

The mutational constraint spectrum quantified from va

Nature

581, 434-443

DOI: [10.1038/s41586-020-2308-7](https://doi.org/10.1038/s41586-020-2308-7)

Citation Report

#	ARTICLE	IF	CITATIONS
1	Future directions for screening and treatment in congenital hearing loss. Precision Clinical Medicine, 2020, 3, 175-186.	1.3	20
2	Heterozygous de novo variants in <i>CSNK1G1</i> are associated with syndromic developmental delay and autism spectrum disorder. Clinical Genetics, 2020, 98, 571-576.	1.0	10
3	Building Infrastructure and Workflows for Clinical Bioinformatics Pipelines. Advances in Molecular Pathology, 2020, 3, 157-167.	0.2	0
4	A MicroRNA Linking Human Positive Selection and Metabolic Disorders. Cell, 2020, 183, 684-701.e14.	13.5	46
5	An enhanced genetic model of relapsed IGH-translocated multiple myeloma evolutionary dynamics. Blood Cancer Journal, 2020, 10, 101.	2.8	11
6	Whole genome sequence analysis of pulmonary function and COPD in 19,996 multi-ethnic participants. Nature Communications, 2020, 11, 5182.	5.8	32
7	Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. Nature Medicine, 2020, 26, 1754-1765.	15.2	84
8	Ultrafast and scalable variant annotation and prioritization with big functional genomics data. Genome Research, 2020, 30, 1789-1801.	2.4	14
9	Use of patient derived urine renal epithelial cells to confirm pathogenicity of PKHD1 alleles. BMC Nephrology, 2020, 21, 435.	0.8	13
10	Immunohistochemistry and Mutation Analysis of SDHx Genes in Carotid Paragangliomas. International Journal of Molecular Sciences, 2020, 21, 6950.	1.8	13
11	An integrated personal and population-based Egyptian genome reference. Nature Communications, 2020, 11, 4719.	5.8	20
12	A large deletion in the COL2A1 gene expands the spectrum of pathogenic variants causing bulldog calf syndrome in cattle. Acta Veterinaria Scandinavica, 2020, 62, 49.	0.5	8
13	Remdesivir and COVID-19. Lancet, The, 2020, 396, 953-954.	6.3	21
14	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	9.4	96
15	Identification of pathogenic missense mutations using protein stability predictors. Scientific Reports, 2020, 10, 15387.	1.6	66
16	Lack of APOE Christchurch variant in five age of onset outliers with PSEN1, PSEN2 Alzheimer's disease and MAPT frontotemporal dementia. Journal of the Neurological Sciences, 2020, 418, 117143.	0.3	2
17	Whole-genome sequencing of acral melanoma reveals genomic complexity and diversity. Nature Communications, 2020, 11, 5259.	5.8	102
18	Clinical advantage of targeted sequencing for unbiased tumor mutational burden estimation in samples with low tumor purity. , 2020, 8, e001199.		7

#	ARTICLE	IF	CITATIONS
19	The Functional Landscape of Patient-Derived RNF43 Mutations Predicts Sensitivity to Wnt Inhibition. <i>Cancer Research</i> , 2020, 80, 5619-5632.	0.4	30
20	Next-generation sequencing of 35 RHD variants in 16% serologically D ⁺ pregnant women in the Finnish population. <i>Blood Advances</i> , 2020, 4, 4994-5001.	2.5	6
21	Characterization of <i>CRB1</i> splicing in retinal organoids derived from a patient with adult-onset rod-cone dystrophy caused by the c.1892A>G and c.2548G>A variants. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1489.	0.6	7
22	Novel insights in the genetics of steroid-sensitive nephrotic syndrome in childhood. <i>Pediatric Nephrology</i> , 2021, 36, 2165-2175.	0.9	11
23	Exome sequencing as a diagnostic tool in chronic kidney disease: ready for clinical application?. <i>Current Opinion in Nephrology and Hypertension</i> , 2020, 29, 608-612.	1.0	1
24	Analysis of the Spectrum of ACE2 Variation Suggests a Possible Influence of Rare and Common Variants on Susceptibility to COVID-19 and Severity of Outcome. <i>Frontiers in Genetics</i> , 2020, 11, 551220.	1.1	32
25	Exome sequencing and characterization of 49,960 individuals in the UK Biobank. <i>Nature</i> , 2020, 586, 749-756.	13.7	369
26	A rare heterozygous variant in FGB (Fibrinogen Merivale) causing hypofibrinogenemia in a Swedish family. <i>Blood Coagulation and Fibrinolysis</i> , 2020, 31, 481-484.	0.5	0
27	High-Throughput Genetic Testing in ALS: The Challenging Path of Variant Classification Considering the ACMG Guidelines. <i>Genes</i> , 2020, 11, 1123.	1.0	15
28	Variants in <i>RABL2A</i> causing male infertility and ciliopathy. <i>Human Molecular Genetics</i> , 2020, 29, 3402-3411.	1.4	11
29	Less Is More, Natural Loss-of-Function Mutation Is a Strategy for Adaptation. <i>Plant Communications</i> , 2020, 1, 100103.	3.6	35
30	Identification of Disease-Associated Variants by Targeted Gene Panel Resequencing in Parkinson's Disease. <i>Frontiers in Neurology</i> , 2020, 11, 576465.	1.1	4
31	Functional analysis of three Nav1.6 mutations causing early infantile epileptic encephalopathy. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2020, 1866, 165959.	1.8	9
32	ROM1 contributes to phenotypic heterogeneity in PRPH2-associated retinal disease. <i>Human Molecular Genetics</i> , 2020, 29, 2708-2722.	1.4	7
33	De novo missense variants disrupting protein-protein interactions affect risk for autism through gene co-expression and protein networks in neuronal cell types. <i>Molecular Autism</i> , 2020, 11, 76.	2.6	19
34	A Data-Driven Approach to Carrier Screening for Common Recessive Diseases. <i>Journal of Personalized Medicine</i> , 2020, 10, 140.	1.1	11
35	FREQMAX provides an alternative approach for determining high-resolution allele frequency thresholds in carrier screening. <i>Human Mutation</i> , 2020, 41, 2078-2086.	1.1	1
36	A CD33 frameshift variant is associated with neuromyelitis optica spectrum disorders. <i>Biomedical Journal</i> , 2021, 44, S93-S100.	1.4	3

#	ARTICLE	IF	CITATIONS
37	Mechanisms of pathogenesis of missense mutations on the KDM6A-H3 interaction in type 2 Kabuki Syndrome. <i>Computational and Structural Biotechnology Journal</i> , 2020, 18, 2033-2042.	1.9	9
38	Sensitive alignment using paralogous sequence variants improves long-read mapping and variant calling in segmental duplications. <i>Nucleic Acids Research</i> , 2020, 48, e114-e114.	6.5	12
39	Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3593-3600.	0.7	16
40	Clinical Implications of the Genetic Architecture of Dilated Cardiomyopathy. <i>Current Cardiology Reports</i> , 2020, 22, 170.	1.3	7
41	Novel <i>F8</i> and <i>F9</i> gene variants from the PedNet hemophilia registry classified according to ACMG/AMP guidelines. <i>Human Mutation</i> , 2020, 41, 2058-2072.	1.1	4
42	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. <i>American Journal of Human Genetics</i> , 2020, 107, 802-814.	2.6	75
43	Inherited causes of clonal haematopoiesis in 97,691 whole genomes. <i>Nature</i> , 2020, 586, 763-768.	13.7	376
44	Evidence for 28 genetic disorders discovered by combining healthcare and research data. <i>Nature</i> , 2020, 586, 757-762.	13.7	343
45	Hematologic presentation and the role of untargeted metabolomics analysis in monitoring treatment for riboflavin transporter deficiency. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2781-2787.	0.7	15
46	Learning disability and myoclonic epilepsy associated with apparently synonymous but splice-disrupting <i>JMJD1C</i> variant that led to 21â€‰bp deletion of the transcript. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 3064-3067.	0.7	0
47	A survey of genetic variants in SARS-CoV-2 interacting domains of ACE2, TMPRSS2 and TLR3/7/8 across populations. <i>Infection, Genetics and Evolution</i> , 2020, 85, 104507.	1.0	31
48	Control of craniofacial and brain development by Cullin3-RING ubiquitin ligases: Lessons from human disease genetics. <i>Experimental Cell Research</i> , 2020, 396, 112300.	1.2	12
49	Modulating gene regulation to treat genetic disorders. <i>Nature Reviews Drug Discovery</i> , 2020, 19, 757-775.	21.5	41
50	Population-specific and trans-ancestry genome-wide analyses identify distinct and shared genetic risk loci for coronary artery disease. <i>Nature Genetics</i> , 2020, 52, 1169-1177.	9.4	206
51	Whole genome, transcriptome and methylome profiling enhances actionable target discovery in high-risk pediatric cancer. <i>Nature Medicine</i> , 2020, 26, 1742-1753.	15.2	185
52	Cross-species regulatory sequence activity prediction. <i>PLoS Computational Biology</i> , 2020, 16, e1008050.	1.5	116
53	Summary of BARD1 Mutations and Precise Estimation of Breast and Ovarian Cancer Risks Associated with the Mutations. <i>Genes</i> , 2020, 11, 798.	1.0	15
54	A novel <i>CACNA1A</i> variant in a child with early stroke and intractable epilepsy. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1383.	0.6	11

#	ARTICLE	IF	CITATIONS
55	Mitochondrial PCK2 Missense Variant in Shetland Sheepdogs with Paroxysmal Exercise-Induced Dyskinesia (PED). <i>Genes</i> , 2020, 11, 774.	1.0	14
56	Mitochondrial Diseases: A Diagnostic Revolution. <i>Trends in Genetics</i> , 2020, 36, 702-717.	2.9	73
57	Novel pathogenic mutations identified in the first Chinese pedigree of complete C6 deficiency. <i>Clinical and Translational Immunology</i> , 2020, 9, e1148.	1.7	3
58	New Insights on Genetic Diagnostics in Cardiomyopathy and Arrhythmia Patients Gained by Stepwise Exome Data Analysis. <i>Journal of Clinical Medicine</i> , 2020, 9, 2168.	1.0	4
59	Bi-allelic Mutations in M1AP Are a Frequent Cause of Meiotic Arrest and Severely Impaired Spermatogenesis Leading to Male Infertility. <i>American Journal of Human Genetics</i> , 2020, 107, 342-351.	2.6	68
60	Lysosome Associated Protein Transmembrane 4B-24 Is the Predominant Protein Isoform in Human Tissues and Undergoes Rapid, Nutrient-Regulated Turnover. <i>American Journal of Pathology</i> , 2020, 190, 2018-2028.	1.9	5
61	Rapid Whole-Exome Sequencing as a Diagnostic Tool in a Neonatal/Pediatric Intensive Care Unit. <i>Journal of Clinical Medicine</i> , 2020, 9, 2220.	1.0	48
62	Ligase IV syndrome can present with microcephaly and radial ray anomalies similar to Fanconi anaemia plus fatal kidney malformations. <i>European Journal of Medical Genetics</i> , 2020, 63, 103974.	0.7	5
63	Description of a family with X-linked oculoauriculovertebral spectrum associated with polyalanine tract expansion in <i>ZIC3</i> . <i>Clinical Genetics</i> , 2020, 98, 384-389.	1.0	11
64	<i>DYNC1H1</i> -related disorders: A description of four new unrelated patients and a comprehensive review of previously reported variants. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2049-2057.	0.7	27
65	Heterozygous missense variant in EIF6 gene: A novel form of Shwachmanâ€“Diamond syndrome?. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2010-2020.	0.7	11
66	Genetic Spectrum of Syndromic and Non-Syndromic Hearing Loss in Pakistani Families. <i>Genes</i> , 2020, 11, 1329.	1.0	7
67	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. <i>American Journal of Human Genetics</i> , 2020, 107, 1078-1095.	2.6	24
68	SURF1 related Leigh syndrome: Clinical and molecular findings of 16 patients from Turkey. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100657.	0.4	10
69	Phased Haplotype Resolution of the SLC6A4 Promoter Using Long-Read Single Molecule Real-Time (SMRT) Sequencing. <i>Genes</i> , 2020, 11, 1333.	1.0	5
70	Stairway Plot 2: demographic history inference with folded SNP frequency spectra. <i>Genome Biology</i> , 2020, 21, 280.	3.8	125
71	Phenotypic Profiling in Subjects Heterozygous for 1 of 2 Rare Variants in the Hypophosphatasia Gene (ALPL). <i>Journal of the Endocrine Society</i> , 2020, 4, bvaa084.	0.1	6
72	Inferring the molecular and phenotypic impact of amino acid variants with MutPred2. <i>Nature Communications</i> , 2020, 11, 5918.	5.8	305

#	ARTICLE	IF	CITATIONS
73	Linking Autism Risk Genes to Disruption of Cortical Development. <i>Cells</i> , 2020, 9, 2500.	1.8	17
74	Integrated Proteogenomic Characterization across Major Histological Types of Pediatric Brain Cancer. <i>Cell</i> , 2020, 183, 1962-1985.e31.	13.5	177
75	The genetic architecture of sporadic and multiple consecutive miscarriage. <i>Nature Communications</i> , 2020, 11, 5980.	5.8	52
76	Expediting rare disease diagnosis: a call to bridge the gap between clinical and functional genomics. <i>Molecular Medicine</i> , 2020, 26, 117.	1.9	10
77	Candidate Gene Discovery in Hereditary Colorectal Cancer and Polyposis Syndromes—Considerations for Future Studies. <i>International Journal of Molecular Sciences</i> , 2020, 21, 8757.	1.8	7
78	Hereditary hyperferritinemia-cataract syndrome in three Czech families: molecular genetic testing and clinical implications. <i>Journal of AAPOS</i> , 2020, 24, 352.e1-352.e5.	0.2	2
79	VarStack: a web tool for data retrieval to interpret somatic variants in cancer. <i>Database: the Journal of Biological Databases and Curation</i> , 2020, 2020, .	1.4	2
80	Structural bioinformatics enhances mechanistic interpretation of genomic variation, demonstrated through the analyses of 935 distinct RAS family mutations. <i>Bioinformatics</i> , 2021, 37, 1367-1375.	1.8	6
81	A Novel Case of Homozygous Interferon Alpha/Beta Receptor Alpha Chain (IFNAR1) Deficiency With Hemophagocytic Lymphohistiocytosis. <i>Clinical Infectious Diseases</i> , 2022, 74, 136-139.	2.9	24
82	A complex DICER1 syndrome phenotype associated with a germline pathogenic variant affecting the RNase IIIa domain of DICER1. <i>Journal of Medical Genetics</i> , 2022, 59, 141-146.	1.5	9
83	Multiple Self-Healing Squamous Epithelioma (MSSE): A Digenic Trait Associated with Loss of Function Mutations in TGFBR1 and Variants at a Second Linked Locus on the Long Arm of Chromosome 9. <i>Genes</i> , 2020, 11, 1410.	1.0	11
84	Peroxisome proliferator-activated receptor δ as a novel therapeutic target for schizophrenia. <i>EBioMedicine</i> , 2020, 62, 103130.	2.7	19
85	Genomic Sequencing for Newborn Screening: Results of the NC NEXUS Project. <i>American Journal of Human Genetics</i> , 2020, 107, 596-611.	2.6	63
86	Systematic Review of Sequencing Studies and Gene Expression Profiling in Familial Meniere Disease. <i>Genes</i> , 2020, 11, 1414.	1.0	15
87	A CNTNAP1 Missense Variant Is Associated with Canine Laryngeal Paralysis and Polyneuropathy. <i>Genes</i> , 2020, 11, 1426.	1.0	9
88	Rare Variant Burden Analysis within Enhancers Identifies CAV1 as an ALS Risk Gene. <i>Cell Reports</i> , 2020, 33, 108456.	2.9	24
89	Interleukin-11 signaling underlies fibrosis, parenchymal dysfunction, and chronic inflammation of the airway. <i>Experimental and Molecular Medicine</i> , 2020, 52, 1871-1878.	3.2	58
90	The 40bp Indel Polymorphism rs150550023 in the MDM2 Promoter is Associated with Intriguing Shifts in Gene Expression in the p53-MDM2 Regulatory Hub. <i>Cancers</i> , 2020, 12, 3363.	1.7	3

#	ARTICLE	IF	CITATIONS
91	Accurate assembly of the olive baboon (<i>Papio anubis</i>) genome using long-read and Hi-C data. <i>GigaScience</i> , 2020, 9, .	3.3	18
92	Novel Variants in LRRK2 and GBA Identified in Latino Parkinson Disease Cohort Enriched for Caribbean Origin. <i>Frontiers in Neurology</i> , 2020, 11, 573733.	1.1	6
93	A comparative genomics multitool for scientific discovery and conservation. <i>Nature</i> , 2020, 587, 240-245.	13.7	216
94	Genetic profiling of Vietnamese population from large-scale genomic analysis of non-invasive prenatal testing data. <i>Scientific Reports</i> , 2020, 10, 19142.	1.6	8
95	Prediction of driver variants in the cancer genome via machine learning methodologies. <i>Briefings in Bioinformatics</i> , 2021, 22, .	3.2	13
96	Carriership of two copies of C9orf72 hexanucleotide repeat intermediate-length alleles is a risk factor for ALS in the Finnish population. <i>Acta Neuropathologica Communications</i> , 2020, 8, 187.	2.4	16
97	ANO3 and early-onset dyskinetic encephalopathy. <i>European Journal of Medical Genetics</i> , 2020, 63, 104085.	0.7	3
98	Finding MEMO—Emerging Evidence for MEMO1's Function in Development and Disease. <i>Genes</i> , 2020, 11, 1316.	1.0	13
99	The FKBP4 Gene, Encoding a Regulator of the Androgen Receptor Signaling Pathway, Is a Novel Candidate Gene for Androgen Insensitivity Syndrome. <i>International Journal of Molecular Sciences</i> , 2020, 21, 8403.	1.8	6
100	Rare Variants in Genes Associated With Cardiomyopathy Are Not Common in Hypoplastic Left Heart Syndrome Patients With Myocardial Dysfunction. <i>Frontiers in Pediatrics</i> , 2020, 8, 596840.	0.9	4
101	Detection of Pathogenic Variants With Germline Genetic Testing Using Deep Learning vs Standard Methods in Patients With Prostate Cancer and Melanoma. <i>JAMA - Journal of the American Medical Association</i> , 2020, 324, 1957.	3.8	33
102	Germline AGO2 mutations impair RNA interference and human neurological development. <i>Nature Communications</i> , 2020, 11, 5797.	5.8	43
103	DYNC2H1 hypomorphic or retina-predominant variants cause nonsyndromic retinal degeneration. <i>Genetics in Medicine</i> , 2020, 22, 2041-2051.	1.1	38
104	Development and validation of a universal blood donor genotyping platform: a multinational prospective study. <i>Blood Advances</i> , 2020, 4, 3495-3506.	2.5	31
105	Genetic and polygenic risk score analysis for Alzheimer's disease in the Chinese population. <i>Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring</i> , 2020, 12, e12074.	1.2	14
106	Epigenomic and Transcriptomic Dynamics During Human Heart Organogenesis. <i>Circulation Research</i> , 2020, 127, e184-e209.	2.0	27
107	Myeloperoxidase Modulates Inflammation in Generalized Pustular Psoriasis and Additional Rare Pustular Skin Diseases. <i>American Journal of Human Genetics</i> , 2020, 107, 527-538.	2.6	53
108	Presence of Genetic Variants Among Young Men With Severe COVID-19. <i>JAMA - Journal of the American Medical Association</i> , 2020, 324, 663.	3.8	626

#	ARTICLE	IF	CITATIONS
109	Absence of mucosal-associated invariant T cells in a person with a homozygous point mutation in <i>MR1</i> . <i>Science Immunology</i> , 2020, 5, .	5.6	50
110	An International Multicenter Evaluation of Inheritance Patterns, Arrhythmic Risks, and Underlying Mechanisms of <i>CASQ2</i> -Catecholaminergic Polymorphic Ventricular Tachycardia. <i>Circulation</i> , 2020, 142, 932-947.	1.6	44
111	Novel manifestations of immune dysregulation and granule defects in gray platelet syndrome. <i>Blood</i> , 2020, 136, 1956-1967.	0.6	34
112	Compound Heterozygous <i>SCN5A</i> Mutations in Severe Sodium Channelopathy With Brugada Syndrome: A Case Report. <i>Frontiers in Cardiovascular Medicine</i> , 2020, 7, 117.	1.1	3
113	Functional Validation of <i>CLDN</i> Variants Identified in a Neural Tube Defect Cohort Demonstrates Their Contribution to Neural Tube Defects. <i>Frontiers in Neuroscience</i> , 2020, 14, 664.	1.4	5
114	A Novel Approach for the Identification of Pharmacogenetic Variants in <i>MT-RNR1</i> through Next-Generation Sequencing Off-Target Data. <i>Journal of Clinical Medicine</i> , 2020, 9, 2082.	1.0	0
115	Protein-Protein Interactions Mediated by Intrinsically Disordered Protein Regions Are Enriched in Missense Mutations. <i>Biomolecules</i> , 2020, 10, 1097.	1.8	22
116	<i>PGAP3</i> Associated with Hyperphosphatasia with Mental Retardation Plays a Novel Role in Brain Morphogenesis and Neuronal Wiring at Early Development. <i>Cells</i> , 2020, 9, 1782.	1.8	14
117	Optimizing Genetic Diagnosis of Neurodevelopmental Disorders in the Clinical Setting. <i>Clinics in Laboratory Medicine</i> , 2020, 40, 231-256.	0.7	11
118	Prenatal clinical manifestations in individuals with <i>COL4A1/2</i> variants. <i>Journal of Medical Genetics</i> , 2021, 58, 505-513.	1.5	22
119	A founder noncoding <i>GALT</i> variant interfering with splicing causes galactosemia. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1199-1204.	1.7	1
120	Compound heterozygous splicing <i>CDON</i> variants result in isolated ocular coloboma. <i>Clinical Genetics</i> , 2020, 98, 486-492.	1.0	4
121	Literature Review of <i>BARD1</i> as a Cancer Predisposing Gene with a Focus on Breast and Ovarian Cancers. <i>Genes</i> , 2020, 11, 856.	1.0	30
122	Predictive and prognostic value of <i>LSP1</i> rs3817198 in sporadic breast cancer in northeastern population of Iran. <i>Experimental and Molecular Pathology</i> , 2020, 116, 104514.	0.9	3
123	Implications of <i>TP53</i> allelic state for genome stability, clinical presentation and outcomes in myelodysplastic syndromes. <i>Nature Medicine</i> , 2020, 26, 1549-1556.	15.2	372
124	Genotype-phenotype correlation at codon 1740 of <i>SETD2</i> . <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2037-2048.	0.7	14
125	Synonymous Mutation in <i>DKC1</i> Causes Telomerase RNA Insufficiency Manifesting as Familial Pulmonary Fibrosis. <i>Chest</i> , 2020, 158, 2449-2457.	0.4	26
126	Vascular inflammation and endothelial injury in SARS-CoV-2 infection: the overlooked regulatory cascades implicated by the <i>ACE2</i> gene cluster. <i>QJM - Monthly Journal of the Association of Physicians</i> , 2020, , .	0.2	11

#	ARTICLE	IF	CITATIONS
127	Precision medicine for pancreatic diseases. <i>Current Opinion in Gastroenterology</i> , 2020, 36, 428-436.	1.0	4
128	Deficiency of plasminogen activator inhibitor-2 results in accelerated tumor growth. <i>Journal of Thrombosis and Haemostasis</i> , 2020, 18, 2968-2975.	1.9	10
129	Whole genome sequence analysis identifies a PAX2 mutation to establish a correct diagnosis for a syndromic form of hyperuricemia. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2521-2528.	0.7	3
130	Promoter CpG Density Predicts Downstream Gene Loss-of-Function Intolerance. <i>American Journal of Human Genetics</i> , 2020, 107, 487-498.	2.6	12
131	The Gnomad Consortium Releases First Studies of Human Genetic Variation. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1999-2000.	0.7	3
132	New Insights into Potocki-Shaffer Syndrome: Report of Two Novel Cases and Literature Review. <i>Brain Sciences</i> , 2020, 10, 788.	1.1	7
133	Patient derived stem cells for discovery and validation of novel pathogenic variants in inherited retinal disease. <i>Progress in Retinal and Eye Research</i> , 2021, 83, 100918.	7.3	16
134	Comprehensive characterization of amino acid positions in protein structures reveals molecular effect of missense variants. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 28201-28211.	3.3	68
135	Genetics and Genomics of Pediatric Pulmonary Arterial Hypertension. <i>Genes</i> , 2020, 11, 1213.	1.0	24
136	SLC19A3 Loss-of-Function Variant in Yorkshire Terriers with Leigh-Like Subacute Necrotizing Encephalopathy. <i>Genes</i> , 2020, 11, 1215.	1.0	4
137	ERCC3, a new ovarian cancer susceptibility gene?. <i>European Journal of Cancer</i> , 2020, 141, 1-8.	1.3	8
138	A novel RFC1 repeat motif (ACAGG) in two Asia-Pacific CANVAS families. <i>Brain</i> , 2020, 143, 2904-2910.	3.7	53
139	Prediction of genome-wide effects of single nucleotide variants on transcription factor binding. <i>Scientific Reports</i> , 2020, 10, 17632.	1.6	11
140	The First Report of Biallelic Missense Mutations in the SFRP4 Gene Causing Pyle Disease in Two Siblings. <i>Frontiers in Genetics</i> , 2020, 11, 593407.	1.1	8
141	ACE2 coding variants in different populations and their potential impact on SARS-CoV-2 binding affinity. <i>Biochemistry and Biophysics Reports</i> , 2020, 24, 100798.	0.7	28
142	Sequencing of the complex CTRB1-CTRB2 locus in chronic pancreatitis. <i>Pancreatology</i> , 2020, 20, 1598-1603.	0.5	5
143	High-depth African genomes inform human migration and health. <i>Nature</i> , 2020, 586, 741-748.	13.7	197
144	Somatic Mutations in UBA1 and Severe Adult-Onset Autoinflammatory Disease. <i>New England Journal of Medicine</i> , 2020, 383, 2628-2638.	13.9	580

#	ARTICLE	IF	CITATIONS
145	Two New Cases of Hypertrophic Cardiomyopathy and Skeletal Muscle Features Associated with ALPK3 Homozygous and Compound Heterozygous Variants. <i>Genes</i> , 2020, 11, 1201.	1.0	20
146	Current Insights in Elucidation of Possible Molecular Mechanisms of the Juvenile Form of Batten Disease. <i>International Journal of Molecular Sciences</i> , 2020, 21, 8055.	1.8	4
147	Computational Modeling of NLRP3 Identifies Enhanced ATP Binding and Multimerization in Cryopyrin-Associated Periodic Syndromes. <i>Frontiers in Immunology</i> , 2020, 11, 584364.	2.2	9
148	Strategic vision for improving human health at The Forefront of Genomics. <i>Nature</i> , 2020, 586, 683-692.	13.7	192
149	LDBlockShow: a fast and convenient tool for visualizing linkage disequilibrium and haplotype blocks based on variant call format files. <i>Briefings in Bioinformatics</i> , 2021, 22, .	3.2	177
150	Mutations in normal tissues—some diagnostic and clinical implications. <i>BMC Medicine</i> , 2020, 18, 283.	2.3	19
151	IndiGenomes: a comprehensive resource of genetic variants from over 1000 Indian genomes. <i>Nucleic Acids Research</i> , 2021, 49, D1225-D1232.	6.5	39
152	Genome sequencing unveils mutational landscape of the familial Mediterranean fever: Potential implications of IL33/ST2 signalling. <i>Journal of Cellular and Molecular Medicine</i> , 2020, 24, 11294-11306.	1.6	7
153	COVID-19 and Genetic Variants of Protein Involved in the SARS-CoV-2 Entry into the Host Cells. <i>Genes</i> , 2020, 11, 1010.	1.0	88
154	Genetic and Clinical Predictors of Age of ESKD in Individuals With Autosomal Dominant Tubulointerstitial Kidney Disease Due to UMOD Mutations. <i>Kidney International Reports</i> , 2020, 5, 1472-1485.	0.4	30
155	The Evolution of the Mammalian <i>ABCA6</i> -like Genes: Analysis of Phylogenetic, Expression, and Population Genetic Data Reveals Complex Evolutionary Histories. <i>Genome Biology and Evolution</i> , 2020, 12, 2093-2106.	1.1	2
156	<i>SYNGAP1</i> Controls the Maturation of Dendrites, Synaptic Function, and Network Activity in Developing Human Neurons. <i>Journal of Neuroscience</i> , 2020, 40, 7980-7994.	1.7	38
157	Variant c.2158-2A>G in MANBA is an important and frequent cause of hereditary hearing loss and beta-mannosidosis among the Czech and Slovak Roma population- evidence for a new ethnic-specific variant. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 222.	1.2	4
158	Sudden infant death with dysgenesis of the testes syndrome in an Amish infant: A case report. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2751-2754.	0.7	4
159	DeepVariant-on-Spark: Small-Scale Genome Analysis Using a Cloud-Based Computing Framework. <i>Computational and Mathematical Methods in Medicine</i> , 2020, 2020, 1-7.	0.7	6
160	Whole exome sequencing highlights variants in association with Keratoconus in Jordanian families. <i>BMC Medical Genetics</i> , 2020, 21, 177.	2.1	6
161	Inherited Rare, Deleterious Variants in ATM Increase Lung Adenocarcinoma Risk. <i>Journal of Thoracic Oncology</i> , 2020, 15, 1871-1879.	0.5	24
162	Circulating testican-2 is a podocyte-derived marker of kidney health. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 25026-25035.	3.3	19

#	ARTICLE	IF	CITATIONS
163	Presynaptic dysfunction in CASK-related neurodevelopmental disorders. <i>Translational Psychiatry</i> , 2020, 10, 312.	2.4	28
164	Evaluating the role of <i>NTHL1</i> p.Q90* allele in inherited breast cancer predisposition. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1493.	0.6	5
165	Two Novel Pathogenic Variants Confirm RMND1 Causative Role in Perrault Syndrome with Renal Involvement. <i>Genes</i> , 2020, 11, 1060.	1.0	16
166	Identification of single nucleotide variants in the Moroccan population by whole-genome sequencing. <i>BMC Genetics</i> , 2020, 21, 111.	2.7	3
167	Effect of 6p21 region on lung function is modified by smoking: a genome-wide interaction study. <i>Scientific Reports</i> , 2020, 10, 13075.	1.6	6
168	Community-based recruitment and exome sequencing indicates high diagnostic yield in adults with intellectual disability. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1439.	0.6	6
169	<p></p>A Homozygous Truncating Mutation in NALCN Causing IHPRF1: Detailed Clinical Manifestations and a Review of Literature</p>. <i>The Application of Clinical Genetics</i> , 2020, Volume 13, 151-157.	1.4	10
170	Annotation of Human Exome Gene Variants with Consensus Pathogenicity. <i>Genes</i> , 2020, 11, 1076.	1.0	4
171	TIE1 as a Candidate Gene for Lymphatic Malformations with or without Lymphedema. <i>International Journal of Molecular Sciences</i> , 2020, 21, 6780.	1.8	11
172	Functional analysis of Sonic Hedgehog variants associated with holoprosencephaly in humans using a CRISPR/Cas9 zebrafish model. <i>Human Mutation</i> , 2020, 41, 2155-2166.	1.1	4
173	Advances in Genomics for Drug Development. <i>Genes</i> , 2020, 11, 942.	1.0	22
174	CHD8 dosage regulates transcription in pluripotency and early murine neural differentiation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2020, 117, 22331-22340.	3.3	27
175	A novel homozygous variant in <i>TRAPPC2L</i> results in a neurodevelopmental disorder and disrupts TRAPP complex function. <i>Journal of Medical Genetics</i> , 2021, 58, 592-601.	1.5	10
176	Monoallelic NTHL1 Loss-of-Function Variants and Risk of Polyposis and Colorectal Cancer. <i>Gastroenterology</i> , 2020, 159, 2241-2243.e6.	0.6	20
177	Assessing performance of pathogenicity predictors using clinically relevant variant datasets. <i>Journal of Medical Genetics</i> , 2021, 58, 547-555.	1.5	57
178	Analysis of APPL1 Gene Polymorphisms in Patients with a Phenotype of Maturity Onset Diabetes of the Young. <i>Journal of Personalized Medicine</i> , 2020, 10, 100.	1.1	13
179	Missense variants in the spectrin repeat domain of DSP are associated with arrhythmogenic cardiomyopathy: A family report and systematic review. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2359-2368.	0.7	6
180	Phenome-wide analyses establish a specific association between aortic valve PALMD expression and calcific aortic valve stenosis. <i>Communications Biology</i> , 2020, 3, 477.	2.0	12

#	ARTICLE	IF	CITATIONS
181	The first reported case of Loey's-Dietz syndrome in a patient with biallelic SMAD3 variants. American Journal of Medical Genetics, Part A, 2020, 182, 2755-2760.	0.7	3
182	Common atrium/atrioventricular canal defect and postaxial polydactyly: A mild clinical subtype of Ellis-van Creveld syndrome caused by hypomorphic mutations in the <i>EVC</i> gene. Human Mutation, 2020, 41, 2087-2093.	1.1	7
183	Amyloid- β 43 cerebrospinal fluid levels and the interpretation of APP, PSEN1 and PSEN2 mutations. Alzheimer's Research and Therapy, 2020, 12, 108.	3.0	17
184	Structural models of human ACE2 variants with SARS-CoV-2 Spike protein for structure-based drug design. Scientific Data, 2020, 7, 309.	2.4	26
185	The Macrophage Migration Inhibitory Factor (MIF) Promoter Polymorphisms (rs3063368, rs755622) Predict Acute Kidney Injury and Death after Cardiac Surgery. Journal of Clinical Medicine, 2020, 9, 2936.	1.0	9
186	Investigating the role of BEST1 and PRPH2 variants in the molecular aetiology of adult-onset vitelliform macular dystrophies. Ophthalmic Genetics, 2020, 41, 585-590.	0.5	3
187	Functional Characterization Reveals the Significance of Rare Coding Variations in Human Organic Anion Transporting Polypeptide 2B1 (<i>SLCO2B1</i>). Molecular Pharmaceutics, 2020, 17, 3966-3978.	2.3	14
188	Diagnostic yield and clinical utility of whole exome sequencing using an automated variant prioritization system, <i>EVIDENCE</i> . Clinical Genetics, 2020, 98, 562-570.	1.0	76
189	Transcriptomic signatures across human tissues identify functional rare genetic variation. Science, 2020, 369, .	6.0	89
190	A Deep Learning Framework Identifies Pathogenic Noncoding Somatic Mutations from Personal Prostate Cancer Genomes. Cancer Research, 2020, 80, 4644-4654.	0.4	9
191	The Membrane Transporter OAT7 (SLC22A9) Is Not a Susceptibility Factor for Osteoporosis in Europeans. Frontiers in Endocrinology, 2020, 11, 532.	1.5	2
192	Polygenic background modifies penetrance of monogenic variants for tier 1 genomic conditions. Nature Communications, 2020, 11, 3635.	5.8	277
193	Transcriptional adaptation: a mechanism underlying genetic robustness. Development (Cambridge), 2020, 147, .	1.2	44
194	The position of nonsense mutations can predict the phenotype severity: A survey on the DMD gene. PLoS ONE, 2020, 15, e0237803.	1.1	25
195	Broad host range of SARS-CoV-2 predicted by comparative and structural analysis of ACE2 in vertebrates. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 22311-22322.	3.3	517
196	Association of Sex With Frequent and Mild <i>ABCA4</i> Alleles in Stargardt Disease. JAMA Ophthalmology, 2020, 138, 1035.	1.4	31
197	A novel homozygous variant in REN in a family presenting with classic features of disorders involving the renin-angiotensin pathway, without renal tubular dysgenesis. American Journal of Medical Genetics, Part A, 2020, 182, 2284-2290.	0.7	3
198	Screening of a large Rubinstein-Taybi cohort identified many novel variants and emphasizes the importance of the CREBBP histone acetyltransferase domain. American Journal of Medical Genetics, Part A, 2020, 182, 2508-2520.	0.7	9

#	ARTICLE	IF	CITATIONS
199	Machine learning for effectively avoiding overfitting is a crucial strategy for the genetic prediction of polygenic psychiatric phenotypes. <i>Translational Psychiatry</i> , 2020, 10, 294.	2.4	11
200	A missense mutation in the CSTF2 gene that impairs the function of the RNA recognition motif and causes defects in 3' end processing is associated with intellectual disability in humans. <i>Nucleic Acids Research</i> , 2020, 48, 9804-9821.	6.5	10
201	Genetic Studies of Hypertrophic Cardiomyopathy in Singaporeans Identify Variants in <i>TNNI3</i> and <i>TNNT2</i> That Are Common in Chinese Patients. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, 424-434.	1.6	18
202	The road ahead in genetics and genomics. <i>Nature Reviews Genetics</i> , 2020, 21, 581-596.	7.7	118
203	The Needle in the Haystack—Searching for Genetic and Epigenetic Differences in Monozygotic Twins Discordant for Tetralogy of Fallot. <i>Journal of Cardiovascular Development and Disease</i> , 2020, 7, 55.	0.8	10
204	PTRH2: an adhesion regulated molecular switch at the nexus of life, death, and differentiation. <i>Cell Death Discovery</i> , 2020, 6, 124.	2.0	9
205	Mutational analysis in familial Alzheimer's disease of Han Chinese in Taiwan with a predominant mutation PSEN1 p.Met146Ile. <i>Scientific Reports</i> , 2020, 10, 19769.	1.6	7
206	Analysis of HLA-G long-read genomic sequences in mother-offspring pairs with preeclampsia. <i>Scientific Reports</i> , 2020, 10, 20027.	1.6	5
207	Nasopharyngeal carcinoma MHC region deep sequencing identifies HLA and novel non-HLA TRIM31 and TRIM39 loci. <i>Communications Biology</i> , 2020, 3, 759.	2.0	17
208	Bayesian Inference Associates Rare <i>KDR</i> Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, .	1.6	29
209	Cystic Fibrosis Polymorphic Variants in a Russian Population. <i>Pharmacogenomics and Personalized Medicine</i> , 2020, Volume 13, 679-686.	0.4	6
210	Mutation Frequency in Main Susceptibility Genes Among Patients With Head and Neck Paragangliomas. <i>Frontiers in Genetics</i> , 2020, 11, 614908.	1.1	16
211	Investigation of Genetic Variations of IL6 and IL6R as Potential Prognostic and Pharmacogenetics Biomarkers: Implications for COVID-19 and Neuroinflammatory Disorders. <i>Life</i> , 2020, 10, 351.	1.1	24
212	The role of polygenic risk and susceptibility genes in breast cancer over the course of life. <i>Nature Communications</i> , 2020, 11, 6383.	5.8	101
213	AsCRISPR: A Web Server for Allele-Specific Single Guide RNA Design in Precision Medicine. <i>CRISPR Journal</i> , 2020, 3, 512-522.	1.4	8
214	PERHAPS: Paired-End short Reads-based HAPlotyping from next-generation Sequencing data. <i>Briefings in Bioinformatics</i> , 2021, 22, .	3.2	5
215	Evaluation of Single-Molecule Sequencing Technologies for Structural Variant Detection in Two Swedish Human Genomes. <i>Genes</i> , 2020, 11, 1444.	1.0	6
216	Creation of bladder assembloids mimicking tissue regeneration and cancer. <i>Nature</i> , 2020, 588, 664-669.	13.7	133

#	ARTICLE	IF	CITATIONS
217	Mutations Causing Mild or No Structural Damage in Interfaces of Multimerization of the Fibrinogen Î³-Module More Likely Confer Negative Dominant Behaviors. <i>International Journal of Molecular Sciences</i> , 2020, 21, 9016.	1.8	3
218	MosaicBase: A Knowledgebase of Postzygotic Mosaic Variants in Noncancer Disease-related and Healthy Human Individuals. <i>Genomics, Proteomics and Bioinformatics</i> , 2020, 18, 140-149.	3.0	10
219	Genome sequencing identifies a rare case of moderate Zellweger spectrum disorder caused by a PEX3 defect: Case report and literature review. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 25, 100664.	0.4	1
220	Familial neonatal seizures caused by the Kv7.3 selectivity filter mutation T313I. <i>Epilepsia Open</i> , 2020, 5, 562-573.	1.3	4
221	Human T-bet Governs Innate and Innate-like Adaptive IFN-Î³ Immunity against Mycobacteria. <i>Cell</i> , 2020, 183, 1826-1847.e31.	13.5	83
222	Missense variant contribution to USP9X-female syndrome. <i>Npj Genomic Medicine</i> , 2020, 5, 53.	1.7	17
223	Structure and Characterization of Phosphoglucomutase 5 from Atlantic and Baltic Herring: An Inactive Enzyme with Intact Substrate Binding. <i>Biomolecules</i> , 2020, 10, 1631.	1.8	4
224	A Multi-Strategy Sequencing Workflow in Inherited Retinal Dystrophies: Routine Diagnosis, Addressing Unsolved Cases and Candidate Genes Identification. <i>International Journal of Molecular Sciences</i> , 2020, 21, 9355.	1.8	5
225	ClassifyCNV: a tool for clinical annotation of copy-number variants. <i>Scientific Reports</i> , 2020, 10, 20375.	1.6	34
226	Human mutational constraint as a tool to understand biology of rare and emerging bone marrow failure syndromes. <i>Blood Advances</i> , 2020, 4, 5232-5245.	2.5	8
227	Germline TP53 Testing in Breast Cancers: Why, When and How?. <i>Cancers</i> , 2020, 12, 3762.	1.7	16
228	Pharmacoresistant Epilepsy in Childhood: Think of the Cerebral Folate Deficiency, a Treatable Disease. <i>Brain Sciences</i> , 2020, 10, 762.	1.1	6
229	GJB2 and GJB6 Genetic Variant Curation in an Argentinean Non-Syndromic Hearing-Impaired Cohort. <i>Genes</i> , 2020, 11, 1233.	1.0	13
230	Environmental Epigenetics of Diesel Particulate Matter Toxicogenomics. <i>International Journal of Environmental Research and Public Health</i> , 2020, 17, 7386.	1.2	0
231	MPEG1/Perforin-2 Haploinsufficiency Associated Polymicrobial Skin Infections and Considerations for Interferon-Î³ Therapy. <i>Frontiers in Immunology</i> , 2020, 11, 601584.	2.2	5
232	PRIM1 deficiency causes a distinctive primordial dwarfism syndrome. <i>Genes and Development</i> , 2020, 34, 1520-1533.	2.7	20
233	<i>Gabra2</i> is a genetic modifier of <i>Scn8a</i> encephalopathy in the mouse*. <i>Epilepsia</i> , 2020, 61, 2847-2856.	2.6	15
234	Bi-Allelic Novel Variants in CLIC5 Identified in a Cameroonian Multiplex Family with Non-Syndromic Hearing Impairment. <i>Genes</i> , 2020, 11, 1249.	1.0	9

#	ARTICLE	IF	CITATIONS
235	Linking Pharmacogenomic Information on Drug Safety and Efficacy with Ethnic Minority Populations. <i>Pharmaceutics</i> , 2020, 12, 1021.	2.0	3
236	From membrane to mineralization: the curious case of the ABCC6 transporter. <i>FEBS Letters</i> , 2020, 594, 4109-4133.	1.3	8
237	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. <i>American Journal of Human Genetics</i> , 2020, 107, 963-976.	2.6	18
238	<i>KMT2B</i> -related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. <i>Brain</i> , 2020, 143, 3242-3261.	3.7	57
239	PRPS-ST: A Protocol-Agnostic Self-training Method for Gene Expression-Based Classification of Blood Cancers. <i>Blood Cancer Discovery</i> , 2020, 1, 244-257.	2.6	4
240	Programmed Cell Death 2-Like (<i>Pdcd2l</i>) Is Required for Mouse Embryonic Development. <i>G3: Genes, Genomes, Genetics</i> , 2020, 10, 4449-4457.	0.8	2
241	Two Aldehyde Clearance Systems Are Essential to Prevent Lethal Formaldehyde Accumulation in Mice and Humans. <i>Molecular Cell</i> , 2020, 80, 996-1012.e9.	4.5	92
242	Congenital Heart Defects Due to <i>TAF1</i> Missense Variants. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, e002843.	1.6	8
243	In silico features of ADAMTS13 contributing to plasmatic ADAMTS13 levels in neonates with congenital heart disease. <i>Thrombosis Research</i> , 2020, 193, 66-76.	0.8	2
244	Exploring human genomic diversity with gnomAD. <i>Nature Reviews Genetics</i> , 2020, 21, 448-448.	7.7	85
245	Evaluating drug targets through human loss-of-function genetic variation. <i>Nature</i> , 2020, 581, 459-464.	13.7	115
246	Hypoparathyroidism, deafness, and renal dysplasia syndrome: 20 Years after the identification of the first <i>GATA3</i> mutations. <i>Human Mutation</i> , 2020, 41, 1341-1350.	1.1	19
247	A structural variation reference for medical and population genetics. <i>Nature</i> , 2020, 581, 444-451.	13.7	614
248	Transcript expression-aware annotation improves rare variant interpretation. <i>Nature</i> , 2020, 581, 452-458.	13.7	142
249	Functional annotation of rare structural variation in the human brain. <i>Nature Communications</i> , 2020, 11, 2990.	5.8	32
250	Expanding the phenotypic spectrum consequent upon de novo <i>WDR37</i> missense variants. <i>Clinical Genetics</i> , 2020, 98, 191-197.	1.0	8
251	Parkinson disease risk variants in East Asian populations. <i>Nature Reviews Neurology</i> , 2020, 16, 461-462.	4.9	3
252	Deleterious mis-splicing of <i>STK11</i> caused by a novel single nucleotide substitution in the 3' polypyrimidine tract of intron five. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1381.	0.6	3

#	ARTICLE	IF	CITATIONS
253	Expanded carrier screening in Chinese patients seeking the help of assisted reproductive technology. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1340.	0.6	22
254	Chromosomal alterations among age-related haematopoietic clones in Japan. <i>Nature</i> , 2020, 584, 130-135.	13.7	102
255	Assembly and annotation of an Ashkenazi human reference genome. <i>Genome Biology</i> , 2020, 21, 129.	3.8	42
256	Consolidated BRCA1/2 Variant Interpretation by MH BRCA Correlates with Predicted PARP Inhibitor Efficacy Association by MH Guide. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3895.	1.8	4
257	Rheostat positions: A new classification of protein positions relevant to pharmacogenomics. <i>Medicinal Chemistry Research</i> , 2020, 29, 1133-1146.	1.1	16
258	Novel loss-of-function mutations in COCH cause autosomal recessive nonsyndromic hearing loss. <i>Human Genetics</i> , 2020, 139, 1565-1574.	1.8	13
259	Transcriptional consequences of MBD5 disruption in mouse brain and CRISPR-derived neurons. <i>Molecular Autism</i> , 2020, 11, 45.	2.6	11
260	Molecular Diagnosis and Genetic Counseling of Cystic Fibrosis and Related Disorders: New Challenges. <i>Genes</i> , 2020, 11, 619.	1.0	15
261	Germline TET2 loss of function causes childhood immunodeficiency and lymphoma. <i>Blood</i> , 2020, 136, 1055-1066.	0.6	58
262	The frontiers of sequencing in undiagnosed neurodevelopmental diseases. <i>Current Opinion in Genetics and Development</i> , 2020, 65, 76-83.	1.5	6
263	Gene constraint and genotype-phenotype correlations in neurodevelopmental disorders. <i>Current Opinion in Genetics and Development</i> , 2020, 65, 69-75.	1.5	7
264	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 1205-1215.	1.2	14
265	A novel VPS13B mutation in Cohen syndrome: a case report and review of literature. <i>BMC Medical Genetics</i> , 2020, 21, 140.	2.1	24
266	Association of <i>APOE</i> With Primary Open-Angle Glaucoma Suggests a Protective Effect for <i>APOE</i> ϵ 4. <i>Investigative Ophthalmology and Visual Science</i> , 2020, 61, 3.		23
267	Loqusdb: added value of an observations database of local genomic variation. <i>BMC Bioinformatics</i> , 2020, 21, 273.	1.2	5
268	lentiMPRA and MPRAflow for high-throughput functional characterization of gene regulatory elements. <i>Nature Protocols</i> , 2020, 15, 2387-2412.	5.5	65
269	Analysis of ACE2 Genetic Variability among Populations Highlights a Possible Link with COVID-19-Related Neurological Complications. <i>Genes</i> , 2020, 11, 741.	1.0	54
270	The Role of <i>FER</i> rs4957796 in the Risk of Developing and Dying from a Bloodstream Infection: A 23-Year Follow-up of the Population-based Nord-Trøndelag Health Study. <i>Clinical Infectious Diseases</i> , 2021, 73, e297-e303.	2.9	1

#	ARTICLE	IF	CITATIONS
271	Mutational and phenotypic characterization of hereditary hemorrhagic telangiectasia. <i>Blood</i> , 2020, 136, 1907-1918.	0.6	40
272	Loss-of-Function <i>CREB3L3</i> Variants in Patients With Severe Hypertriglyceridemia. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2020, 40, 1935-1941.	1.1	19
273	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A (<i>KIF1A</i>) in patients with developmental delay and intellectual disability. <i>Journal of Medical Genetics</i> , 2020, 57, 100-106.	1.1	16
274	Embracing human genetics: a primer for developmental biologists. <i>Development (Cambridge)</i> , 2020, 147, .	1.2	3
275	Concurrent sodium channelopathies and amyotrophic lateral sclerosis supports shared pathogenesis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2020, 21, 627-630.	1.1	5
276	A homozygous pathogenic variant in <i>SHROOM3</i> associated with anencephaly and cleft lip and palate. <i>Clinical Genetics</i> , 2020, 98, 299-302.	1.0	8
277	Clinical interpretation of variants identified in RNU4ATAC, a non-coding spliceosomal gene. <i>PLoS ONE</i> , 2020, 15, e0235655.	1.1	8
278	VarFish: comprehensive DNA variant analysis for diagnostics and research. <i>Nucleic Acids Research</i> , 2020, 48, W162-W169.	6.5	39
279	Breakpoint Mapping of Symptomatic Balanced Translocations Links the EPHA6, KLF13 and UBR3 Genes to Novel Disease Phenotype. <i>Journal of Clinical Medicine</i> , 2020, 9, 1245.	1.0	4
280	Clinical spectrum, prognosis and estimated prevalence of DNAJB11-kidney disease. <i>Kidney International</i> , 2020, 98, 476-487.	2.6	38
281	Emerging Methods and Resources for Biological Interrogation of Neuropsychiatric Polygenic Signal. <i>Biological Psychiatry</i> , 2021, 89, 41-53.	0.7	38
282	Detecting pathogenic variants in autoimmune diseases using high-throughput sequencing. <i>Immunology and Cell Biology</i> , 2021, 99, 146-156.	1.0	11
283	PredCID: prediction of driver frameshift indels in human cancer. <i>Briefings in Bioinformatics</i> , 2021, 22, .	3.2	29
284	Clinical and Genetic Findings in CTNNA1-Associated Macular Pattern Dystrophy. <i>Ophthalmology</i> , 2021, 128, 952-955.	2.5	8
285	Pathogenic 12-kb copy-neutral inversion in syndromic intellectual disability identified by high-fidelity long-read sequencing. <i>Genomics</i> , 2021, 113, 1044-1053.	1.3	11
286	Ensembl 2021. <i>Nucleic Acids Research</i> , 2021, 49, D884-D891.	6.5	1,231
287	A review on age-related cancer risks in <i>PTEN</i> hamartoma tumor syndrome. <i>Clinical Genetics</i> , 2021, 99, 219-225.	1.0	42
288	Misannotated Multi-Nucleotide Variants in Public Cancer Genomics Datasets Lead to Inaccurate Mutation Calls with Significant Implications. <i>Cancer Research</i> , 2021, 81, 282-288.	0.4	7

#	ARTICLE	IF	CITATIONS
289	<scp>Poirierâ€™s“Bienvenu</scp> neurodevelopmental syndrome: A report of a patient with a pathogenic variant in <scp><i>CSNK2B</i></scp> with abnormal linear growth. American Journal of Medical Genetics, Part A, 2021, 185, 539-543.	0.7	12
290	MobiDetails: online DNA variants interpretation. European Journal of Human Genetics, 2021, 29, 356-360.	1.4	34
291	<scp>Witteveenâ€™s“Kolk</scp> syndrome: The first patient from Turkey. American Journal of Medical Genetics, Part A, 2021, 185, 617-619.	0.7	3
292	Fabry disease: GLA deletion alters a canonical splice site in a family with neuropsychiatric manifestations. Metabolic Brain Disease, 2021, 36, 265-272.	1.4	5
293	Genetic variant burden and adverse outcomes in pediatric cardiomyopathy. Pediatric Research, 2021, 89, 1470-1476.	1.1	9
294	Deciphering the complexity of simple chromosomal insertions by genome sequencing. Human Genetics, 2021, 140, 361-380.	1.8	15
295	Integrative genomic analysis in African American children with asthma finds three novel loci associated with lung function. Genetic Epidemiology, 2021, 45, 190-208.	0.6	4
296	Prevalence of comprehensive <scp>DNA</scp> damage repair gene germline mutations in Chinese prostate cancer patients. International Journal of Cancer, 2021, 148, 673-681.	2.3	20
297	De novo variant in AMOTL1 in infant with cleft lip and palate, imperforate anus and dysmorphic features. American Journal of Medical Genetics, Part A, 2021, 185, 190-195.	0.7	3
298	Haploinsufficiency of <scp><i>ATP6V0C</i></scp> possibly underlies 16p13.3 deletions that cause microcephaly, seizures, and neurodevelopmental disorder. American Journal of Medical Genetics, Part A, 2021, 185, 196-202.	0.7	9
299	A review of UMAP in population genetics. Journal of Human Genetics, 2021, 66, 85-91.	1.1	73
300	Genome sequencing increases diagnostic yield in clinically diagnosed Alagille syndrome patients with previously negative test results. Genetics in Medicine, 2021, 23, 323-330.	1.1	17
301	Exome sequencing identifies novel missense and deletion variants in <scp><i>RTN4IP1</i></scp> associated with optic atrophy, global developmental delay, epilepsy, ataxia, and choreoathetosis. American Journal of Medical Genetics, Part A, 2021, 185, 203-207.	0.7	5
302	Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. Genetics in Medicine, 2021, 23, 341-351.	1.1	16
303	Population based frequency of naturally occurring lossâ€™ofâ€™function variants in genes associated with platelet disorders. Journal of Thrombosis and Haemostasis, 2021, 19, 248-254.	1.9	13
304	Open Targets Genetics: systematic identification of trait-associated genes using large-scale genetics and functional genomics. Nucleic Acids Research, 2021, 49, D1311-D1320.	6.5	295
305	Triâ€™Allelic Haplotypes Determine and Differentiate Functionally Normal Allele <i>CYP2D6*2</i> and Impaired Allele <i>CYP2D6*41</i>. Clinical Pharmacology and Therapeutics, 2021, 109, 1256-1264.	2.3	7
306	KALRN: A central regulator of synaptic function and synaptopathies. Gene, 2021, 768, 145306.	1.0	22

#	ARTICLE	IF	CITATIONS
307	Isolated dystonia: clinical and genetic updates. <i>Journal of Neural Transmission</i> , 2021, 128, 405-416.	1.4	18
308	<i>USP8</i> and <i>TP53</i> Drivers are Associated with CNV in a Corticotroph Adenoma Cohort Enriched for Aggressive Tumors. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, 826-842.	1.8	34
309	Missense substitutions at a conserved 14-3-3 binding site in HDAC4 cause a novel intellectual disability syndrome. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100015.	1.0	6
310	Recurrent prenatal PIEZO1-related lymphatic dysplasia: Expanding molecular and ultrasound findings. <i>European Journal of Medical Genetics</i> , 2021, 64, 104106.	0.7	7
311	Clinical and Molecular Characterization of Microphthalmia-associated Transcription Factor (MITF)-related Renal Cell Carcinoma. <i>Urology</i> , 2021, 149, 89-97.	0.5	22
312	A homozygous truncating variant in <i>CCDC186</i> in an individual with epileptic encephalopathy. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 278-283.	1.7	4
313	To NMD or Not To NMD: Nonsense-Mediated mRNA Decay in Cancer and Other Genetic Diseases. <i>Trends in Genetics</i> , 2021, 37, 657-668.	2.9	124
314	GPCRdb in 2021: integrating GPCR sequence, structure and function. <i>Nucleic Acids Research</i> , 2021, 49, D335-D343.	6.5	254
315	Heterozygous recurrent <i>HNF4A</i> variant p.Arg85Trp causes Fanconi renotubular syndrome 4 with maturity onset diabetes of the young, an autosomal dominant phenocopy of Fanconi Bickel syndrome with colobomas. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 566-570.	0.7	7
316	Benralizumab for Prednisone-Dependent Eosinophilic Asthma Associated With Novel STAT3 Loss of Function Mutation. <i>Chest</i> , 2021, 159, e181-e184.	0.4	6
317	Histone isoforms and the oncohistone code. <i>Current Opinion in Genetics and Development</i> , 2021, 67, 61-66.	1.5	15
318	c.451dupT in <i>KLKB1</i> is common in Nigerians, confirming a higher prevalence of severe prekallikrein deficiency in Africans compared to Europeans. <i>Journal of Thrombosis and Haemostasis</i> , 2021, 19, 147-152.	1.9	7
319	Genetic Ancestry Contributes to Somatic Mutations in Lung Cancers from Admixed Latin American Populations. <i>Cancer Discovery</i> , 2021, 11, 591-598.	7.7	69
320	Heterozygous loss of <i>WBP11</i> function causes multiple congenital defects in humans and mice. <i>Human Molecular Genetics</i> , 2021, 29, 3662-3678.	1.4	14
321	A Collaborative Effort to Define Classification Criteria for <i>ATM</i> Variants in Hereditary Cancer Patients. <i>Clinical Chemistry</i> , 2021, 67, 518-533.	1.5	14
322	Mutation analysis of TMEM family members for early-onset Parkinson's disease in Chinese population. <i>Neurobiology of Aging</i> , 2021, 101, 299.e1-299.e6.	1.5	14
323	Brain circuits at risk in psychiatric diseases and pharmacological pathways. <i>Therapie</i> , 2021, 76, 75-86.	0.6	2
324	Forkhead Transcription Factors in Health and Disease. <i>Trends in Genetics</i> , 2021, 37, 460-475.	2.9	65

#	ARTICLE	IF	CITATIONS
325	Revisiting the role of factor H in age-related macular degeneration: Insights from complement-mediated renal disease and rare genetic variants. <i>Survey of Ophthalmology</i> , 2021, 66, 378-401.	1.7	19
326	Biallelic variants in HPDL, encoding 4-hydroxyphenylpyruvate dioxygenase-like protein, lead to an infantile neurodegenerative condition. <i>Genetics in Medicine</i> , 2021, 23, 524-533.	1.1	17
327	Novel variants broaden the phenotypic spectrum of PLEKHG5-associated neuropathies. <i>European Journal of Neurology</i> , 2021, 28, 1344-1355.	1.7	4
328	Mutations in G Protein-Coupled Receptors: Mechanisms, Pathophysiology and Potential Therapeutic Approaches. <i>Pharmacological Reviews</i> , 2021, 73, 89-119.	7.1	60
329	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. <i>Genetics in Medicine</i> , 2021, 23, 384-395.	1.1	4
330	TrkA mediates effect of novel KIDINS220 mutation in human brain ventriculomegaly. <i>Human Molecular Genetics</i> , 2021, 29, 3757-3764.	1.4	10
331	Asymmetron: a toolkit for the identification of strand asymmetry patterns in biological sequences. <i>Nucleic Acids Research</i> , 2021, 49, e4-e4.	6.5	5
332	Evaluating variants classified as pathogenic in ClinVar in the DDD Study. <i>Genetics in Medicine</i> , 2021, 23, 571-575.	1.1	16
333	OncoVar: an integrated database and analysis platform for oncogenic driver variants in cancers. <i>Nucleic Acids Research</i> , 2021, 49, D1289-D1301.	6.5	64
334	Multi-parametric analysis of 57 SYNGAP1 variants reveal impacts on GTPase signaling, localization, and protein stability. <i>American Journal of Human Genetics</i> , 2021, 108, 148-162.	2.6	6
335	Clinical and structural insights into potential dominant negative triggers of proximal urea cycle disorders. <i>Biochimie</i> , 2021, 183, 89-99.	1.3	4
336	Genomic Characterization of <i>de novo</i> Metastatic Breast Cancer. <i>Clinical Cancer Research</i> , 2021, 27, 1105-1118.	3.2	24
337	Pharmacogenetics of inflammatory bowel disease. <i>Pharmacogenomics</i> , 2021, 22, 55-66.	0.6	8
338	<i>CHRNB1</i> -associated congenital myasthenia syndrome: Expanding the clinical spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 827-835.	0.7	6
339	New clinical and molecular evidence linking mutations in <i>ARSG</i> to Usher's syndrome type IV. <i>Human Mutation</i> , 2021, 42, 261-271.	1.1	23
340	GDF5 mutation case report and a systematic review of molecular and clinical spectrum: Expanding current knowledge on genotype-phenotype correlations. <i>Bone</i> , 2021, 144, 115803.	1.4	7
341	COVID-19: The Effect of Host Genetic Variations on Host-Virus Interactions. <i>Journal of Proteome Research</i> , 2021, 20, 139-153.	1.8	14
342	Computational Tools for Causal Inference in Genetics. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2021, 11, a039248.	2.9	3

#	ARTICLE	IF	CITATIONS
343	Collagen remodelling and plasma ascorbic acid levels in patients suspected of inherited bleeding disorders harbouring germline variants in collagen-related genes. <i>Haemophilia</i> , 2021, 27, e69-e77.	1.0	0
344	Description of the first mutation in the human tissue factor gene associated with a bleeding tendency. <i>Journal of Thrombosis and Haemostasis</i> , 2021, 19, 3-6.	1.9	1
345	Modeling Human TBX5 Haploinsufficiency Predicts Regulatory Networks for Congenital Heart Disease. <i>Developmental Cell</i> , 2021, 56, 292-309.e9.	3.1	63
346	The New "Wholly Trinity" in the Diagnosis and Management of Inborn Errors of Immunity. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 613-625.	2.0	10
347	Ichthyosis prematurity syndrome in two Omani siblings, caused by homozygous c.1AA>ÂG mutation in the FATP4 gene. <i>International Journal of Dermatology</i> , 2021, 60, 368-371.	0.5	3
348	Benefits of clinical criteria and high-throughput sequencing for diagnosing children with syndromic craniosynostosis. <i>European Journal of Human Genetics</i> , 2021, 29, 920-929.	1.4	13
349	Spectrum-frequency and genotype-phenotype analysis of rhodopsin variants. <i>Experimental Eye Research</i> , 2021, 203, 108405.	1.2	14
350	The UCSC Genome Browser database: 2021 update. <i>Nucleic Acids Research</i> , 2021, 49, D1046-D1057.	6.5	354
351	TMEM218 dysfunction causes ciliopathies, including Joubert and Meckel syndromes. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100016.	1.0	7
352	Pathogenic paternally inherited NLGN4X deletion in a female with autism spectrum disorder: Clinical, cytogenetic, and molecular characterization. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 894-900.	0.7	5
353	Flype: Software for enabling personalized medicine. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2021, 187, 37-47.	0.7	8
354	Characterization of the <sc><i>GABRB2</i></sc>-Associated Neurodevelopmental Disorders. <i>Annals of Neurology</i> , 2021, 89, 573-586.	2.8	14
355	Update of the Pompe variant database for the prediction of clinical phenotypes: Novel disease-associated variants, common sequence variants, and results from newborn screening. <i>Human Mutation</i> , 2021, 42, 119-134.	1.1	19
356	Functional and clinical implications of genetic structure in 1686 Italian exomes. <i>Human Mutation</i> , 2021, 42, 272-289.	1.1	5
357	Two cases of carbonic anhydrase <sc><i>VA</i></sc> deficiency- An ultrarare metabolic decompensation syndrome presenting with hyperammonemia, lactic acidosis, ketonuria, and good clinical outcome. <i>JIMD Reports</i> , 2021, 57, 9-14.	0.7	7
358	Early truncation of the N-terminal variable region of <i>EYA4</i> gene causes dominant hearing loss without cardiac phenotype. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1569.	0.6	7
359	Dominant variants in <sc><i>PRR12</i></sc> result in unilateral or bilateral complex microphthalmia. <i>Clinical Genetics</i> , 2021, 99, 437-442.	1.0	5
360	The prevalent I686T human variant and loss-of-function mutations in the cardiomyocyte-specific kinase gene TNNI3K cause adverse contractility and concentric remodeling in mice. <i>Human Molecular Genetics</i> , 2021, 29, 3504-3515.	1.4	9

#	ARTICLE	IF	CITATIONS
361	<i>ECHS1</i> disease in two unrelated families of Samoan descent: Common variant rare disorder. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 157-167.	0.7	13
362	Molecular Analysis of the <i>CYP11B2</i> Gene in 62 Patients with Hypoaldosteronism Due to Aldosterone Synthase Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e182-e191.	1.8	6
363	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 119-133.	0.7	17
364	An expanded analysis framework for multivariate GWAS connects inflammatory biomarkers to functional variants and disease. <i>European Journal of Human Genetics</i> , 2021, 29, 309-324.	1.4	19
365	Variable expressivity in patients with autosomal recessive retinitis pigmentosa associated with the gene <i>CNGB1</i> . <i>Ophthalmic Genetics</i> , 2021, 42, 15-22.	0.5	7
366	Variants in <i>NAA15</i> cause pediatric hypertrophic cardiomyopathy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 228-233.	0.7	10
367	Analysis of DNA variants in miRNAs and miRNA 3'UTR binding sites in female infertility patients. <i>Laboratory Investigation</i> , 2021, 101, 503-512.	1.7	10
368	De novo variants in <i>CELF2</i> that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. <i>Human Mutation</i> , 2021, 42, 66-76.	1.1	16
369	Heterozygous variants that disturb the transcriptional repressor activity of <i>FOXP4</i> cause a developmental disorder with speech/language delays and multiple congenital abnormalities. <i>Genetics in Medicine</i> , 2021, 23, 534-542.	1.1	17
370	Germline variants in <i>HEY2</i> functional domains lead to congenital heart defects and thoracic aortic aneurysms. <i>Genetics in Medicine</i> , 2021, 23, 103-110.	1.1	7
371	Telangiectasia-ectodermal dysplasia-brachydactyly-cardiac anomaly syndrome is caused by de novo mutations in <i>protein kinase D1</i> . <i>Journal of Medical Genetics</i> , 2021, 58, 415-421.	1.5	5
372	Functional evaluation of 16 <i>SCHAD</i> missense variants: Only amino acid substitutions causing congenital hyperinsulinism of infancy lead to loss of function phenotypes in vitro. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 240-252.	1.7	1
373	rs641738C>T near <i>MBOAT7</i> is associated with liver fat, ALT and fibrosis in NAFLD: A meta-analysis. <i>Journal of Hepatology</i> , 2021, 74, 20-30.	1.8	77
374	Non-coding variants in <i>MYH11</i> , <i>FZD3</i> , and <i>SORCS3</i> are associated with dementia in women. <i>Alzheimer's and Dementia</i> , 2021, 17, 215-225.	0.4	18
375	Integrated genetic and epigenetic analyses uncover <i>MSI2</i> association with allergic inflammation. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 147, 1453-1463.	1.5	11
376	Deficiency of Adenosine Deaminase 2 in Adults and Children: Experience From India. <i>Arthritis and Rheumatology</i> , 2021, 73, 276-285.	2.9	43
377	Cancer health disparities in racial/ethnic minorities in the United States. <i>British Journal of Cancer</i> , 2021, 124, 315-332.	2.9	447
378	Effect Sizes of Deletions and Duplications on Autism Risk Across the Genome. <i>American Journal of Psychiatry</i> , 2021, 178, 87-98.	4.0	50

#	ARTICLE	IF	CITATIONS
379	Loss-of-function variants in <i>POT1</i> predispose to uveal melanoma. <i>Journal of Medical Genetics</i> , 2021, 58, 234-236.	1.5	3
380	Novel RPL13 Variants and Variable Clinical Expressivity in a Human Ribosomopathy With Spondyloepimetaphyseal Dysplasia. <i>Journal of Bone and Mineral Research</i> , 2020, 36, 283-297.	3.1	12
381	Search for Genetic Predictors of Adult Autoimmune Polyendocrine Syndrome in Monozygotic Twins. <i>Clinical Medicine Insights: Endocrinology and Diabetes</i> , 2021, 14, 117955142110097.	1.0	1
382	Missense3D-DB web catalogue: an atom-based analysis and repository of 4M human protein-coding genetic variants. <i>Human Genetics</i> , 2021, 140, 805-812.	1.8	39
383	Phenotypic continuum between Waardenburg syndrome and idiopathic hypogonadotropic hypogonadism in humans with SOX10 variants. <i>Genetics in Medicine</i> , 2021, 23, 629-636.	1.1	9
384	Whole exome sequencing and trio analysis to broaden the variant spectrum of genes in idiopathic hypogonadotropic hypogonadism. <i>Asian Journal of Andrology</i> , 2021, 23, 288.	0.8	5
385	Mutational Landscape for Indian Hereditary Breast and Ovarian Cancer Cohort Suggests Need for Identifying Population Specific Genes and Biomarkers for Screening. <i>Frontiers in Oncology</i> , 2020, 10, 568786.	1.3	6
386	Genetic Etiology of Left-Sided Obstructive Heart Lesions: A Story in Development. <i>Journal of the American Heart Association</i> , 2021, 10, e019006.	1.6	23
388	Unexpected role of SIX1 variants in craniosynostosis: expanding the phenotype of SIX1-related disorders. <i>Journal of Medical Genetics</i> , 2021, , jmedgenet-2020-107459.	1.5	5
390	Cardiomyopathic troponin mutations predominantly occur at its interface with actin and tropomyosin. <i>Journal of General Physiology</i> , 2021, 153, .	0.9	24
391	Congenital cervical spine malformation due to allelic <i>RIPPLY2</i> variants in spondylocostal dysostosis type 6. <i>Clinical Genetics</i> , 2021, 99, 565-571.	1.0	4
392	Loss of MANF Causes Childhood-Onset Syndromic Diabetes Due to Increased Endoplasmic Reticulum Stress. <i>Diabetes</i> , 2021, 70, 1006-1018.	0.3	37
393	Investigating the causal role of MRE11A p.E506* in breast and ovarian cancer. <i>Scientific Reports</i> , 2021, 11, 2409.	1.6	5
394	Challenges and opportunities in rare diseases research. , 2021, , 263-284.		0
396	Mutations causing Lopes-Maciel-Rodan syndrome are huntingtin hypomorphs. <i>Human Molecular Genetics</i> , 2021, 30, 135-148.	1.4	24
397	Synaptic processes and immune-related pathways implicated in Tourette syndrome. <i>Translational Psychiatry</i> , 2021, 11, 56.	2.4	31
398	Evidence that <i>FGFRL1</i> contributes to congenital diaphragmatic hernia development in humans. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 836-840.	0.7	8
399	A rare coding mutation in the MAST2 gene causes venous thrombosis in a French family with unexplained thrombophilia: The Breizh MAST2 Arg89Gln variant. <i>PLoS Genetics</i> , 2021, 17, e1009284.	1.5	2

#	ARTICLE	IF	CITATIONS
400	IsoMiRmap: fast, deterministic and exhaustive mining of isomiRs from short RNA-seq datasets. <i>Bioinformatics</i> , 2021, 37, 1828-1838.	1.8	11
401	Detection of aberrant splicing events in RNA-seq data using FRASER. <i>Nature Communications</i> , 2021, 12, 529.	5.8	78
402	The Parkinson's Disease <sc>DNA</sc> Variant Browser. <i>Movement Disorders</i> , 2021, 36, 1250-1258.	2.2	11
403	Herpes simplex encephalitis in a patient with a distinctive form of inherited IFNAR1 deficiency. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	64
404	Enhancer redundancy in development and disease. <i>Nature Reviews Genetics</i> , 2021, 22, 324-336.	7.7	128
405	Diverse viral proteases activate the NLRP1 inflammasome. <i>ELife</i> , 2021, 10, .	2.8	100
406	The Molecular Basis of Glucose Galactose Malabsorption in a Large Swedish Pedigree. <i>Function</i> , 2021, 2, zqab040.	1.1	4
407	Whole-exome sequencing reveals a role of HTRA1 and EGFL8 in brain white matter hyperintensities. <i>Brain</i> , 2021, 144, 2670-2682.	3.7	21
409	Prevalence of <i>MYOC</i> risk variants for glaucoma in different populations. <i>Acta Ophthalmologica</i> , 2021, 99, e1090-e1097.	0.6	7
410	Genome diversity in Ukraine. <i>GigaScience</i> , 2021, 10, .	3.3	9
411	Increasing involvement of CAPN1 variants in spastic ataxias and phenotype-genotype correlations. <i>Neurogenetics</i> , 2021, 22, 71-79.	0.7	11
414	239-kb Microdeletion Spanning <i>KMT2E</i> in a Child with Developmental Delay: Further Delineation of the Phenotype. <i>Molecular Syndromology</i> , 2021, 12, 321-326.	0.3	0
415	Rare genetic variants prioritize molecular pathways for semaphorin interactions in Alzheimer's disease patients. <i>Biotechnology and Biotechnological Equipment</i> , 2021, 35, 1256-1262.	0.5	3
416	The Mutation Spectrum of Maturity Onset Diabetes of the Young (MODY)-Associated Genes among Western Siberia Patients. <i>Journal of Personalized Medicine</i> , 2021, 11, 57.	1.1	12
417	Comprehensive analysis of important pharmacogenes in Koreans using the DMET platform. <i>Translational and Clinical Pharmacology</i> , 2021, 29, 135.	0.3	3
419	Reference flow: reducing reference bias using multiple population genomes. <i>Genome Biology</i> , 2021, 22, 8.	3.8	44
420	Patterns of de novo tandem repeat mutations and their role in autism. <i>Nature</i> , 2021, 589, 246-250.	13.7	114
421	A semisupervised model to predict regulatory effects of genetic variants at single nucleotide resolution using massively parallel reporter assays. <i>Bioinformatics</i> , 2021, , .	1.8	2

#	ARTICLE	IF	CITATIONS
423	The Desmin (DES) Mutation p.A337P Is Associated with Left-Ventricular Non-Compaction Cardiomyopathy. <i>Genes</i> , 2021, 12, 121.	1.0	26
424	Human genetic variants disrupt RGS14 nuclear shuttling and regulation of LTP in hippocampal neurons. <i>Journal of Biological Chemistry</i> , 2021, 296, 100024.	1.6	9
426	Breast Tumor Microenvironment in Black Women: A Distinct Signature of CD8+ T-Cell Exhaustion. <i>Journal of the National Cancer Institute</i> , 2021, 113, 1036-1043.	3.0	50
427	Hereditäre Netzhautdystrophien aufgrund von RPE65-Varianten: Von der genetischen Diagnostik zur Therapie. <i>Karger Kompass Ophthalmologie</i> , 2021, 7, 115-123.	0.0	0
428	The influence of evolutionary history on human health and disease. <i>Nature Reviews Genetics</i> , 2021, 22, 269-283.	7.7	133
429	Deep neural networks identify sequence context features predictive of transcription factor binding. <i>Nature Machine Intelligence</i> , 2021, 3, 172-180.	8.3	55
430	Lost in Translation: Lack of CD4 Expression due to a Novel Genetic Defect. <i>Journal of Infectious Diseases</i> , 2021, 223, 645-654.	1.9	10
431	Exploiting the GTEx resources to decipher the mechanisms at GWAS loci. <i>Genome Biology</i> , 2021, 22, 49.	3.8	150
432	Biallelic variants in <i>SLC38A3</i> encoding a glutamine transporter cause epileptic encephalopathy. <i>Brain</i> , 2022, 145, 909-924.	3.7	17
433	Genotyping of Eight Human Platelet Antigen Systems in Serbian Blood Donors: Foundation for Platelet Apheresis Registry. <i>Transfusion Medicine and Hemotherapy</i> , 2021, 48, 228-233.	0.7	5
434	Common allotypes of ER aminopeptidase 1 have substrate-dependent and highly variable enzymatic properties. <i>Journal of Biological Chemistry</i> , 2021, 296, 100443.	1.6	20
436	The origins and consequences of UPF1 variants in pancreatic adenosquamous carcinoma. <i>ELife</i> , 2021, 10, .	2.8	8
437	Functional Common and Rare <i>ERBB2</i> Germline Variants Cooperate in Familial and Sporadic Cancer Susceptibility. <i>Cancer Prevention Research</i> , 2021, 14, 441-454.	0.7	0
439	Biallelic truncating variants in <i>MAPKAPK5</i> cause a new developmental disorder involving neurological, cardiac, and facial anomalies combined with synpolydactyly. <i>Genetics in Medicine</i> , 2021, 23, 679-688.	1.1	7
440	Ultrasound and genetic detection of fetal hypertrophic cardiomyopathy in second trimester of pregnancy. <i>BMJ Case Reports</i> , 2021, 14, e239773.	0.2	0
442	Transcription-translation error: In-silico investigation of the structural and functional impact of deleterious single nucleotide polymorphisms in <i>GULP1</i> gene. <i>Informatics in Medicine Unlocked</i> , 2021, 22, 100503.	1.9	0
444	The phenotypic spectrum in a patient with Glycine to Serine mutation in the <i>COL2A1</i> gene: overview study. <i>AIMS Molecular Science</i> , 2021, 8, 76-85.	0.3	0
445	The landscape of somatic mutation in cerebral cortex of autistic and neurotypical individuals revealed by ultra-deep whole-genome sequencing. <i>Nature Neuroscience</i> , 2021, 24, 176-185.	7.1	73

#	ARTICLE	IF	CITATIONS
446	Rare functional missense variants in CACNA1H: What can we learn from Writerâ€™s cramp?. Molecular Brain, 2021, 14, 18.	1.3	3
447	Efficient phasing and imputation of low-coverage sequencing data using large reference panels. Nature Genetics, 2021, 53, 120-126.	9.4	184
448	Exome-wide evaluation of rare coding variants using electronic health records identifies new geneâ€™phenotype associations. Nature Medicine, 2021, 27, 66-72.	15.2	44
449	Deleterious variants in genes regulating mammalian reproduction in Neanderthals, Denisovans and extant humans. Human Reproduction, 2021, 36, 734-755.	0.4	5
451	To Be or No B2: A Rare Cause of Stridor and Weakness in a Toddler. Child Neurology Open, 2021, 8, 2329048X2110307.	0.5	2
453	Next-generation sequencing analysis suggests varied multistep mutational pathogenesis for endocrine mucin-producing sweat gland carcinoma with comments on INSM1 and MUC2 suggesting a conjunctival origin. Journal of the American Academy of Dermatology, 2022, 86, 1072-1079.	0.6	8
454	Human variant of scavenger receptor BI (R174C) exhibits impaired cholesterol transport functions. Journal of Lipid Research, 2021, 62, 100045.	2.0	8
455	Detection of aberrant gene expression events in RNA sequencing data. Nature Protocols, 2021, 16, 1276-1296.	5.5	58
456	Personalized Medicine Through GPCR Pharmacogenomics. , 2021, , .		2
457	PIGF deficiency causes a phenotype overlapping with DOORS syndrome. Human Genetics, 2021, 140, 879-884.	1.8	2
458	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	3.9	87
459	Hunting for the perfect test: Neuromuscular diagnosis in the age of genomic bounty. Muscle and Nerve, 2021, 63, 282-284.	1.0	1
461	Genotypeâ€™phenotype correlation in <sc><i>GNB1</i></sc>â€™related neurodevelopmental disorder: Potential association of p.<sc>Leu95Pro</sc> with cleft palate. American Journal of Medical Genetics, Part A, 2021, 185, 1341-1343.	0.7	5
462	STOX1 deficiency is associated with renin-mediated gestational hypertension and placental defects. JCI Insight, 2021, 6, .	2.3	4
463	CORONAVIRUSES â€™ HOW PROTEIN INTERACTIONS CHANGED OUR PERCEPTION OF THE WORLD. Postepy Mikrobiologii, 2021, 60, 121-135.	0.1	0
464	Functional evaluation of human ion channel variants using automated electrophysiology. Methods in Enzymology, 2021, 654, 383-405.	0.4	9
465	Autism Spectrum Disorder Genetics and the Search for Pathological Mechanisms. American Journal of Psychiatry, 2021, 178, 30-38.	4.0	70
466	MVP predicts theâ€™pathogenicity of missense variants by deep learning. Nature Communications, 2021, 12, 510.	5.8	85

#	ARTICLE	IF	CITATIONS
467	Negative selection on human genes underlying inborn errors depends on disease outcome and both the mode and mechanism of inheritance. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	33
468	Biallelic SORD pathogenic variants cause Chinese patients with distal hereditary motor neuropathy. Npj Genomic Medicine, 2021, 6, 1.	1.7	32
469	Reprogramming translation for gene therapy. Progress in Molecular Biology and Translational Science, 2021, 182, 439-476.	0.9	5
470	Expression defect of the rare variant/Brugada mutation R1512W depends upon the SCN5A splice variant background and can be rescued by mexiletine and the common polymorphism H558R. Channels, 2021, 15, 253-261.	1.5	3
471	Distinguishing between PTEN clinical phenotypes through mutation analysis. Computational and Structural Biotechnology Journal, 2021, 19, 3097-3109.	1.9	20
473	BARD1 Pathogenic Variants Are Associated with Triple-Negative Breast Cancer in a Spanish Hereditary Breast and Ovarian Cancer Cohort. Genes, 2021, 12, 150.	1.0	11
474	Methods and feasibility study for exome sequencing as a universal second-tier test in newborn screening. Genetics in Medicine, 2021, 23, 767-776.	1.1	8
475	Probing the aggregated effects of purifying selection per individual on 1,380 medical phenotypes in the UK Biobank. PLoS Genetics, 2021, 17, e1009337.	1.5	2
477	Association of rs12722 COL5A1 with pulmonary tuberculosis: a preliminary case-control study in a Kazakhstani population. Molecular Biology Reports, 2021, 48, 691-699.	1.0	2
478	Regulation of purine metabolism connects KCTD13 to a metabolic disorder with autistic features. IScience, 2021, 24, 101935.	1.9	7
479	Biallelic loss-of-function HACD1 variants are a bona fide cause of congenital myopathy. Clinical Genetics, 2021, 99, 513-518.	1.0	5
480	Genomic imbalances in the placenta are associated with poor fetal growth. Molecular Medicine, 2021, 27, 3.	1.9	13
481	Massively parallel functional testing of MSH2 missense variants conferring Lynch syndrome risk. American Journal of Human Genetics, 2021, 108, 163-175.	2.6	66
482	Familial neurohypophyseal diabetes insipidus: clinical, genetic and functional studies of novel mutations in the arginine vasopressin gene. Pituitary, 2021, 24, 400-411.	1.6	4
483	Breast cancer and DDT: putative interactions, associated gene alterations, and molecular pathways. Environmental Science and Pollution Research, 2021, 28, 27162-27173.	2.7	2
484	Dominant and sporadic de novo disorders. , 2021, , 117-135.		0
485	Methods to Study Genomic DNA Sequence Variation. , 2021, , 59-92.		0
486	Genetics of Cellular Immunodeficiencies. Rare Diseases of the Immune System, 2021, , 5-24.	0.1	0

#	ARTICLE	IF	CITATIONS
487	A platform for curated products from novel open reading frames prompts reinterpretation of disease variants. <i>Genome Research</i> , 2021, 31, 327-336.	2.4	17
488	SCISSOR: a framework for identifying structural changes in RNA transcripts. <i>Nature Communications</i> , 2021, 12, 286.	5.8	10
489	TMSNP: a web server to predict pathogenesis of missense mutations in the transmembrane region of membrane proteins. <i>NAR Genomics and Bioinformatics</i> , 2021, 3, lqab008.	1.5	7
490	Missense variants in the N-terminal domain of the A isoform of FHF2/FGF13 cause an X-linked developmental and epileptic encephalopathy. <i>American Journal of Human Genetics</i> , 2021, 108, 176-185.	2.6	20
491	Long-read transcriptome sequencing reveals abundant promoter diversity in distinct molecular subtypes of gastric cancer. <i>Genome Biology</i> , 2021, 22, 44.	3.8	46
492	Genome-wide landscape of RNA-binding protein target site dysregulation reveals a major impact on psychiatric disorder risk. <i>Nature Genetics</i> , 2021, 53, 166-173.	9.4	49
493	A pathogenic UFSP2 variant in an autosomal recessive form of pediatric neurodevelopmental anomalies and epilepsy. <i>Genetics in Medicine</i> , 2021, 23, 900-908.	1.1	14
494	Generation of human induced pluripotent stem cell (iPSC) lines (UUMC <i>Bi</i> 001-A, UUMC <i>Bi</i> 002-A) from two healthy donors. <i>Stem Cell Research</i> , 2021, 50, 102114.	0.3	1
495	Identification and characterization of novel <i>ACD</i> variants: modulation of TPP1 protein level offsets the impact of germline loss-of-function variants on telomere length. <i>Journal of Physical Education and Sports Management</i> , 2021, 7, a005454.	0.5	8
496	An emerging spectrum of variants and clinical features in <i>KCNMA1</i> -linked channelopathy. <i>Channels</i> , 2021, 15, 447-464.	1.5	41
497	Genetic variant effect prediction by supervised nonnegative matrix tri-factorization. <i>Molecular Omics</i> , 2021, 17, 740-751.	1.4	1
498	Genome-Wide Association Study of Korean Asthmatics: A Comparison With UK Asthmatics. <i>Allergy, Asthma and Immunology Research</i> , 2021, 13, 609.	1.1	4
499	X-linked serotonin 2C receptor is associated with a non-canonical pathway for sudden unexpected death in epilepsy. <i>Brain Communications</i> , 2021, 3, fcab149.	1.5	13
502	Female and male perspectives on male partner roles in expanded carrier screening. <i>Journal of Assisted Reproduction and Genetics</i> , 2021, 38, 375-385.	1.2	5
503	Pulmonary arterial hypertension-associated genetic variants in combined postcapillary and precapillary pulmonary hypertension: a case report. <i>Pulmonary Circulation</i> , 2021, 11, 1-5.	0.8	0
504	Founder variants and population genomes—Toward precision medicine. <i>Advances in Genetics</i> , 2021, 107, 121-152.	0.8	2
505	Whole-genome sequencing reveals <i>KRTAP1-1</i> as a novel genetic variant associated with antidepressant treatment outcomes. <i>Scientific Reports</i> , 2021, 11, 4552.	1.6	3
506	Akt Is S-Palmitoylated: A New Layer of Regulation for Akt. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 626404.	1.8	20

#	ARTICLE	IF	CITATIONS
508	Alazami syndrome: Report of three Indian patients with phenotypic spectrum from adolescence to adulthood. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1606-1609.	0.7	4
510	A multiplex PCR amplicon sequencing assay to screen genetic hearing loss variants in newborns. <i>BMC Medical Genomics</i> , 2021, 14, 61.	0.7	5
511	Genotype and Long-term Clinical Course of Bietti Crystalline Dystrophy in Korean and Japanese Patients. <i>Ophthalmology Retina</i> , 2021, 5, 1269-1279.	1.2	6
512	Altered structure and dynamics of pathogenic cytochrome <i>c</i> variants correlate with increased apoptotic activity. <i>Biochemical Journal</i> , 2021, 478, 669-684.	1.7	8
514	A Workflow for Selection of Single Nucleotide Polymorphic Markers for Studying of Genetics of Ischemic Stroke Outcomes. <i>Genes</i> , 2021, 12, 328.	1.0	6
515	Autosomal dominant familial acanthosis nigricans caused by a C-terminal nonsense mutation of FGFR3. <i>Journal of Human Genetics</i> , 2021, 66, 831-834.	1.1	3
516	SLC22A4 Gene in Hereditary Non-syndromic Hearing Loss: Recurrence and Incomplete Penetrance of the p.C113Y Mutation in Northwest Africa. <i>Frontiers in Genetics</i> , 2021, 12, 606630.	1.1	7
517	Mutations in PRDM15 Are a Novel Cause of Galloway-Mowat Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2021, 32, 580-596.	3.0	15
518	Interleukin-1 (IL-1) receptor cleavage by the rhomboid protease RHBDL2 induces IL-1 trans-signaling. <i>FASEB Journal</i> , 2021, 35, e21380.	0.2	20
519	Clinical and laboratory reporting impact of ACMG-AMP and modified ClinGen variant classification frameworks in MYH7-related cardiomyopathy. <i>Genetics in Medicine</i> , 2021, 23, 1108-1115.	1.1	14
520	Rare Recurrent Variants in Noncoding Regions Impact Attention-Deficit Hyperactivity Disorder (ADHD) Gene Networks in Children of both African American and European American Ancestry. <i>Genes</i> , 2021, 12, 310.	1.0	10
521	Network-Based Target Prioritization and Drug Candidate Identification for Multiple Sclerosis: From Analyzing Omics Data to Druggability Simulations. <i>ACS Chemical Neuroscience</i> , 2021, 12, 917-929.	1.7	5
522	GeneBreaker: Variant simulation to improve the diagnosis of Mendelian rare genetic diseases. <i>Human Mutation</i> , 2021, 42, 346-358.	1.1	3
523	ACE2 and FURIN variants are potential predictors of SARS-CoV-2 outcome: A time to implement precision medicine against COVID-19. <i>Heliyon</i> , 2021, 7, e06133.	1.4	26
524	A Novel Germline TP53 Mutation in a Patient With Li-Fraumeni Syndrome: Resolving a Variant of Uncertain Significance. <i>Journal of Pediatric Hematology/Oncology</i> , 2021, 43, e1220-e1222.	0.3	1
525	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. <i>Genetics in Medicine</i> , 2021, 23, 1075-1085.	1.1	16
526	Structural genetics of circulating variants affecting the SARS-CoV-2 spike/human ACE2 complex. <i>Journal of Biomolecular Structure and Dynamics</i> , 2022, 40, 6545-6555.	2.0	40
527	Clinical characterization and identification of rare genetic variants in atypical hemolytic uremic syndrome: A Swedish retrospective observational study. <i>Therapeutic Apheresis and Dialysis</i> , 2021, 25, 988-1000.	0.4	4

#	ARTICLE	IF	CITATIONS
528	Prevalence and cardiometabolic correlates of ketohexokinase gene variants among UK Biobank participants. <i>PLoS ONE</i> , 2021, 16, e0247683.	1.1	3
529	Pinpointing the PRDM9-PRDM7 Gene Duplication Event During Primate Divergence. <i>Frontiers in Genetics</i> , 2021, 12, 593725.	1.1	0
530	A phenotypically diverse family with an atypical 22q11.2 deletion due to an unbalanced 18q23;22q11.2 translocation. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1532-1537.	0.7	1
531	Unexplained sudden death: next-generation sequencing to the rescue?. <i>Europace</i> , 2021, 23, 327-328.	0.7	3
532	Loss-of-Function Variants in the Tumor-Suppressor Gene <i>PTPN14</i> Confer Increased Cancer Risk. <i>Cancer Research</i> , 2021, 81, 1954-1964.	0.4	15
533	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with <i>SATB1</i> dysfunction. <i>American Journal of Human Genetics</i> , 2021, 108, 346-356.	2.6	30
534	The genomic landscape of pediatric rheumatology disorders in the Middle East. <i>Human Mutation</i> , 2021, 42, e1-e14.	1.1	12
535	Allelic Dropout Is a Common Phenomenon That Reduces the Diagnostic Yield of PCR-Based Sequencing of Targeted Gene Panels. <i>Frontiers in Genetics</i> , 2021, 12, 620337.	1.1	26
536	ClinSV: clinical grade structural and copy number variant detection from whole genome sequencing data. <i>Genome Medicine</i> , 2021, 13, 32.	3.6	36
537	Life-Threatening Influenza, Hemophagocytic Lymphohistiocytosis and Probable Vaccine-Strain Varicella in a Novel Case of Homozygous <i>STAT2</i> Deficiency. <i>Frontiers in Immunology</i> , 2020, 11, 624415.	2.2	21
538	CADD-Splice—improving genome-wide variant effect prediction using deep learning-derived splice scores. <i>Genome Medicine</i> , 2021, 13, 31.	3.6	375
539	Comprehensive Mutational Analysis of the <i>BRCA1</i> -Associated DNA Helicase and Tumor-Suppressor <i>FANCI/BACH1/BRIP1</i> . <i>Molecular Cancer Research</i> , 2021, 19, 1015-1025.	1.5	15
541	Neurodevelopmental disorder in an Egyptian family with a biallelic <i>ALKBH8</i> variant. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1288-1293.	0.7	13
542	Alteration of DNA mismatch repair capacity underlying the co-occurrence of non-small-cell lung cancer and nonmedullary thyroid cancer. <i>Scientific Reports</i> , 2021, 11, 3597.	1.6	6
543	The zebrafish <i>grime</i> mutant uncovers an evolutionarily conserved role for <i>Tmem161b</i> in the control of cardiac rhythm. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	12
544	Whole genome sequencing of 45 Japanese patients with intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1468-1480.	0.7	13
545	Breaching the Bacterial Envelope: The Pivotal Role of Perforin-2 (<i>MPEG1</i>) Within Phagocytes. <i>Frontiers in Immunology</i> , 2021, 12, 597951.	2.2	9
546	The population genomics of adaptive loss of function. <i>Heredity</i> , 2021, 126, 383-395.	1.2	33

#	ARTICLE	IF	CITATIONS
548	Insights into the pathogenicity of missense variants in the forkhead domain of FOX proteins underlying Mendelian disorders. <i>Human Genetics</i> , 2021, 140, 999-1010.	1.8	2
549	Optic Atrophy and Inner Retinal Thinning in CACNA1F-Related Congenital Stationary Night Blindness. <i>Genes</i> , 2021, 12, 330.	1.0	6
550	RCL1 copy number variants are associated with a range of neuropsychiatric phenotypes. <i>Molecular Psychiatry</i> , 2021, 26, 1706-1718.	4.1	10
551	Role of an Atypical Cadherin Gene, <i>Cdh23</i> in Prepulse Inhibition, and Implication of <i>CDH23</i> in Schizophrenia. <i>Schizophrenia Bulletin</i> , 2021, 47, 1190-1200.	2.3	7
552	Evidence for Non-Mendelian Inheritance in Spastic Paraplegia 7. <i>Movement Disorders</i> , 2021, 36, 1664-1675.	2.2	11
553	Defects in the cytoplasmic assembly of axonemal dynein arms cause morphological abnormalities and dysmotility in sperm cells leading to male infertility. <i>PLoS Genetics</i> , 2021, 17, e1009306.	1.5	50
554	PopDel identifies medium-size deletions simultaneously in tens of thousands of genomes. <i>Nature Communications</i> , 2021, 12, 730.	5.8	9
555	Functional analysis of a gene-edited mouse model to gain insights into the disease mechanisms of a titin missense variant. <i>Basic Research in Cardiology</i> , 2021, 116, 14.	2.5	16
556	Dyshomeostatic modulation of Ca ²⁺ -activated K ⁺ channels in a human neuronal model of KCNQ2 encephalopathy. <i>ELife</i> , 2021, 10, .	2.8	23
558	Unravelling the Sequential Interplay of Mutational Mechanisms during Clonal Evolution in Relapsed Pediatric Acute Lymphoblastic Leukemia. <i>Genes</i> , 2021, 12, 214.	1.0	6
561	Location matters – Genotype-phenotype correlation in LRSAM1 mutations associated with rare Charcot-Marie-Tooth neuropathy CMT2P. <i>Neuromuscular Disorders</i> , 2021, 31, 123-133.	0.3	2
563	A heterozygous mutation in the <i>CCDC88C</i> gene likely causes early-onset pure hereditary spastic paraplegia: a case report. <i>BMC Neurology</i> , 2021, 21, 78.	0.8	8
564	Germline and Somatic Whole-Exome Sequencing Identifies New Candidate Genes Involved in Familial Predisposition to Serrated Polyposis Syndrome. <i>Cancers</i> , 2021, 13, 929.	1.7	12
565	Huntington's Disease Pathogenesis: Two Sequential Components. <i>Journal of Huntington's Disease</i> , 2021, 10, 35-51.	0.9	49
566	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. <i>American Journal of Human Genetics</i> , 2021, 108, 357-367.	2.6	14
567	The integrity of the U12 snRNA 3' stem-loop is necessary for its overall stability. <i>Nucleic Acids Research</i> , 2021, 49, 2835-2847.	6.5	7
568	ERBB4 exonic deletions on chromosome 2q34 in patients with intellectual disability or epilepsy. <i>European Journal of Human Genetics</i> , 2021, 29, 1377-1383.	1.4	9
569	DNA Mismatch Repair and its Role in Huntington's Disease. <i>Journal of Huntington's Disease</i> , 2021, 10, 75-94.	0.9	47

#	ARTICLE	IF	CITATIONS
570	StellarPGx: A Nextflow Pipeline for Calling Star Alleles in Cytochrome P450 Genes. <i>Clinical Pharmacology and Therapeutics</i> , 2021, 110, 741-749.	2.3	28
571	Mapping Out Autoimmunity Control in Primary Immune Regulatory Disorders. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 653-659.	2.0	3
572	Functional significance of germline EPAS1 variants. <i>Endocrine-Related Cancer</i> , 2021, 28, 97-109.	1.6	6
574	Protein Stability Perturbation Contributes to the Loss of Function in Haploinsufficient Genes. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 620793.	1.6	20
575	Further delineation of the <sc><i>NTHL1</i></sc> associated syndrome: A report from the French Oncogenetic Consortium. <i>Clinical Genetics</i> , 2021, 99, 662-672.	1.0	7
576	Clinicopathological and Genomic Characterization of a Simmental Calf with Generalized Bovine Juvenile Angiomatosis. <i>Animals</i> , 2021, 11, 624.	1.0	3
577	APC Splicing Mutations Leading to In-Frame Exon 12 or Exon 13 Skipping Are Rare Events in FAP Pathogenesis and Define the Clinical Outcome. <i>Genes</i> , 2021, 12, 353.	1.0	2
578	A unique missense variant in the E1A-binding protein P400 gene is implicated in schizophrenia by whole-exome sequencing and mutant mouse models. <i>Translational Psychiatry</i> , 2021, 11, 132.	2.4	0
579	Prevalence and Characterization of Biallelic and Monoallelic <i>NTHL1</i> and <i>MSH3</i> Variant Carriers From a Pan-Cancer Patient Population. <i>JCO Precision Oncology</i> , 2021, 5, 455-465.	1.5	10
581	Ethnic-specific association of amylase gene copy number with adiposity traits in a large Middle Eastern biobank. <i>Npj Genomic Medicine</i> , 2021, 6, 8.	1.7	8
582	14q32.11 microdeletion including <sc><i>CALM1</i></sc>, <sc><i>TTC7B</i></sc>, <sc><i>PSMC1</i></sc>, and <sc><i>RPS6KA5</i></sc>: A new potential cause of developmental and language delay in three unrelated patients. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1519-1524.	0.7	2
583	Neuroacanthocytosis Syndromes in an Italian Cohort: Clinical Spectrum, High Genetic Variability and Muscle Involvement. <i>Genes</i> , 2021, 12, 344.	1.0	6
585	A case of retinitis pigmentosa homozygous for a rare CNGA1 causal variant. <i>Scientific Reports</i> , 2021, 11, 4681.	1.6	6
586	More for less: predicting and maximizing genomic variant discovery via Bayesian nonparametrics. <i>Biometrika</i> , 2022, 109, 17-32.	1.3	5
588	Evaluating the molecular diagnostic yield of joint genotyping-based approach for detecting rare germline pathogenic and putative loss-of-function variants. <i>Genetics in Medicine</i> , 2021, 23, 918-926.	1.1	5
590	The mutational landscape of human olfactory G protein-coupled receptors. <i>BMC Biology</i> , 2021, 19, 21.	1.7	14
591	Human myelomeningocele risk and ultra-rare deleterious variants in genes associated with cilium, WNT-signaling, ECM, cytoskeleton and cell migration. <i>Scientific Reports</i> , 2021, 11, 3639.	1.6	8
592	Structure-Based Approaches to Classify the Functional Impact of ZBTB18 Missense Variants in Health and Disease. <i>ACS Chemical Neuroscience</i> , 2021, 12, 979-989.	1.7	4

#	ARTICLE	IF	CITATIONS
593	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. <i>Nature Communications</i> , 2021, 12, 833.	5.8	41
594	Current knowledge on genetic variants shaping placental transcriptome and their link to gestational and postnatal health. <i>Placenta</i> , 2021, 116, 2-11.	0.7	6
596	Bi-allelic loss of function variants in SLC30A5 as cause of perinatal lethal cardiomyopathy. <i>European Journal of Human Genetics</i> , 2021, 29, 808-815.	1.4	9
597	Oculocutaneous albinism type 1B associated with a functionally significant tyrosinase gene polymorphism detected with Whole Exome Sequencing. <i>Ophthalmic Genetics</i> , 2021, 42, 291-295.	0.5	3
598	Leveraging population-based exome screening to impact clinical care: The evolution of variant assessment in the Geisinger MyCode research project. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2021, 187, 83-94.	0.7	21
599	FARS1-related disorders caused by bi-allelic mutations in cytosolic phenylalanyl-tRNA synthetase genes: Look beyond the lungs!. <i>Clinical Genetics</i> , 2021, 99, 789-801.	1.0	16
600	Meta-analysis of tumor- and T cell-intrinsic mechanisms of sensitization to checkpoint inhibition. <i>Cell</i> , 2021, 184, 596-614.e14.	13.5	485
602	Massively Parallel Sequencing for Rare Genetic Disorders: Potential and Pitfalls. <i>Frontiers in Endocrinology</i> , 2020, 11, 628946.	1.5	15
603	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. <i>Nature Genetics</i> , 2021, 53, 294-303.	9.4	198
604	Identification of Germline Genetic Variants that Increase Prostate Cancer Risk and Influence Development of Aggressive Disease. <i>Cancers</i> , 2021, 13, 760.	1.7	22
605	Novel REST Truncation Mutations Causing Hereditary Gingival Fibromatosis. <i>Journal of Dental Research</i> , 2021, 100, 002203452199662.	2.5	8
606	Loss-of-Function Variants in EFEMP1 Cause a Recognizable Connective Tissue Disorder Characterized by Cutis Laxa and Multiple Herniations. <i>Genes</i> , 2021, 12, 510.	1.0	6
607	Heterogeneous Pulmonary Phenotypes in Filamin A Mutation-Related Lung Disease. <i>Pediatric, Allergy, Immunology, and Pulmonology</i> , 2021, 34, 7-14.	0.3	5
609	Genome-wide association study of psychiatric and substance use comorbidity in Mexican individuals. <i>Scientific Reports</i> , 2021, 11, 6771.	1.6	3
610	A Survey of Compound Heterozygous Variants in Pediatric Cancers and Structural Birth Defects. <i>Frontiers in Genetics</i> , 2021, 12, 640242.	1.1	9
611	Next-generation sequencing for inborn errors of immunity. <i>Human Immunology</i> , 2021, 82, 871-882.	1.2	12
613	Excision Repair Cross-Complementation Group 6 Gene Polymorphism Is Associated with the Response to FOLFIRINOX Chemotherapy in Asian Patients with Pancreatic Cancer. <i>Cancers</i> , 2021, 13, 1196.	1.7	0
614	Association of SLC32A1 Missense Variants With Genetic Epilepsy With Febrile Seizures Plus. <i>Neurology</i> , 2021, 96, e2251-e2260.	1.5	13

#	ARTICLE	IF	CITATIONS
615	<i>De novo</i> variants in neurodevelopmental disordersâ€™ experiences from a tertiary care center. <i>Clinical Genetics</i> , 2021, 100, 14-28.	1.0	64
616	Cobalamin J disease detected on newborn screening: Novel variant and normal neurodevelopmental course. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1870-1874.	0.7	0
617	NUDT15: A bench to bedside success story. <i>Clinical Biochemistry</i> , 2021, 92, 1-8.	0.8	7
618	Evolutionary Constraint on Visual and Nonvisual Mammalian Opsins. <i>Journal of Biological Rhythms</i> , 2021, 36, 109-126.	1.4	22
619	Trans-ethnic variation in germline variants of patients with renal cell carcinoma. <i>Cell Reports</i> , 2021, 34, 108926.	2.9	16
620	Transitioning the Molecular Tumor Board from Proof of Concept to Clinical Routine: A German Single-Center Analysis. <i>Cancers</i> , 2021, 13, 1151.	1.7	27
621	<i>TRIM33</i> gene somatic mutations identified by next generation sequencing in neoplasms of patients with anti-TIF1 β positive cancer-associated dermatomyositis. <i>Rheumatology</i> , 2021, 60, 5863-5867.	0.9	10
622	Somatic CAG expansion in Huntington's disease is dependent on the MLH3 endonuclease domain, which can be excluded via splice redirection. <i>Nucleic Acids Research</i> , 2021, 49, 3907-3918.	6.5	20
623	An international virtual hackathon to build tools for the analysis of structural variants within species ranging from coronaviruses to vertebrates. <i>F1000Research</i> , 2021, 10, 246.	0.8	3
624	<i>CACNA1</i> gain-of-function mutations differentially affect channel gating and cause neurodevelopmental disorders. <i>Brain</i> , 2021, 144, 2092-2106.	3.7	26
625	High mutation burden in the checkpoint and micro-RNA processing genes in myelodysplastic syndrome. <i>PLoS ONE</i> , 2021, 16, e0248430.	1.1	5
626	Genetic alterations in Wnt family of genes and their putative association with head and neck squamous cell carcinoma. <i>Genomics and Informatics</i> , 2021, 19, e5.	0.4	5
628	Functional Characterisation of Three Glycine N-Acyltransferase Variants and the Effect on Glycine Conjugation to Benzoylâ€‘CoA. <i>International Journal of Molecular Sciences</i> , 2021, 22, 3129.	1.8	4
629	PALLD mutation in a European family conveys a stromal predisposition for familial pancreatic cancer. <i>JCI Insight</i> , 2021, 6, .	2.3	7
633	Interpreting Sequence Variation in PDAC-Predisposing Genes Using a Multi-Tier Annotation Approach Performed at the Gene, Patient, and Cohort Level. <i>Frontiers in Oncology</i> , 2021, 11, 606820.	1.3	4
634	MutSpliceDB: A database of splice sites variants with RNA-seq based evidence on effects on splicing. <i>Human Mutation</i> , 2021, 42, 342-345.	1.1	9
635	<scp><i>GCH1</i></scp> mutations in hereditary spastic paraplegia. <i>Clinical Genetics</i> , 2021, 100, 51-58.	1.0	5
636	<i>ZNF91</i> deletion in human embryonic stem cells leads to ectopic activation of SVA retrotransposons and up-regulation of KRAB zinc finger gene clusters. <i>Genome Research</i> , 2021, 31, 551-563.	2.4	22

#	ARTICLE	IF	CITATIONS
637	Germline and Tumor Sequencing as a Diagnostic Tool To Resolve Suspected Lynch Syndrome. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 358-371.	1.2	12
638	Common Genetic Variation in Humans Impacts In Vitro Susceptibility to SARS-CoV-2 Infection. <i>Stem Cell Reports</i> , 2021, 16, 505-518.	2.3	39
639	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemann-Steiner syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1649-1665.	0.7	34
640	Voltage-Gated Ca ²⁺ -Channel β 1-Subunit de novo Missense Mutations: Gain or Loss of Function – Implications for Potential Therapies. <i>Frontiers in Synaptic Neuroscience</i> , 2021, 13, 634760.	1.3	32
641	Exploring a Region on Chromosome 8p23.1 Displaying Positive Selection Signals in Brazilian Admixed Populations: Additional Insights Into Predisposition to Obesity and Related Disorders. <i>Frontiers in Genetics</i> , 2021, 12, 636542.	1.1	4
642	Vitamin D Status, Bone Mineral Density, and VDR Gene Polymorphism in a Cohort of Belarusian Postmenopausal Women. <i>Nutrients</i> , 2021, 13, 837.	1.7	19
643	Novel bi-allelic variants expand the SPTBN4-related genetic and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2021, 29, 1121-1128.	1.4	9
644	Bi-allelic loss of function variants in COX20 gene cause autosomal recessive sensory neuropathy. <i>Brain</i> , 2021, 144, 2457-2470.	3.7	11
645	Known Mutations at the Cause of Alpha-1 Antitrypsin Deficiency an Updated Overview of SERPINA1 Variation Spectrum. <i>The Application of Clinical Genetics</i> , 2021, Volume 14, 173-194.	1.4	57
647	Prioritization of candidate genes for a South African family with Parkinson's disease using in-silico tools. <i>PLoS ONE</i> , 2021, 16, e0249324.	1.1	9
648	Diagnostic Analyses of Retinal Dystrophy Genes: Current Status and Perspective. <i>Klinische Monatsblätter Für Augenheilkunde</i> , 2021, 238, 261-266.	0.3	2
649	Hunting for Familial Parkinson's Disease Mutations in the Post Genome Era. <i>Genes</i> , 2021, 12, 430.	1.0	4
650	Disrupting upstream translation in mRNAs is associated with human disease. <i>Nature Communications</i> , 2021, 12, 1515.	5.8	37
651	A data set of variants derived from 1455 clinical and research exomes is efficient in variant prioritization for early-onset monogenic disorders in Indians. <i>Human Mutation</i> , 2021, 42, e15-e61.	1.1	25
652	Landmarks of human embryonic development inscribed in somatic mutations. <i>Science</i> , 2021, 371, 1249-1253.	6.0	65
655	Pathogenic alleles in microtubule, secretory granule and extracellular matrix-related genes in familial keratoconus. <i>Human Molecular Genetics</i> , 2021, 30, 658-671.	1.4	12
656	Myoclonic dystonia phenotype related to a novel calmodulin-binding transcription activator 1 sequence variant. <i>Neurogenetics</i> , 2021, 22, 137-141.	0.7	3
658	MTSplice predicts effects of genetic variants on tissue-specific splicing. <i>Genome Biology</i> , 2021, 22, 94.	3.8	23

#	ARTICLE	IF	CITATIONS
659	<sc><i>PLXNA2</i></sc> and <sc><i>LRRC40</i></sc> as candidate genes in autism spectrum disorder. <i>Autism Research</i> , 2021, 14, 1088-1100.	2.1	5
660	Sequencing at lymphoid neoplasm susceptibility loci maps six myeloma risk genes. <i>Human Molecular Genetics</i> , 2021, 30, 1142-1153.	1.4	2
661	Genetic Analysis of ZNF Protein Family Members for Early-Onset Parkinsonâ€™s Disease in Chinese Population. <i>Molecular Neurobiology</i> , 2021, 58, 3435-3442.	1.9	12
662	Genetic basis of mitochondrial diseases. <i>FEBS Letters</i> , 2021, 595, 1132-1158.	1.3	36
664	ATAV: a comprehensive platform for population-scale genomic analyses. <i>BMC Bioinformatics</i> , 2021, 22, 149.	1.2	20
665	Comprehensive identification of somatic nucleotide variants in human brain tissue. <i>Genome Biology</i> , 2021, 22, 92.	3.8	26
666	Identifying Primate ACE2 Variants That Confer Resistance to SARS-CoV-2. <i>Molecular Biology and Evolution</i> , 2021, 38, 2715-2731.	3.5	22
667	Intronic variant screening with targeted next-generation sequencing reveals first pseudoexon in LDLR in familial hypercholesterolemia. <i>Atherosclerosis</i> , 2021, 321, 14-20.	0.4	10
668	Variant curation expert panel recommendations for RYR1 pathogenicity classifications in malignant hyperthermia susceptibility. <i>Genetics in Medicine</i> , 2021, 23, 1288-1295.	1.1	46
669	The Role of Z-disc Proteins in Myopathy and Cardiomyopathy. <i>International Journal of Molecular Sciences</i> , 2021, 22, 3058.	1.8	29
670	A homozygous <i>POC1B</i> variant causes recessive cone-rod dystrophy. <i>Ophthalmic Genetics</i> , 2021, 42, 349-353.	0.5	5
671	Interactions of COMT and ALDH2 Genetic Polymorphisms on Symptoms of Parkinsonâ€™s Disease. <i>Brain Sciences</i> , 2021, 11, 361.	1.1	9
672	Whole-Genome Sequencing in Diagnostics of Selected Slovenian Undiagnosed Patients with Rare Disorders. <i>Life</i> , 2021, 11, 205.	1.1	5
673	Identification of a missense variant in SPDL1 associated with idiopathic pulmonary fibrosis. <i>Communications Biology</i> , 2021, 4, 392.	2.0	28
676	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 136.	1.2	5
677	The Ok blood group system: an update. <i>Immunohematology</i> , 2021, 37, 18-19.	0.2	1
678	Considerations for using population frequency data in germline variant interpretation: Cancer syndrome genes as a model. <i>Human Mutation</i> , 2021, 42, 530-536.	1.1	8
679	Autosomal Recessive Cerebellar Ataxia 1: First Case Report Depicting a Variant in SYNE1 Gene in a Chilean Patient. <i>Cerebellum</i> , 2021, 20, 938-941.	1.4	1

#	ARTICLE	IF	CITATIONS
680	An Assessment of GUCA1C Variants in Primary Congenital Glaucoma. <i>Genes</i> , 2021, 12, 359.	1.0	2
681	Novel BTK mutation in X-linked agammaglobulinemia: Report of a 17-year-old male. <i>Allergologia Et Immunopathologia</i> , 2021, 49, 80-83.	1.0	3
682	Widespread haploid-biased gene expression enables sperm-level natural selection. <i>Science</i> , 2021, 371, .	6.0	28
683	Expanding the Genotypic Spectrum of Congenital Sensory and Autonomic Neuropathies Using Whole-Exome Sequencing. <i>Neurology: Genetics</i> , 2021, 7, e568.	0.9	6
684	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. <i>Genetics in Medicine</i> , 2021, 23, 1028-1040.	1.1	34
685	A <i>Peruvian</i> patient carrying the novel <i>RHCE</i> * <i>cE</i> (<i>c.382G>>C</i>) missense allele in the <i>RH</i> blood group system. <i>Transfusion</i> , 2021, 61, E41-E43.		1
686	Novel single nucleotide deletion in <i>ART4</i> accounts for the Gy(a ⁺) phenotype in a woman of <i>Lebanese</i> origin. <i>Transfusion</i> , 2021, 61, E39-E40.	0.8	0
687	Common X-chromosome Variants Are Associated with Parkinson Disease Risk. <i>Annals of Neurology</i> , 2021, 90, 22-34.	2.8	28
688	Targeted sequencing panels in Italian ALS patients support different etiologies in the ALS/FTD continuum. <i>Journal of Neurology</i> , 2021, 268, 3766-3776.	1.8	12
689	A multidimensional computational exploration of congenital myasthenic syndrome causing mutations in human choline acetyltransferase. <i>Journal of Cellular Biochemistry</i> , 2021, 122, 787-800.	1.2	1
691	Novel somatic mutations in UBA1 as a cause of VEXAS syndrome. <i>Blood</i> , 2021, 137, 3676-3681.	0.6	115
692	Laminin β 2 variants associated with isolated nephropathy that impact matrix regulation. <i>JCI Insight</i> , 2021, 6, .	2.3	2
693	A novel homozygous RIPK4 variant in a family with severe Bartsocas-Papas syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1691-1699.	0.7	2
694	The Molecular Function of PURA and Its Implications in Neurological Diseases. <i>Frontiers in Genetics</i> , 2021, 12, 638217.	1.1	21
695	Evaluation of a Custom Design Gene Panel as a Diagnostic Tool for Human Non-Syndromic Infertility. <i>Genes</i> , 2021, 12, 410.	1.0	5
697	Molecular characterization of pathogenic <i>OTOA</i> gene conversions in hearing loss patients. <i>Human Mutation</i> , 2021, 42, 373-377.	1.1	10
698	HINT1 neuropathy in Norway: clinical, genetic and functional profiling. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 116.	1.2	8
699	Whole-genome sequencing of African Americans implicates differential genetic architecture in inflammatory bowel disease. <i>American Journal of Human Genetics</i> , 2021, 108, 431-445.	2.6	21

#	ARTICLE	IF	CITATIONS
700	Mutation analysis of seven SLC family transporters for early-onset Parkinson's disease in Chinese population. <i>Neurobiology of Aging</i> , 2021, 103, 152.e1-152.e6.	1.5	10
701	The Obesity Risk SNP (rs17782313) near the MC4R Gene Is Not Associated with Brain Glucose Uptake during Insulin Clamp—A Study in Finns. <i>Journal of Clinical Medicine</i> , 2021, 10, 1312.	1.0	1
702	Novel frameshift mutation in PURA gene causes severe encephalopathy of unclear cause. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1622.	0.6	5
703	A Novel Truncating Mutation in HOMER2 Causes Nonsyndromic Progressive DFNA68 Hearing Loss in a Spanish Family. <i>Genes</i> , 2021, 12, 411.	1.0	5
704	Mutational Profile and Clonal Evolution of Relapsed/Refractory Diffuse Large B-Cell Lymphoma. <i>Frontiers in Oncology</i> , 2021, 11, 628807.	1.3	13
706	Low Diversity of Human Variation Despite Mostly Mild Functional Impact of De Novo Variants. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 635382.	1.6	2
707	Genetic Variation in Enhancers Modifies Cardiomyopathy Gene Expression and Progression. <i>Circulation</i> , 2021, 143, 1302-1316.	1.6	36
708	A Novel Titin Truncation Variant Linked to Familial Dilated Cardiomyopathy Found in a Japanese Family and Its Functional Analysis in Genome-Edited Model Cells. <i>International Heart Journal</i> , 2021, 62, 359-366.	0.5	6
709	Biallelic loss-of-function variants in PLD1 cause congenital right-sided cardiac valve defects and neonatal cardiomyopathy. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	16
710	Genetic variation analyses indicate conserved SARS-CoV-2 host interaction and varied genetic adaptation in immune response factors in modern human evolution. <i>Development Growth and Differentiation</i> , 2021, 63, 219-227.	0.6	10
711	Aicardi-Goutières syndrome may present with positive newborn screen for X-linked adrenoleukodystrophy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1848-1853.	0.7	8
714	Large-scale Identification of Clonal Hematopoiesis and Mutations Recurrent in Blood Cancers. <i>Blood Cancer Discovery</i> , 2021, 2, 226-237.	2.6	22
716	Australia and New Zealand renal gene panel testing in routine clinical practice of 542 families. <i>Npj Genomic Medicine</i> , 2021, 6, 20.	1.7	11
717	The severity of the pathogen-induced acute sickness response is affected by polymorphisms in genes of the NLRP3 inflammasome pathway. <i>Brain, Behavior, and Immunity</i> , 2021, 93, 186-193.	2.0	5
718	Is it time to report carrier state for recessive disorders in every microarray analysis?—A pilot model based on hearing loss genes deletions. <i>European Journal of Human Genetics</i> , 2021, 29, 1292-1300.	1.4	1
719	Autism-linked Cullin3 germline haploinsufficiency impacts cytoskeletal dynamics and cortical neurogenesis through RhoA signaling. <i>Molecular Psychiatry</i> , 2021, 26, 3586-3613.	4.1	26
724	Why are rare variants hard to impute? Coalescent models reveal theoretical limits in existing algorithms. <i>Genetics</i> , 2021, 217, .	1.2	11
725	ActiveDriverDB: Interpreting Genetic Variation in Human and Cancer Genomes Using Post-translational Modification Sites and Signaling Networks (2021 Update). <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 626821.	1.8	12

#	ARTICLE	IF	CITATIONS
726	Emerging Role of ODC1 in Neurodevelopmental Disorders and Brain Development. <i>Genes</i> , 2021, 12, 470.	1.0	15
727	Molecular characterisation of rare loss-of-function NPAS3 and NPAS4 variants identified in individuals with neurodevelopmental disorders. <i>Scientific Reports</i> , 2021, 11, 6602.	1.6	6
728	Extended gene panel testing in lobular breast cancer. <i>Familial Cancer</i> , 2022, 21, 129-136.	0.9	1
729	The Impact of Modern Technologies on Molecular Diagnostic Success Rates, with a Focus on Inherited Retinal Dystrophy and Hearing Loss. <i>International Journal of Molecular Sciences</i> , 2021, 22, 2943.	1.8	6
730	PrecisionProDB: improving the proteomics performance for precision medicine. <i>Bioinformatics</i> , 2021, 37, 3361-3363.	1.8	4
731	Allele frequency of variants reported to cause adenine phosphoribosyltransferase deficiency. <i>European Journal of Human Genetics</i> , 2021, 29, 1061-1070.	1.4	5
732	Next-Generation Sequencing in the Field of Primary Immunodeficiencies: Current Yield, Challenges, and Future Perspectives. <i>Clinical Reviews in Allergy and Immunology</i> , 2021, 61, 212-225.	2.9	17
733	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , 2021, 13, 40.	3.6	116
735	New Findings of Immunodysregulation, Polyendocrinopathy, and Enteropathy X-linked Syndrome (IPEX); Granulomas in Lung and Duodenum. <i>Pediatric and Developmental Pathology</i> , 2021, 24, 252-257.	0.5	1
736	Novel variants in TUBA1A cause congenital fibrosis of the extraocular muscles with or without malformations of cortical brain development. <i>European Journal of Human Genetics</i> , 2021, 29, 816-826.	1.4	13
737	Ending a diagnostic odysseyâ€”The first case of Takenouchiâ€™Kosaki syndrome in an African patient. <i>Clinical Case Reports (discontinued)</i> , 2021, 9, 2144-2148.	0.2	4
738	Whole Genome Interpretation for a Family of Five. <i>Frontiers in Genetics</i> , 2021, 12, 535123.	1.1	3
739	A novel variant in <i>DMXL2</i> gene is associated with autosomal dominant non-syndromic hearing impairment (DFNA71) in a Cameroonian family. <i>Experimental Biology and Medicine</i> , 2021, 246, 1524-1532.	1.1	8
740	Gene variants of coagulation related proteins that interact with SARS-CoV-2. <i>PLoS Computational Biology</i> , 2021, 17, e1008805.	1.5	18
741	A rare case of an NLRP12-associated autoinflammatory disease. <i>European Journal of Medical Genetics</i> , 2021, 64, 104168.	0.7	5
743	Variable Expressivity and Allelic Heterogeneity in Type 2 Familial Partial Lipodystrophy: The p.(Thr528Met) LMNA Variant. <i>Journal of Clinical Medicine</i> , 2021, 10, 1497.	1.0	5
745	Investigation of the Wilson gene ATP7B transcriptional start site and the effect of core promoter alterations. <i>Scientific Reports</i> , 2021, 11, 7674.	1.6	2
746	A New Insight for the Identification of Oncogenic Variants in Breast and Prostate Cancers in Diverse Human Populations, With a Focus on Latinos. <i>Frontiers in Pharmacology</i> , 2021, 12, 630658.	1.6	3

#	ARTICLE	IF	CITATIONS
747	PKR activity modulation by phosphomimetic mutations of serine residues located three aminoacids upstream of double-stranded RNA binding motifs. <i>Scientific Reports</i> , 2021, 11, 9188.	1.6	9
748	Non-Penetrance for Ocular Phenotype in Two Individuals Carrying Heterozygous Loss-of-Function ZEB1 Alleles. <i>Genes</i> , 2021, 12, 677.	1.0	3
749	MR Locus: Identifying causal genes mediating a trait through Bayesian estimation of allelic heterogeneity. <i>PLoS Genetics</i> , 2021, 17, e1009455.	1.5	24
750	A novel age-informed approach for genetic association analysis in Alzheimer's disease. <i>Alzheimer's Research and Therapy</i> , 2021, 13, 72.	3.0	17
752	Genetic variability in COVID-19-related genes in the Brazilian population. <i>Human Genome Variation</i> , 2021, 8, 15.	0.4	29
753	Carrier frequency and incidence estimation of Smith-Lemli-Opitz syndrome in East Asian populations by Genome Aggregation Database (gnomAD) based analysis. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 166.	1.2	4
754	Whole genome and exome sequencing identify <i>NDUFV2</i> mutations as a new cause of progressive cavitating leukoencephalopathy. <i>Journal of Medical Genetics</i> , 2022, 59, 351-357.	1.5	5
755	A family harboring an MLKL loss of function variant implicates impaired necroptosis in diabetes. <i>Cell Death and Disease</i> , 2021, 12, 345.	2.7	26
756	A Novel Mutation in the Thyroglobulin Gene Resulting in Neonatal Goiter and Congenital Hypothyroidism in an Eritrean Infant. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , 2022, 14, 221-226.	0.4	6
757	Advancing drug discovery using the power of the human genome. <i>Journal of Pathology</i> , 2021, 254, 418-429.	2.1	11
758	A homozygous truncating variant in GDF9 in siblings with primary ovarian insufficiency. <i>Journal of Assisted Reproduction and Genetics</i> , 2021, 38, 1539-1543.	1.2	3
759	A Review of the Literature Organized Into a New Database: RHeference. <i>Transfusion Medicine Reviews</i> , 2021, 35, 70-77.	0.9	8
760	Janus-faced EPHB4-associated disorders: novel pathogenic variants and unreported intrafamilial overlapping phenotypes. <i>Genetics in Medicine</i> , 2021, 23, 1315-1324.	1.1	6
761	Wolfram-like syndrome with bicuspid aortic valve due to a homozygous missense variant in CDK13. <i>Journal of Human Genetics</i> , 2021, 66, 1009-1018.	1.1	4
762	Proteogenomic and metabolomic characterization of human glioblastoma. <i>Cancer Cell</i> , 2021, 39, 509-528.e20.	7.7	327
763	Spectrum of splicing variants in disease genes and the ability of RNA analysis to reduce uncertainty in clinical interpretation. <i>American Journal of Human Genetics</i> , 2021, 108, 696-708.	2.6	43
764	Somatic mutation landscapes at single-molecule resolution. <i>Nature</i> , 2021, 593, 405-410.	13.7	254
765	A Rare Autosomal Dominant Variant in Regulator of Calcineurin Type 1 (RCAN1) Gene Confers Enhanced Calcineurin Activity and May Cause FSGS. <i>Journal of the American Society of Nephrology: JASN</i> , 2021, 32, 1682-1695.	3.0	3

#	ARTICLE	IF	CITATIONS
766	ATP1A3-Related Disorders: An Ever-Expanding Clinical Spectrum. <i>Frontiers in Neurology</i> , 2021, 12, 637890.	1.1	28
769	<i>BBS1</i> branchpoint variant is associated with non-syndromic retinitis pigmentosa. <i>Journal of Medical Genetics</i> , 2022, 59, 438-444.	1.5	13
771	Systemic Screening for 22q11.2 Copy Number Variations in Hungarian Pediatric and Adult Patients With Congenital Heart Diseases Identified Rare Pathogenic Patterns in the Region. <i>Frontiers in Genetics</i> , 2021, 12, 635480.	1.1	0
772	Functional testing for variant prioritization in a family with long QT syndrome. <i>Molecular Genetics and Genomics</i> , 2021, 296, 823-836.	1.0	3
773	Safety, Antitumor Activity, and Biomarker Analysis in a Phase I Trial of the Once-daily Wee1 Inhibitor Adavosertib (AZD1775) in Patients with Advanced Solid Tumors. <i>Clinical Cancer Research</i> , 2021, 27, 3834-3844.	3.2	36
774	Penile Cancer-Derived Cells Molecularly Characterized as Models to Guide Targeted Therapies. <i>Cells</i> , 2021, 10, 814.	1.8	9
776	In Silico Analysis of the Molecular-Level Impact of SMPD1 Variants on Niemann-Pick Disease Severity. <i>International Journal of Molecular Sciences</i> , 2021, 22, 4516.	1.8	4
777	Molecular Insights into Myocilin and Its Glaucoma-Causing Misfolded Olfactomedin Domain Variants. <i>Accounts of Chemical Research</i> , 2021, 54, 2205-2215.	7.6	13
779	Incidental detection of acquired variants in germline genetic and genomic testing: a points to consider statement of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2021, 23, 1179-1184.	1.1	13
780	<i>FGFR2</i> Extracellular Domain In-Frame Deletions Are Therapeutically Targetable Genomic Alterations That Function as Oncogenic Drivers in Cholangiocarcinoma. <i>Cancer Discovery</i> , 2021, 11, 2488-2505.	7.7	46
781	Massive parallel sequencing in a family with rectal cancer. <i>Hereditary Cancer in Clinical Practice</i> , 2021, 19, 23.	0.6	3
783	Phenotype and genotype characteristics of 58 patients showing a prolonged effect of succinylcholine: A four-year experience. <i>Anaesthesia, Critical Care & Pain Medicine</i> , 2021, 40, 100847.	0.6	0
784	The Role of Knockout Olfactory Receptor Genes in Odor Discrimination. <i>Genes</i> , 2021, 12, 631.	1.0	3
787	Biallelic variants in the <i>SORD</i> gene are one of the most common causes of hereditary neuropathy among Czech patients. <i>Scientific Reports</i> , 2021, 11, 8443.	1.6	14
789	Multiplexed assays reveal effects of missense variants in <i>MSH2</i> and cancer predisposition. <i>PLoS Genetics</i> , 2021, 17, e1009496.	1.5	13
791	Low-level variant calling for non-matched samples using a position-based and nucleotide-specific approach. <i>BMC Bioinformatics</i> , 2021, 22, 181.	1.2	0
793	Multi-system neurological disorder associated with a <i>CRYAB</i> variant. <i>Neurogenetics</i> , 2021, 22, 117-125.	0.7	1
795	Whole-exome sequencing identifies genes associated with Tourette's disorder in multiplex families. <i>Molecular Psychiatry</i> , 2021, , .	4.1	16

#	ARTICLE	IF	CITATIONS
796	Human ACE2 receptor polymorphisms and altered susceptibility to SARS-CoV-2. <i>Communications Biology</i> , 2021, 4, 475.	2.0	126
798	Progressive myoclonus epilepsies—Residual unsolved cases have marked genetic heterogeneity including dolichol-dependent protein glycosylation pathway genes. <i>American Journal of Human Genetics</i> , 2021, 108, 722-738.	2.6	41
799	Genome-Wide Association Study of Non-syndromic Orofacial Clefts in a Multiethnic Sample of Families and Controls Identifies Novel Regions. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 621482.	1.8	16
800	Burden of rare variants in synaptic genes in patients with severe tinnitus: An exome based extreme phenotype study. <i>EBioMedicine</i> , 2021, 66, 103309.	2.7	25
801	Personalized Therapy for Mycophenolate: Consensus Report by the International Association of Therapeutic Drug Monitoring and Clinical Toxicology. <i>Therapeutic Drug Monitoring</i> , 2021, 43, 150-200.	1.0	89
802	Heterozygous variants in SPTBN1 cause intellectual disability and autism. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2037-2045.	0.7	9
803	Delineation of epileptic and neurodevelopmental phenotypes associated with variants in STX1B. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 87, 25-29.	0.9	6
804	Germline sequence analysis of RABL3 in a large series of pancreatic ductal adenocarcinoma patients reveals no evidence of deleterious variants. <i>Genes Chromosomes and Cancer</i> , 2021, 60, 559-564.	1.5	3
805	A rare missense variant in the <i>ATP2C2</i> gene is associated with language impairment and related measures. <i>Human Molecular Genetics</i> , 2021, 30, 1160-1171.	1.4	10
806	Assessing acquired resistance to IDH1 inhibitor therapy by full-exon <i>IDH1</i> sequencing and structural modeling. <i>Journal of Physical Education and Sports Management</i> , 2021, 7, a006007.	0.5	10
807	Notch3 Signaling and Aggregation as Targets for the Treatment of CADASIL and Other NOTCH3-Associated Small-Vessel Diseases. <i>American Journal of Pathology</i> , 2021, 191, 1856-1870.	1.9	17
808	Haploinsufficiency of PRR12 causes a spectrum of neurodevelopmental, eye, and multisystem abnormalities. <i>Genetics in Medicine</i> , 2021, 23, 1234-1245.	1.1	6
811	Significant variants of type 2 diabetes in the Arabian Region through an Integration of exome databases. <i>PLoS ONE</i> , 2021, 16, e0249226.	1.1	0
812	From bedside to bench: regulation of host factors in SARS-CoV-2 infection. <i>Experimental and Molecular Medicine</i> , 2021, 53, 483-494.	3.2	6
814	A novel proline substitution (Arg201Pro) in alpha helix 8 of TMEM98 causes autosomal dominant nanophthalmos-4, closed angle glaucoma and attenuated visual acuity. <i>Experimental Eye Research</i> , 2021, 205, 108497.	1.2	3
815	Digenic Inheritance and Gene-Environment Interaction in a Patient With Hypertriglyceridemia and Acute Pancreatitis. <i>Frontiers in Genetics</i> , 2021, 12, 640859.	1.1	7
817	Multiregional Sequencing of IDH-WT Glioblastoma Reveals High Genetic Heterogeneity and a Dynamic Evolutionary History. <i>Cancers</i> , 2021, 13, 2044.	1.7	5
818	Analysis of Brugada syndrome loci reveals that fine-mapping clustered GWAS hits enhances the annotation of disease-relevant variants. <i>Cell Reports Medicine</i> , 2021, 2, 100250.	3.3	4

#	ARTICLE	IF	CITATIONS
819	More than meets the eye: Expanding and reviewing the clinical and mutational spectrum of brittle cornea syndrome. <i>Human Mutation</i> , 2021, 42, 711-730.	1.1	19
820	MutationTaster2021. <i>Nucleic Acids Research</i> , 2021, 49, W446-W451.	6.5	122
824	Biallelic variants in TMEM222 cause a new autosomal recessive neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2021, 23, 1246-1254.	1.1	5
825	Application of Genome Sequencing from Blood to Diagnose Mitochondrial Diseases. <i>Genes</i> , 2021, 12, 607.	1.0	8
826	CDH1 pathogenic variants and cancer risk in an unselected patient population. <i>Familial Cancer</i> , 2022, 21, 235-239.	0.9	5
828	DPP9: Comprehensive In Silico Analyses of Loss of Function Gene Variants and Associated Gene Expression Signatures in Human Hepatocellular Carcinoma. <i>Cancers</i> , 2021, 13, 1637.	1.7	9
830	Rare, Protein-Altering Variants in <i>AS3MT</i> and Arsenic Metabolism Efficiency: A Multi-Population Association Study. <i>Environmental Health Perspectives</i> , 2021, 129, 47007.	2.8	9
831	Whole exome sequencing, a hypothesis-free approach to investigate recurrent early miscarriage. <i>Reproductive BioMedicine Online</i> , 2021, 42, 789-798.	1.1	3
832	Genomic Evolution in a Patient With Lung Adenocarcinoma With a Germline EGFR T790M Mutation. <i>JTO Clinical and Research Reports</i> , 2021, 2, 100146.	0.6	0
834	Whole-exome sequencing with targeted analysis and epilepsy after acute symptomatic neonatal seizures. <i>Pediatric Research</i> , 2021, , .	1.1	6
836	Deletion of CTCF sites in the SHH locus alters enhancer-promoter interactions and leads to acheiropodia. <i>Nature Communications</i> , 2021, 12, 2282.	5.8	37
837	Genetic analyses identify widespread sex-differential participation bias. <i>Nature Genetics</i> , 2021, 53, 663-671.	9.4	124
838	Possible ethnic associations in primary hyperoxaluria type-III-associated HOGA1 sequence variants. <i>Molecular Biology Reports</i> , 2021, 48, 3841-3844.	1.0	5
839	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. <i>Genome Medicine</i> , 2021, 13, 55.	3.6	16
841	Genotype-phenotype correlations of <i>KIF5A</i> stalk domain variants. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2021, 22, 561-570.	1.1	9
842	<i>SASH3</i> variants cause a novel form of X-linked combined immunodeficiency with immune dysregulation. <i>Blood</i> , 2021, 138, 1019-1033.	0.6	28
844	Low-coverage sequencing cost-effectively detects known and novel variation in underrepresented populations. <i>American Journal of Human Genetics</i> , 2021, 108, 656-668.	2.6	49
848	Age-related differences of genetic susceptibility to patients with acute lymphoblastic leukemia. <i>Aging</i> , 2021, 13, 12456-12465.	1.4	2

#	ARTICLE	IF	CITATIONS
850	Neurological Disorders Associated with WWOX Germline Mutations—A Comprehensive Overview. <i>Cells</i> , 2021, 10, 824.	1.8	15
851	Hypersociability associated with developmental delay, macrocephaly and facial dysmorphism points to CHD3 mutations. <i>European Journal of Medical Genetics</i> , 2021, 64, 104166.	0.7	6
852	Evolution of DNA methylation in the human brain. <i>Nature Communications</i> , 2021, 12, 2021.	5.8	53
853	A homozygous variant in <i>TBPL2</i> was identified in women with oocyte maturation defects and infertility. <i>Human Reproduction</i> , 2021, 36, 2011-2019.	0.4	14
854	Progress in Defining the Genetic Contribution to Type 2 Diabetes in Individuals of East Asian Ancestry. <i>Current Diabetes Reports</i> , 2021, 21, 17.	1.7	5
855	Titin Circular RNAs Create a Back-Splice Motif Essential for SRSF10 Splicing. <i>Circulation</i> , 2021, 143, 1502-1512.	1.6	18
856	Profiling SARS-CoV-2 mutation fingerprints that range from the viral pangenome to individual infection quasispecies. <i>Genome Medicine</i> , 2021, 13, 62.	3.6	18
857	Variants in <i>PRKAR1B</i> cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. <i>Genetics in Medicine</i> , 2021, 23, 1465-1473.	1.1	10
858	Rare deleterious <i>de novo</i> missense variants in <i>Rnf2/Ring2</i> are associated with a neurodevelopmental disorder with unique clinical features. <i>Human Molecular Genetics</i> , 2021, 30, 1283-1292.	1.4	17
859	Whole-genome sequencing reveals new Alzheimer's disease-associated rare variants in loci related to synaptic function and neuronal development. <i>Alzheimer's and Dementia</i> , 2021, 17, 1509-1527.	0.4	50
861	New insights into hallux valgus by whole exome sequencing study. <i>Experimental Biology and Medicine</i> , 2021, 246, 1607-1616.	1.1	2
863	The impact of cell type and context-dependent regulatory variants on human immune traits. <i>Genome Biology</i> , 2021, 22, 122.	3.8	32
864	Opportunities and challenges for the computational interpretation of rare variation in clinically important genes. <i>American Journal of Human Genetics</i> , 2021, 108, 535-548.	2.6	40
865	The landscape of autosomal-recessive pathogenic variants in European populations reveals phenotype-specific effects. <i>American Journal of Human Genetics</i> , 2021, 108, 608-619.	2.6	36
866	Whole genome variation in 27 Mexican indigenous populations, demographic and biomedical insights. <i>PLoS ONE</i> , 2021, 16, e0249773.	1.1	8
868	Integrated Genomic Analysis Identifies Driver Genes and Cisplatin-Resistant Progenitor Phenotype in Pediatric Liver Cancer. <i>Cancer Discovery</i> , 2021, 11, 2524-2543.	7.7	41
870	The PI3K/mTOR Pathway Is Targeted by Rare Germline Variants in Patients with Both Melanoma and Renal Cell Carcinoma. <i>Cancers</i> , 2021, 13, 2243.	1.7	2
872	Secondary findings in 622 Turkish clinical exome sequencing data. <i>Journal of Human Genetics</i> , 2021, 66, 1113-1119.	1.1	4

#	ARTICLE	IF	CITATIONS
873	Angiotensin System Polymorphisms™ in SARS-CoV-2 Positive Patients: Assessment Between Symptomatic and Asymptomatic Patients: A Pilot Study. <i>Pharmacogenomics and Personalized Medicine</i> , 2021, Volume 14, 621-629.	0.4	37
874	Screening for extremely rare pathogenic variants of monogenic diabetes using targeted panel sequencing. <i>Endocrine</i> , 2021, 73, 752-757.	1.1	3
875	Precision Medicine in Catecholaminergic Polymorphic Ventricular Tachycardia. <i>Journal of the American College of Cardiology</i> , 2021, 77, 2592-2612.	1.2	26
876	Paired-like homeobox gene (PHOX2B) nonpolyalanine repeat expansion mutations (NPARMs): genotype-phenotype correlation in congenital central hypoventilation syndrome (CCHS). <i>Genetics in Medicine</i> , 2021, 23, 1656-1663.	1.1	16
877	Skeletal and molecular findings in 51 Cleidocranial dysplasia patients from Turkey. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2488-2495.	0.7	8
880	Pan-genomic characterization of high-risk pediatric papillary thyroid carcinoma. <i>Endocrine-Related Cancer</i> , 2021, 28, 337-351.	1.6	11
881	DNACJ12 deficiency in patients with unexplained hyperphenylalaninemia: two new patients and a novel variant. <i>Metabolic Brain Disease</i> , 2021, 36, 1405-1410.	1.4	8
882	Activity of PD-1 blockade with nivolumab among patients with recurrent atypical/anaplastic meningioma: phase II trial results. <i>Neuro-Oncology</i> , 2022, 24, 101-113.	0.6	38
884	Functional dynamic genetic effects on gene regulation are specific to particular cell types and environmental conditions. <i>ELife</i> , 2021, 10, .	2.8	41
885	Rare Missense Functional Variants at <i>COL4A1</i> and <i>COL4A2</i> in Sporadic Intracerebral Hemorrhage. <i>Neurology</i> , 2021, 97, .	1.5	6
886	Comprehensive phenotypic and functional analysis of dominant and recessive <i>FOXE3</i> alleles in ocular developmental disorders. <i>Human Molecular Genetics</i> , 2021, 30, 1591-1606.	1.4	6
887	Incomplete penetrance of a novel <i>SDHD</i> variation causing familial head and neck paraganglioma. <i>Clinical Otolaryngology</i> , 2021, 46, 1044-1049.	0.6	4
888	Dissecting <i>ELANE</i> neutropenia pathogenicity by human HSC gene editing. <i>Cell Stem Cell</i> , 2021, 28, 833-845.e5.	5.2	23
889	Missense and truncating variants in <i>CHD5</i> in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. <i>Human Genetics</i> , 2021, 140, 1109-1120.	1.8	18
890	Comprehensive analysis of <i>ADA2</i> genetic variants and estimation of carrier frequency driven by a function-based approach. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 149, 379-387.	1.5	27
891	Clinical, neuroimaging, and molecular spectrum of <i>TECPR2</i> -associated hereditary sensory and autonomic neuropathy with intellectual disability. <i>Human Mutation</i> , 2021, 42, 762-776.	1.1	18
892	Endometrial Carcinoma as the Presenting Malignancy in a Teenager With a Pathogenic <i>TP53</i> Germline Mutation: A Case Report and Literature Review. <i>International Journal of Gynecological Pathology</i> , 2022, 41, 258-267.	0.9	3
893	The Cause of Hereditary Hearing Loss in <i>GJB2</i> Heterozygotes - A Comprehensive Study of the <i>GJB2/DFNB1</i> Region. <i>Genes</i> , 2021, 12, 684.	1.0	8

#	ARTICLE	IF	CITATIONS
894	SMA Identified: Clinical and Molecular Findings From a Sponsored Testing Program for Spinal Muscular Atrophy in More Than 2,000 Individuals. <i>Frontiers in Neurology</i> , 2021, 12, 663911.	1.1	7
896	Genoppi is an open-source software for robust and standardized integration of proteomic and genetic data. <i>Nature Communications</i> , 2021, 12, 2580.	5.8	15
898	Multigene Panel Testing in Individuals With Hepatocellular Carcinoma Identifies Pathogenic Germline Variants. <i>JCO Precision Oncology</i> , 2021, 5, 988-1000.	1.5	10
899	A novel de novo intronic variant in ITPR1 causes Gillespie syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2315-2324.	0.7	2
900	A de novo missense variant in <i>MED13</i> in a patient with global developmental delay, marked facial dysmorphism, macroglossia, short stature, and macrocephaly. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2586-2592.	0.7	4
901	Factors Affecting Migration to GRCh38 in Laboratories Performing Clinical Next-Generation Sequencing. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 651-657.	1.2	13
902	Strategies in Rapid Genetic Diagnostics of Critically Ill Children: Experiences From a Dutch University Hospital. <i>Frontiers in Pediatrics</i> , 2021, 9, 600556.	0.9	6
905	The causes of Fanconi anemia in South Asia and the Middle East: A case series and review of the literature. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1693.	0.6	2
906	aScan: A Novel Method for the Study of Allele Specific Expression in Single Individuals. <i>Journal of Molecular Biology</i> , 2021, 433, 166829.	2.0	1
907	Molecular mechanisms of metabotropic GABA _B receptor function. <i>Science Advances</i> , 2021, 7, .	4.7	46
908	Familial Occurrence of Adult Granulosa Cell Tumors: Analysis of Whole-Genome Germline Variants. <i>Cancers</i> , 2021, 13, 2430.	1.7	1
909	Biallelic <i>ASCC1</i> variants including a novel intronic variant result in expanded phenotypic spectrum of spinal muscular atrophy with congenital bone fractures 2 (<i>SMABF2</i>). <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2190-2197.	0.7	4
910	Dissection of contiguous gene effects for deletions around ERF on chromosome 19. <i>Human Mutation</i> , 2021, 42, 811-817.	1.1	2
911	Swarm: A federated cloud framework for large-scale variant analysis. <i>PLoS Computational Biology</i> , 2021, 17, e1008977.	1.5	2
914	Molecular genetics of renal ciliopathies. <i>Biochemical Society Transactions</i> , 2021, 49, 1205-1220.	1.6	15
915	Decreased ATM Function Causes Delayed DNA Repair and Apoptosis in Common Variable Immunodeficiency Disorders. <i>Journal of Clinical Immunology</i> , 2021, 41, 1315-1330.	2.0	6
916	Phenotype-driven variant filtration strategy in exome sequencing toward a high diagnostic yield and identification of 85 novel variants in 400 patients with rare Mendelian disorders. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2561-2571.	0.7	24
917	KCNV2-Associated Retinopathy: Genetics, Electrophysiology, and Clinical Course—KCNV2 Study Group Report 1. <i>American Journal of Ophthalmology</i> , 2021, 225, 95-107.	1.7	17

#	ARTICLE	IF	CITATIONS
918	Immunodeficiency and bone marrow failure with mosaic and germline TLR8 gain of function. <i>Blood</i> , 2021, 137, 2450-2462.	0.6	47
919	Whole-exome sequencing identifies functional classes of gene mutations associated with bone marrow failure in pediatric Fanconi Anemia patients. <i>European Journal of Haematology</i> , 2021, 107, 293-294.	1.1	0
920	Low frequency variants associated with leukocyte telomere length in the Singapore Chinese population. <i>Communications Biology</i> , 2021, 4, 519.	2.0	15
921	L2HGDH Missense Variant in a Cat with L-2-Hydroxyglutaric Aciduria. <i>Genes</i> , 2021, 12, 682.	1.0	1
923	Feasibility of predicting allele specific expression from DNA sequencing using machine learning. <i>Scientific Reports</i> , 2021, 11, 10606.	1.6	4
924	Identification of missense <i>MAB21L1</i> variants in microphthalmia and aniridia. <i>Human Mutation</i> , 2021, 42, 877-890.	1.1	13
925	New cases that expand the genotypic and phenotypic spectrum of Congenital NAD Deficiency Disorder. <i>Human Mutation</i> , 2021, 42, 862-876.	1.1	16
926	Bi-allelic <i>VPS16</i> variants limit HOPS/CORVET levels and cause a mucopolysaccharidosis-like disease. <i>EMBO Molecular Medicine</i> , 2021, 13, e13376.	3.3	16
927	The DBSAV Database: Predicting Deleteriousness of Single Amino Acid Variations in the Human Proteome. <i>Journal of Molecular Biology</i> , 2021, 433, 166915.	2.0	15
928	A form of muscular dystrophy associated with pathogenic variants in <i>JAG2</i> . <i>American Journal of Human Genetics</i> , 2021, 108, 840-856.	2.6	15
930	CNVxplorer: a web tool to assist clinical interpretation of CNVs in rare disease patients. <i>Nucleic Acids Research</i> , 2021, 49, W93-W103.	6.5	14
932	Identification of a Splice Variant (c.5074+3A>C) of <i>BRCA1</i> by RNA Sequencing and TOPO Cloning. <i>Genes</i> , 2021, 12, 810.	1.0	0
934	Dysgenesis and Dysfunction of the Pancreas and Pituitary Due to <i>FOXA2</i> Gene Defects. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e4142-e4154.	1.8	6
935	Novel Gene Mutations Regulating Immune Responses in Autoimmune Polyglandular Syndrome With an Atypical Course. <i>Journal of the Endocrine Society</i> , 2021, 5, bvab077.	0.1	2
936	Expectations and blind spots for structural variation detection from long-read assemblies and short-read genome sequencing technologies. <i>American Journal of Human Genetics</i> , 2021, 108, 919-928.	2.6	72
937	Short-coupled ventricular fibrillation represents a distinct phenotype among latent causes of unexplained cardiac arrest: a report from the CASPER registry. <i>European Heart Journal</i> , 2021, 42, 2827-2838.	1.0	54
938	The analysis of ancestry with small-scale forensic panels of genetic markers. <i>Emerging Topics in Life Sciences</i> , 2021, 5, 443-453.	1.1	4
939	Novel ultra-rare exonic variants identified in a founder population implicate cadherins in schizophrenia. <i>Neuron</i> , 2021, 109, 1465-1478.e4.	3.8	21

#	ARTICLE	IF	CITATIONS
940	Functional and genetic determinants of mutation rate variability in regulatory elements of cancer genomes. <i>Genome Biology</i> , 2021, 22, 133.	3.8	12
941	Computational Studies of the Structural Basis of Human RPS19 Mutations Associated With Diamond-Blackfan Anemia. <i>Frontiers in Genetics</i> , 2021, 12, 650897.	1.1	3
942	Next-Generation Sequencing for Congenital Nephrotic Syndrome: A Multi-Center Cross-Sectional Study from India. <i>Indian Pediatrics</i> , 2021, 58, 445-451.	0.2	8
943	Accelerating Medicines Partnership: Parkinson's Disease. <i>Genetic Resource. Movement Disorders</i> , 2021, 36, 1795-1804.	2.2	60
944	The Polygenic and Monogenic Basis of Paediatric Fractures. <i>Current Osteoporosis Reports</i> , 2021, 19, 481-493.	1.5	2
947	Biallelic variants in <i>VPS50</i> cause a neurodevelopmental disorder with neonatal cholestasis. <i>Brain</i> , 2021, 144, 3036-3049.	3.7	4
948	Neurodevelopmental phenotypes associated with pathogenic variants in <i>SLC6A1</i> . <i>Journal of Medical Genetics</i> , 2022, 59, 536-543.	1.5	18
950	Clinical Manifestations, Mutational Analysis, and Immunological Phenotype in Patients with RAG1/2 Mutations: First Cases Series from Mexico and Description of Two Novel Mutations. <i>Journal of Clinical Immunology</i> , 2021, 41, 1291-1302.	2.0	2
951	Genetics and genomics of arrhythmic risk: current and future strategies to prevent sudden cardiac death. <i>Nature Reviews Cardiology</i> , 2021, 18, 774-784.	6.1	15
954	Association of FAAH p.Pro129Thr and COMT p.Ala72Ser with schizophrenia and comorbid substance use through next-generation sequencing: an exploratory analysis. <i>Revista Brasileira De Psiquiatria</i> , 2021, , .	0.9	2
955	Genetic analysis of GLT8D1 and ARPP21 in Australian familial and sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2021, 101, 297.e9-297.e11.	1.5	6
956	Different types of disease-causing noncoding variants revealed by genomic and gene expression analyses in families with X-linked intellectual disability. <i>Human Mutation</i> , 2021, 42, 835-847.	1.1	0
957	Whole-Exome Sequencing Reveals a Rare Variant of OTOF Gene Causing Congenital Non-syndromic Hearing Loss Among Large Muslim Families Favoring Consanguinity. <i>Frontiers in Genetics</i> , 2021, 12, 641925.	1.1	5
958	The Novel KIF1A Missense Variant (R169T) Strongly Reduces Microtubule Stimulated ATPase Activity and Is Associated With NESCAV Syndrome. <i>Frontiers in Neuroscience</i> , 2021, 15, 618098.	1.4	11
959	Familial pseudohyperkalemia induces significantly higher levels of extracellular potassium in early storage of red cell concentrates without affecting other standard measures of quality: A case control and allele frequency study. <i>Transfusion</i> , 2021, 61, 2439-2449.	0.8	9
960	Rho-GTPase Activating Protein myosin MYO9A identified as a novel candidate gene for monogenic focal segmental glomerulosclerosis. <i>Kidney International</i> , 2021, 99, 1102-1117.	2.6	12
962	Genotype-phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 90.	3.6	16
963	Effect of MACC1 Genetic Polymorphisms and Environmental Risk Factors in the Occurrence of Oral Squamous Cell Carcinoma. <i>Journal of Personalized Medicine</i> , 2021, 11, 490.	1.1	5

#	ARTICLE	IF	CITATIONS
964	A Challenging Diagnosis: PTEN Hamartoma Tumor Syndrome Presenting as Isolated Soft-tissue Vascular Anomalies. <i>Journal of Vascular Anomalies</i> , 2021, 2, e011.	0.1	0
965	Alternative splicing during mammalian organ development. <i>Nature Genetics</i> , 2021, 53, 925-934.	9.4	93
966	“H-syndrome”: a multisystem genetic disorder with cutaneous clues. <i>BMJ Case Reports</i> , 2021, 14, e238973.	0.2	3
968	Whole genome sequencing reveals a frameshift mutation and a large deletion in YY1AP1 in a girl with a panvascular artery disease. <i>Human Genomics</i> , 2021, 15, 28.	1.4	5
969	Discordant Reporting of a Previously Undescribed Pathogenic Germline BRCA2 Variant in Blood and Tumor Tissue in a Patient With Pancreatic Adenocarcinoma. <i>JCO Precision Oncology</i> , 2021, 5, 974-980.	1.5	1
970	Knowledge bases and software support for variant interpretation in precision oncology. <i>Briefings in Bioinformatics</i> , 2021, 22, .	3.2	9
971	Whole-genome sequencing. <i>Practical Neurology</i> , 2021, 21, 322-327.	0.5	3
972	A novel variant in the COX15 gene causing a fatal infantile cardioencephalomyopathy: A case report with clinical and molecular review. <i>European Journal of Medical Genetics</i> , 2021, 64, 104195.	0.7	1
973	Biallelic mutations in <i>RNF220</i> cause laminopathies featuring leukodystrophy, ataxia and deafness. <i>Brain</i> , 2021, 144, 3020-3035.	3.7	11
974	Molecular genetics of inherited retinal degenerations in Icelandic patients. <i>Clinical Genetics</i> , 2021, 100, 156-167.	1.0	4
975	Loss of function variants in <i>Kv11.1</i> cardiac channels as a biomarker for SUDEP. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 1422-1432.	1.7	9
976	The pluripotent stem cell-specific transcript <i>ESRG</i> is dispensable for human pluripotency. <i>PLoS Genetics</i> , 2021, 17, e1009587.	1.5	20
977	Refinement of the clinical variant interpretation framework by statistical evidence and machine learning. <i>Med</i> , 2021, 2, 611-632.e9.	2.2	1
979	Subcutaneous vitamin B12 administration using a portable infusion pump in cobalamin-related remethylation disorders: a gentle and easy to use alternative to intramuscular injections. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 215.	1.2	1
980	TRIM71 Deficiency Causes Germ Cell Loss During Mouse Embryogenesis and Is Associated With Human Male Infertility. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 658966.	1.8	17
981	High Concordance of Genomic Profiles between Primary and Metastatic Colorectal Cancer. <i>International Journal of Molecular Sciences</i> , 2021, 22, 5561.	1.8	4
982	Missense variants in <i>DPYSL5</i> cause a neurodevelopmental disorder with corpus callosum agenesis and cerebellar abnormalities. <i>American Journal of Human Genetics</i> , 2021, 108, 951-961.	2.6	26
983	<i>MTR3D</i> : identifying regions within protein tertiary structures under purifying selection. <i>Nucleic Acids Research</i> , 2021, 49, W438-W445.	6.5	17

#	ARTICLE	IF	CITATIONS
984	Analysis and Interpretation of the Impact of Missense Variants in Cancer. <i>International Journal of Molecular Sciences</i> , 2021, 22, 5416.	1.8	28
985	The clinical and functional effects of <i>TERT</i> variants in myelodysplastic syndrome. <i>Blood</i> , 2021, 138, 898-911.	0.6	27
986	One in seven pathogenic variants can be challenging to detect by NGS: an analysis of 450,000 patients with implications for clinical sensitivity and genetic test implementation. <i>Genetics in Medicine</i> , 2021, 23, 1673-1680.	1.1	40
987	The Complex and Diverse Genetic Architecture of Dilated Cardiomyopathy. <i>Circulation Research</i> , 2021, 128, 1514-1532.	2.0	49
989	Population-scale tissue transcriptomics maps long non-coding RNAs to complex disease. <i>Cell</i> , 2021, 184, 2633-2648.e19.	13.5	94
990	Identification of a variant in NDP associated with X-linked retinal dysplasia in the English cocker spaniel dog. <i>PLoS ONE</i> , 2021, 16, e0251071.	1.1	2
991	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. <i>American Journal of Human Genetics</i> , 2021, 108, 857-873.	2.6	19
992	Prospects for personalization of depression treatment with genome sequencing. <i>British Journal of Pharmacology</i> , 2022, 179, 4220-4232.	2.7	3
993	Pathogenic variants in TNNC2 cause congenital myopathy due to an impaired force response to calcium. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	11
994	Identification of CYP2D6 Haplotypes that Interfere with Commonly Used Assays for Copy Number Variation Characterization. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 577-588.	1.2	10
996	Occurrence of Amyotrophic Lateral Sclerosis in Type 1 Gaucher Disease. <i>Neurology: Genetics</i> , 2021, 7, e600.	0.9	3
998	Evidence-Based Assessment of Genes in Dilated Cardiomyopathy. <i>Circulation</i> , 2021, 144, 7-19.	1.6	213
999	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. <i>Nature Medicine</i> , 2021, 27, 1197-1204.	15.2	96
1002	Biallelic splicing variants in the nucleolar 60S assembly factor RBM28 cause the ribosomopathy ANE syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	12
1005	Genetic investigations of 100 inherited cardiac disease-related genes in deceased individuals with schizophrenia. <i>International Journal of Legal Medicine</i> , 2021, 135, 1395-1405.	1.2	4
1006	Discovery of human ACE2 variants with altered recognition by the SARS-CoV-2 spike protein. <i>PLoS ONE</i> , 2021, 16, e0251585.	1.1	11
1007	A prospective trial of abiraterone acetate plus prednisone in Black and White men with metastatic castrate-resistant prostate cancer. <i>Cancer</i> , 2021, 127, 2954-2965.	2.0	21
1008	Identifying genetic modifiers of age-associated penetrance in X-linked dystonia-parkinsonism. <i>Nature Communications</i> , 2021, 12, 3216.	5.8	34

#	ARTICLE	IF	CITATIONS
1009	Mapping OMIM Disease-Related Variations on Protein Domains Reveals an Association Among Variation Type, Pfam Models, and Disease Classes. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 617016.	1.6	5
1010	African Americans and European Americans exhibit distinct gene expression patterns across tissues and tumors associated with immunologic functions and environmental exposures. <i>Scientific Reports</i> , 2021, 11, 9905.	1.6	15
1011	InÂvivo gene editing via homology-independent targeted integration for adrenoleukodystrophy treatment. <i>Molecular Therapy</i> , 2022, 30, 119-129.	3.7	9
1012	<i>CSNK2B</i> : A broad spectrum of neurodevelopmental disability and epilepsy severity. <i>Epilepsia</i> , 2021, 62, e103-e109.	2.6	13
1016	Hypomyelinating leukodystrophy - NKX6 gene variant as a cause. <i>Brain Disorders</i> , 2021, 2, 100006.	1.1	1
1017	Oligogenic Inheritance of Monoallelic TRIP11, FKBP10, NEK1, TBX5, and NBAS Variants Leading to a Phenotype Similar to Odontochondrodysplasia. <i>Frontiers in Genetics</i> , 2021, 12, 680838.	1.1	6
1018	Common and rare myocilin variants: Predicting glaucoma pathogenicity based on genetics, clinical, and laboratory misfolding data. <i>Human Mutation</i> , 2021, 42, 903-946.	1.1	13
1019	Upstream ORF frameshift variants in the <i>PAX6</i> 5'UTR cause congenital aniridia. <i>Human Mutation</i> , 2021, 42, 1053-1065.	1.1	11
1020	Analysis of Worldwide Carrier Frequency and Predicted Genetic Prevalence of Autosomal Recessive Congenital Hypothyroidism Based on a General Population Database. <i>Genes</i> , 2021, 12, 863.	1.0	8
1021	Challenges in providing residual risks in carrier testing. <i>Prenatal Diagnosis</i> , 2021, 41, 1049-1056.	1.1	5
1022	Adult diagnosis of congenital serine biosynthesis defect: A treatable cause of progressive neuropathy. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2102-2107.	0.7	9
1023	Strategies to Uplift Novel Mendelian Gene Discovery for Improved Clinical Outcomes. <i>Frontiers in Genetics</i> , 2021, 12, 674295.	1.1	23
1025	The Genetics of Adverse Drug Outcomes in Type 2 Diabetes: A Systematic Review. <i>Frontiers in Genetics</i> , 2021, 12, 675053.	1.1	6
1026	Mulibrey Nanism and the Real Time Use of Genome and Biobank Engines to Inform Clinical Care in an Ultrarare Disease. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003430.	1.6	0
1027	<i>CRY2</i> missense mutations suppress P53 and enhance cell growth. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	17
1028	Deep sequencing of 1320 genes reveals the landscape of protein-truncating variants and their contribution to psoriasis in 19,973 Chinese individuals. <i>Genome Research</i> , 2021, 31, 1150-1158.	2.4	5
1029	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. <i>American Journal of Human Genetics</i> , 2021, 108, 965-982.	2.6	35
1030	Biased pathogenic assertions of loss of function variants challenge molecular diagnosis of admixed individuals. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2021, 187, 357-363.	0.7	4

#	ARTICLE	IF	CITATIONS
1032	Carrier frequency and predicted genetic prevalence of Pompe disease based on a general population database. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 27, 100734.	0.4	16
1034	Exome sequencing in obsessive-compulsive disorder reveals a burden of rare damaging coding variants. <i>Nature Neuroscience</i> , 2021, 24, 1071-1076.	7.1	35
1035	Mechnetor: a web server for exploring protein mechanism and the functional context of genetic variants. <i>Nucleic Acids Research</i> , 2021, 49, W366-W374.	6.5	3
1036	Global spectrum of population-specific common missense variation in cytochrome P450 pharmacogenes. <i>Human Mutation</i> , 2021, 42, 1107-1123.	1.1	1
1037	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. <i>American Journal of Human Genetics</i> , 2021, 108, 1053-1068.	2.6	31
1038	mSWI/SNF promotes Polycomb repression both directly and through genome-wide redistribution. <i>Nature Structural and Molecular Biology</i> , 2021, 28, 501-511.	3.6	50
1039	Phenotypic diversity, disease progression, and pathogenicity of <i>MVK</i> missense variants in mevalonic aciduria. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1272-1287.	1.7	17
1040	Recurrent <i>KCNT2</i> missense variants affecting p.Arg190 result in a recognizable phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3083-3091.	0.7	7
1041	Mutational Landscape of Pirin and Elucidation of the Impact of Most Detrimental Missense Variants That Accelerate the Breast Cancer Pathways: A Computational Modelling Study. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 692835.	1.6	27
1042	Genetic architecture of 11 organ traits derived from abdominal MRI using deep learning. <i>ELife</i> , 2021, 10, .	2.8	102
1045	Understanding the genetics of adult-onset dilated cardiomyopathy: what a clinician needs to know. <i>European Heart Journal</i> , 2021, 42, 2384-2396.	1.0	28
1046	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , 2021, 108, 1138-1150.	2.6	17
1047	Prevalence and spectrum of DNA mismatch repair gene variation in the general Chinese population. <i>Journal of Medical Genetics</i> , 2022, 59, 652-661.	1.5	9
1050	Molecular Inversion Probe-Based Sequencing of USH2A Exons and Splice Sites as a Cost-Effective Screening Tool in USH2 and arRP Cases. <i>International Journal of Molecular Sciences</i> , 2021, 22, 6419.	1.8	8
1051	Germline ERBB2/HER2 Coding Variants Are Associated with Increased Risk of Myeloproliferative Neoplasms. <i>Cancers</i> , 2021, 13, 3246.	1.7	5
1053	Operative list of genes associated with autism and neurodevelopmental disorders based on database review. <i>Molecular and Cellular Neurosciences</i> , 2021, 113, 103623.	1.0	51
1054	Machine Learning-Based Approach Highlights the Use of a Genomic Variant Profile for Precision Medicine in Ovarian Failure. <i>Journal of Personalized Medicine</i> , 2021, 11, 609.	1.1	6
1055	TEAD family transcription factors in development and disease. <i>Development (Cambridge)</i> , 2021, 148, .	1.2	37

#	ARTICLE	IF	CITATIONS
1056	Internal Standardization of the Interpretation and Reporting of Sequence Variants in Hematologic Neoplasms. <i>Molecular Diagnosis and Therapy</i> , 2021, 25, 517-526.	1.6	4
1058	Whole exome sequencing of large populations: identification of loss of function alleles and implications for inherited kidney diseases. <i>Kidney International</i> , 2021, 99, 1255-1259.	2.6	2
1059	Mosaic de novo <i>SNRPN</i> gene variant associated with Prader-Willi syndrome. <i>Journal of Medical Genetics</i> , 2022, 59, 719-722.	1.5	6
1060	The prevalence, genetic complexity and population-specific founder effects of human autosomal recessive disorders. <i>Npj Genomic Medicine</i> , 2021, 6, 41.	1.7	17
1061	Computational Tools to Assess the Functional Consequences of Rare and Noncoding Pharmacogenetic Variability. <i>Clinical Pharmacology and Therapeutics</i> , 2021, 110, 626-636.	2.3	16
1062	Eight novel susceptibility loci and putative causal variants in atopic dermatitis. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 1293-1306.	1.5	32
1064	Loss of GSK-3 β mediated phosphorylation in HtrA2 contributes to uncontrolled cell death with Parkinsonian phenotype. <i>International Journal of Biological Macromolecules</i> , 2021, 180, 97-111.	3.6	3
1065	Unraveling the genetic complexities of combined retinal dystrophy and hearing impairment. <i>Human Genetics</i> , 2022, 141, 785-803.	1.8	6
1066	Increased diagnosis of enlarged vestibular aqueduct by multiplex PCR enrichment and next-generation sequencing of the <i>SLC26A4</i> gene. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1734.	0.6	10
1067	Monogenic Childhood Diabetes: Dissecting Clinical Heterogeneity by Next-Generation Sequencing in Maturity-Onset Diabetes of the Young. <i>OMICS A Journal of Integrative Biology</i> , 2021, 25, 431-449.	1.0	12
1068	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. <i>Nature Communications</i> , 2021, 12, 3505.	5.8	49
1069	Plasma Mucin-1 (CA15-3) Levels in Autosomal Dominant Tubulointerstitial Kidney Disease due to <i>MUC1</i> Mutations. <i>American Journal of Nephrology</i> , 2021, 52, 378-387.	1.4	4
1070	Maternal iron deficiency perturbs embryonic cardiovascular development in mice. <i>Nature Communications</i> , 2021, 12, 3447.	5.8	17
1072	Effect of rare coding variants of charged amino acid residues on the function of human organic anion transporting polypeptide 1B3 (SLCO1B3). <i>Biochemical and Biophysical Research Communications</i> , 2021, 557, 1-7.	1.0	4
1073	The Combined Human Genotype of Truncating TTN and RBM20 Mutations Is Associated with Severe and Early Onset of Dilated Cardiomyopathy. <i>Genes</i> , 2021, 12, 883.	1.0	15
1074	An identical-by-descent novel splice-donor variant in <i>PRUNE1</i> causes a neurodevelopmental syndrome with prominent dystonia in two consanguineous Sudanese families. <i>Annals of Human Genetics</i> , 2021, 85, 186-195.	0.3	5
1075	ADAMTS1, MPDZ, MVD, and SEZ6: candidate genes for autosomal recessive nonsyndromic hearing impairment. <i>European Journal of Human Genetics</i> , 2021, , .	1.4	6
1076	Soluble CD14 Levels in the Jackson Heart Study: Associations With Cardiovascular Disease Risk and Genetic Variants. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2021, 41, e369-e378.	1.1	5

#	ARTICLE	IF	CITATIONS
1077	Genetic Characterization of Cancer of Unknown Primary Using Liquid Biopsy Approaches. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 666156.	1.8	12
1078	Evaluating the clinical utility of early exome sequencing in diverse pediatric outpatient populations in the North Carolina Clinical Genomic Evaluation of Next-generation Exome Sequencing (NCGENES) 2 study: a randomized controlled trial. <i>Trials</i> , 2021, 22, 395.	0.7	5
1080	Identifying the Lipidomic Effects of a Rare Loss-of-Function Deletion in <i>ANGPTL3</i> . <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003232.	1.6	3
1083	Leveraging supervised learning for functionally informed fine-mapping of cis-eQTLs identifies an additional 20,913 putative causal eQTLs. <i>Nature Communications</i> , 2021, 12, 3394.	5.8	44
1084	Role of calcium/calmodulin-dependent kinase 2 in neurodevelopmental disorders. <i>Brain Research Bulletin</i> , 2021, 171, 209-220.	1.4	13
1085	A mosaic PIK3CA variant in a young adult with diffuse gastric cancer: case report. <i>European Journal of Human Genetics</i> , 2021, 29, 1354-1358.	1.4	9
1086	Non-coding region variants upstream of MEF2C cause severe developmental disorder through three distinct loss-of-function mechanisms. <i>American Journal of Human Genetics</i> , 2021, 108, 1083-1094.	2.6	42
1088	Pseudoendocrine Sarcoma. <i>American Journal of Surgical Pathology</i> , 2022, 46, 33-43.	2.1	16
1090	ALG3-CDG: a patient with novel variants and review of the genetic and ophthalmic findings. <i>BMC Ophthalmology</i> , 2021, 21, 249.	0.6	4
1091	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. <i>American Journal of Human Genetics</i> , 2021, 108, 1069-1082.	2.6	8
1092	Common polymorphic <i>OTC</i> variants can act as genetic modifiers of enzymatic activity. <i>Human Mutation</i> , 2021, 42, 978-989.	1.1	6
1093	RNA-Seq Data for Reliable SNP Detection and Genotype Calling: Interest for Coding Variant Characterization and Cis-Regulation Analysis by Allele-Specific Expression in Livestock Species. <i>Frontiers in Genetics</i> , 2021, 12, 655707.	1.1	30
1094	Genome sequencing of 320 Chinese children with epilepsy: a clinical and molecular study. <i>Brain</i> , 2021, 144, 3623-3634.	3.7	13
1095	Association between NF- κ B polymorphism and age-related macular degeneration in a high-altitude population. <i>PLoS ONE</i> , 2021, 16, e0251931.	1.1	5
1096	The Diagnostic Journey of a Patient with Prader-Willi-Like Syndrome and a Unique Homozygous SNURF-SNRPN Variant; Bio-Molecular Analysis and Review of the Literature. <i>Genes</i> , 2021, 12, 875.	1.0	4
1097	Variant Reinterpretation in Survivors of Cardiac Arrest With Preserved Ejection Fraction (the Cardiac Tj ETQq1 1 0.784314 rgBT /Over Laboratories. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003235.	1.6	10
1099	Variant analyses of candidate genes in orofacial clefts in multiethnic populations. <i>Oral Diseases</i> , 2022, 28, 1921-1935.	1.5	3
1100	Systematic analysis of exonic germline and postzygotic de novo mutations in bipolar disorder. <i>Nature Communications</i> , 2021, 12, 3750.	5.8	15

#	ARTICLE	IF	CITATIONS
1102	Novel Variant in CLDN16: A Further Step in the Diagnosis of Familial Hypomagnesemia with Hypercalciuria and Nephrocalcinosisâ€”A Case Report. <i>Uro</i> , 2021, 1, 76-81.	0.3	0
1103	Functional characterization of 105 factor H variants associated with aHUS: lessons for variant classification. <i>Blood</i> , 2021, 138, 2185-2201.	0.6	29
1104	Germline POT1 variants can predispose to myeloid and lymphoid neoplasms. <i>Leukemia</i> , 2022, 36, 283-287.	3.3	17
1105	Genome-wide association analysis of metabolic syndrome quantitative traits in the GENNID multiethnic family study. <i>Diabetology and Metabolic Syndrome</i> , 2021, 13, 59.	1.2	9
1106	A new method to accurately identify single nucleotide variants using small FFPE breast samples. <i>Briefings in Bioinformatics</i> , 2021, 22, .	3.2	4
1107	<i>CDKN2A</i> Alterations and Response to Immunotherapy in Solid Tumors. <i>Clinical Cancer Research</i> , 2021, 27, 4025-4035.	3.2	51
1108	Rapid genotype imputation from sequence with reference panels. <i>Nature Genetics</i> , 2021, 53, 1104-1111.	9.4	47
1109	Genotype-Phenotype Associations in Patients With Type-1, Type-2, and Atypical NF1 Microdeletions. <i>Frontiers in Genetics</i> , 2021, 12, 673025.	1.1	11
1110	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. <i>Genetics in Medicine</i> , 2021, 23, 1901-1911.	1.1	9
1111	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. <i>European Journal of Human Genetics</i> , 2021, 29, 1325-1331.	1.4	49
1112	MRGPRX4 in Cholestatic Pruritus. <i>Seminars in Liver Disease</i> , 2021, 41, 358-367.	1.8	6
1113	A deep learning approach to identify gene targets of a therapeutic for human splicing disorders. <i>Nature Communications</i> , 2021, 12, 3332.	5.8	26
1114	Modern diagnostic and therapeutic approaches in familial maculopathy with reference to North Carolina macular dystrophy. <i>Biomedical Papers of the Medical Faculty of the University Palacky&#x0301;, Olomouc, Czechoslovakia</i> , 2022, 166, 418-427.	0.2	2
1115	Mutational Landscape of the Proglucagon-Derived Peptides. <i>Frontiers in Endocrinology</i> , 2021, 12, 698511.	1.5	7
1116	A novel, germline, deactivating CBL variant p.L493F alters domain orientation and is associated with multiple childhood cancers. <i>Cancer Genetics</i> , 2021, 254-255, 18-24.	0.2	2
1120	Biallelic loss of <i>ERGIC1</i> causes relatively mild arthrogryposis. <i>Clinical Genetics</i> , 2021, 100, 329-333.	1.0	2
1121	Heterozygous <i>OAS1</i> gain-of-function variants cause an autoinflammatory immunodeficiency. <i>Science Immunology</i> , 2021, 6, .	5.6	36
1122	Surveillance recommendations for DICER1 pathogenic variant carriers: a report from the SIOPE Host Genome Working Group and CanGene-CanVar Clinical Guideline Working Group. <i>Familial Cancer</i> , 2021, 20, 337-348.	0.9	19

#	ARTICLE	IF	CITATIONS
1123	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. <i>Genetics in Medicine</i> , 2021, 23, 1889-1900.	1.1	13
1124	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. <i>Genetics in Medicine</i> , 2021, 23, 1922-1932.	1.1	16
1125	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. <i>Genetics in Medicine</i> , 2021, 23, 1952-1960.	1.1	7
1128	Report of two siblings with spondylodysplastic Ehlers-Danlos syndrome and B4GALT7 deficiency. <i>BMC Pediatrics</i> , 2021, 21, 293.	0.7	2
1130	Association of Vitamin D, Zinc and Selenium Related Genetic Variants With COVID-19 Disease Severity. <i>Frontiers in Nutrition</i> , 2021, 8, 689419.	1.6	22
1131	<sc>Î±â€šynuclein</sc> Deposition in Sympathetic Nerve Fibers in Genetic Forms of Parkinson's Disease. <i>Movement Disorders</i> , 2021, 36, 2346-2357.	2.2	11
1132	Proximal variants in <sc><i>CCND2</i></sc> associated with microcephaly, short stature, and developmental delay: A case series and review of inverse brain growth phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2719-2738.	0.7	14
1135	Loss-of-function variants in <i>DNM1</i> cause a specific form of developmental and epileptic encephalopathy only in biallelic state. <i>Journal of Medical Genetics</i> , 2022, 59, 549-553.	1.5	9
1136	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. <i>Genetics in Medicine</i> , 2021, 23, 1705-1714.	1.1	22
1137	Biallelic truncating variants in <i>ATP9A</i> cause a novel neurodevelopmental disorder involving postnatal microcephaly and failure to thrive. <i>Journal of Medical Genetics</i> , 2022, 59, 662-668.	1.5	9
1140	BRCA1 and BRCA2 Variation in Taiwanese General Population and the Cancer Cohort. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 685174.	1.6	9
1141	Homozygous <sc><i>WNT9B</i></sc> variants in two families with bilateral renal agenesis/hypoplasia/dysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3005-3011.	0.7	5
1142	Advances in clinical genetics and genomics. <i>Intelligent Medicine</i> , 2021, 1, 128-133.	1.6	4
1143	Revisiting the pathogenic mechanism of the CJB1 5â€š™ UTR c.-103Câ€š%â€šT mutation causing CMTX1. <i>Neurogenetics</i> , 2021, 22, 149-160.	0.7	1
1145	Saturation mutagenesis defines novel mouse models of severe spine deformity. <i>DMM Disease Models and Mechanisms</i> , 2021, 14, .	1.2	4
1146	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. <i>European Journal of Human Genetics</i> , 2021, 29, 1337-1347.	1.4	34
1147	Gabra2 is a genetic modifier of Dravet syndrome in mice. <i>Mammalian Genome</i> , 2021, 32, 350-363.	1.0	11
1148	Pharmacogene Sequencing of a Gabonese Population with Severe Plasmodium falciparum Malaria Reveals Multiple Novel Variants with Putative Relevance for Antimalarial Treatment. <i>Antimicrobial Agents and Chemotherapy</i> , 2021, 65, e0027521.	1.4	6

#	ARTICLE	IF	CITATIONS
1149	Quantitative Alterations in Complement Alternative Pathway and Related Genetic Analysis in Severe Phenotype Preeclampsia. <i>Kidney360</i> , 2021, 2, 1463-1472.	0.9	2
1150	The genetic architecture of Plakophilin 2 cardiomyopathy. <i>Genetics in Medicine</i> , 2021, 23, 1961-1968.	1.1	13
1151	Advancing human genetics research and drug discovery through exome sequencing of the UK Biobank. <i>Nature Genetics</i> , 2021, 53, 942-948.	9.4	234
1152	Contribution of Multiple Inherited Variants to Autism Spectrum Disorder (ASD) in a Family with 3 Affected Siblings. <i>Genes</i> , 2021, 12, 1053.	1.0	12
1153	Genomic characteristics of invasive mucinous adenocarcinoma of the lung with multiple pulmonary sites of involvement. <i>Modern Pathology</i> , 2022, 35, 202-209.	2.9	12
1154	An overview of germline variations in genes of primary immunodeficiencies through integrative analysis of ClinVar, HGMD® and dbSNP databases. <i>Human Genetics</i> , 2021, 140, 1379-1393.	1.8	3
1155	Clinical likelihood ratios and balanced accuracy for 44 in silico tools against multiple large-scale functional assays of cancer susceptibility genes. <i>Genetics in Medicine</i> , 2021, 23, 2096-2104.	1.1	41
1156	A Curriculum for Genomic Education of Molecular Genetic Pathology Fellows. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 1218-1240.	1.2	4
1157	A COVID-associated variant in the ciliogenesis protein <i>CCDC28B</i> disrupts immune synapse assembly. <i>Cell Death and Differentiation</i> , 2022, 29, 65-81.	5.0	5
1158	Towards personalized medicine for amyotrophic lateral sclerosis. <i>Trends in Endocrinology and Metabolism</i> , 2021, 32, 839-841.	3.1	1
1159	Delineating the genotypic and phenotypic spectrum of <i>HECW2</i> -related neurodevelopmental disorders. <i>Journal of Medical Genetics</i> , 2022, 59, 669-677.	1.5	5
1160	Multiple Acyl-CoA Dehydrogenase Deficiency with Variable Presentation Due to a Homozygous Mutation in a Bedouin Tribe. <i>Genes</i> , 2021, 12, 1140.	1.0	1
1161	Genetic and Molecular Aspects of Drug-Induced QT Interval Prolongation. <i>International Journal of Molecular Sciences</i> , 2021, 22, 8090.	1.8	17
1162	The first insight into the genetic structure of the population of modern Serbia. <i>Scientific Reports</i> , 2021, 11, 13995.	1.6	1
1163	Molecular and Pathological Profiling of Corresponding Treatment-Naïve and Neoadjuvant Pazopanib-Treated High-Risk Soft Tissue Sarcoma Samples of the GISG-04/NOPASS Study. <i>Biology</i> , 2021, 10, 639.	1.3	1
1164	Clinical and genomic characterization of 8p cytogenomic disorders. <i>Genetics in Medicine</i> , 2021, 23, 2342-2351.	1.1	3
1166	Immune Dysregulation in Human <i>ITCH</i> Deficiency Successfully Treated with Hematopoietic Cell Transplantation. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2021, 9, 2885-2893.e3.	2.0	4
1167	Contribution of <i>HCN1</i> variant to sinus bradycardia: A case report. <i>Journal of Arrhythmia</i> , 2021, 37, 1337-1347.	0.5	2

#	ARTICLE	IF	CITATIONS
1168	Mutation spectrum of the OPA1 gene in a large cohort of patients with suspected dominant optic atrophy: Identification and classification of 48 novel variants. PLoS ONE, 2021, 16, e0253987.	1.1	18
1169	Exome variant discrepancies due to reference-genome differences. American Journal of Human Genetics, 2021, 108, 1239-1250.	2.6	36
1170	Germ-line mutations in <i>WDR77</i> predispose to familial papillary thyroid cancer. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	13
1171	Prevalence of POC5 Coding Variants in French-Canadian and British AIS Cohort. Genes, 2021, 12, 1032.	1.0	4
1173	A Multi-center Genome-wide Association Study of Cervical Dystonia. Movement Disorders, 2021, 36, 2795-2801.	2.2	5
1174	Interpretable deep learning uncovers cellular properties in label-free live cell images that are predictive of highly metastatic melanoma. Cell Systems, 2021, 12, 733-747.e6.	2.9	48
1175	Myopathy can be a key phenotype of membrin (GOSR2) deficiency. Human Mutation, 2021, 42, 1101-1106.	1.1	3
1176	Association of Rare Genetic Variants and Early-Onset Atrial Fibrillation in Ethnic Minority Individuals. JAMA Cardiology, 2021, 6, 811.	3.0	30
1177	Functional and structural analyses of novel Smith-Kingsmore Syndrome-Associated MTOR variants reveal potential new mechanisms and predictors of pathogenicity. PLoS Genetics, 2021, 17, e1009651.	1.5	9
1179	Three-dimensional missense tolerance ratio analysis. Genome Research, 2021, 31, 1447-1461.	2.4	14
1180	GIGYF1 loss of function is associated with clonal mosaicism and adverse metabolic health. Nature Communications, 2021, 12, 4178.	5.8	20
1181	Identification of autosomal recessive nonsyndromic hearing impairment genes through the study of consanguineous and non-consanguineous families: past, present, and future. Human Genetics, 2022, 141, 413-430.	1.8	2
1183	Epilepsy Syndromes in the First Year of Life and Usefulness of Genetic Testing for Precision Therapy. Genes, 2021, 12, 1051.	1.0	36
1184	Further evidence for <i>de novo</i> variants in <i>SYNCRIP</i> as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	1.1	9
1185	Comparative Analysis of Mammal Genomes Unveils Key Genomic Variability for Human Life Span. Molecular Biology and Evolution, 2021, 38, 4948-4961.	3.5	15
1186	A molecular genetics view on Mucopolysaccharidosis Type II. Mutation Research - Reviews in Mutation Research, 2021, 788, 108392.	2.4	9
1187	<i>PLXNA2</i> as a candidate gene in patients with intellectual disability. American Journal of Medical Genetics, Part A, 2021, 185, 3859-3865.	0.7	6
1188	Differential Domain Distribution of gnomAD- and Disease-Linked Connexin Missense Variants. International Journal of Molecular Sciences, 2021, 22, 7832.	1.8	7

#	ARTICLE	IF	CITATIONS
1190	Comprehensive Genomic and Transcriptomic Analysis of Three Synchronous Primary Tumours and a Recurrence from a Head and Neck Cancer Patient. <i>International Journal of Molecular Sciences</i> , 2021, 22, 7583.	1.8	3
1191	Human autoinflammatory disease reveals ELF4 as a transcriptional regulator of inflammation. <i>Nature Immunology</i> , 2021, 22, 1118-1126.	7.0	30
1192	Inherited Retinal Diseases Due to RPE65 Variants: From Genetic Diagnostic Management to Therapy. <i>International Journal of Molecular Sciences</i> , 2021, 22, 7207.	1.8	19
1193	Neptune: an environment for the delivery of genomic medicine. <i>Genetics in Medicine</i> , 2021, 23, 1838-1846.	1.1	3
1197	Multimodal imaging of Hypotrichosis with juvenile macular dystrophy: a case report. <i>BMC Ophthalmology</i> , 2021, 21, 284.	0.6	1
1199	P2T2: Protein Panoramic annoTation Tool for the interpretation of protein coding genetic variants. <i>JAMIA Open</i> , 2021, 4, ooab065.	1.0	1
1200	Hearing Impairment with Monoallelic GJB2 Variants. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 1279-1291.	1.2	10
1206	Copy Number Variation in Parkinson's Disease: An Update from <sc>Sub-Saharan</sc> Africa. <i>Movement Disorders</i> , 2021, 36, 2442-2444.	2.2	4
1207	Splicing in the Diagnosis of Rare Disease: Advances and Challenges. <i>Frontiers in Genetics</i> , 2021, 12, 689892.	1.1	41
1208	Prevalence of cancer susceptibility variants in patients with multiple Lynch syndrome related cancers. <i>Scientific Reports</i> , 2021, 11, 14807.	1.6	8
1209	Association of the <i>MYOC</i> p.(Gln368Ter) Variant With Glaucoma in a Finnish Population. <i>JAMA Ophthalmology</i> , 2021, 139, 762.	1.4	7
1210	Unveiling the genetic variation of severe continuous/mixed-type ossification of the posterior longitudinal ligament by whole-exome sequencing and bioinformatic analysis. <i>Spine Journal</i> , 2021, 21, 1847-1856.	0.6	5
1211	MYH7-related disorders in two Bulgarian families: Novel variants in the same region associated with different clinical manifestation and disease penetrance. <i>Neuromuscular Disorders</i> , 2021, 31, 633-641.	0.3	2
1212	Kallmann syndrome in a patient with Weissâ€Kruszka syndrome and a de novo deletion in 9q31.2. <i>European Journal of Endocrinology</i> , 2021, 185, 57-66.	1.9	8
1213	Quantitative disease risk scores from EHR with applications to clinical risk stratification and genetic studies. <i>Npj Digital Medicine</i> , 2021, 4, 116.	5.7	7
1214	Ligand-directed bias of G protein signaling at the dopamine D2 receptor. <i>Cell Chemical Biology</i> , 2022, 29, 226-238.e4.	2.5	14
1215	Genetics of Paroxysmal Dyskinesia: Novel Variants Corroborate the Role of KCNA1 in Paroxysmal Dyskinesia and Highlight the Diverse Phenotypic Spectrum of KCNA1- and SLC2A1-Related Disorders. <i>Frontiers in Neurology</i> , 2021, 12, 701351.	1.1	3
1216	NUDT15 polymorphism influences the metabolism and therapeutic effects of acyclovir and ganciclovir. <i>Nature Communications</i> , 2021, 12, 4181.	5.8	11

#	ARTICLE	IF	CITATIONS
1218	ZTTK syndrome: Clinical and molecular findings of 15 cases and a review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3740-3753.	0.7	11
1219	Discovery of genomic variation across a generation. <i>Human Molecular Genetics</i> , 2021, 30, R174-R186.	1.4	9
1221	Meta-Analysis of Circulating Cell-Free DNA's Role in the Prognosis of Pancreatic Cancer. <i>Cancers</i> , 2021, 13, 3378.	1.7	9
1222	DECODE: a Deep-learning framework for Connecting enhancers and refining boundaries with large-scale functional assays. <i>Bioinformatics</i> , 2021, 37, i280-i288.	1.8	8
1224	Case Report: Two Distinct Focal Congenital Hyperinsulinism Lesions Resulting From Separate Genetic Events. <i>Frontiers in Pediatrics</i> , 2021, 9, 699129.	0.9	3
1225	Mutants of human ACE2 differentially promote SARS-CoV and SARS-CoV-2 spike mediated infection. <i>PLoS Pathogens</i> , 2021, 17, e1009715.	2.1	24
1226	Genomic frontiers in congenital heart disease. <i>Nature Reviews Cardiology</i> , 2022, 19, 26-42.	6.1	93
1227	Somatic mutations in benign breast disease tissues and association with breast cancer risk. <i>BMC Medical Genomics</i> , 2021, 14, 185.	0.7	2
1228	Second patient with GNB2-related neurodevelopmental disease: Further evidence for a gene-disease association. <i>European Journal of Medical Genetics</i> , 2021, 64, 104243.	0.7	0
1229	Re-analysis of whole-exome sequencing data reveals a novel splicing variant in the SLC2A1 in a patient with GLUT1 Deficiency Syndrome 1 accompanied by hemangioma: a case report. <i>BMC Medical Genomics</i> , 2021, 14, 197.	0.7	1
1230	Germline ERBB3 mutation in familial non-small-cell lung carcinoma: expanding ErbB's role in oncogenesis. <i>Human Molecular Genetics</i> , 2021, 30, 2393-2401.	1.4	3
1231	Whole-exome sequencing reveals a monogenic cause in 56% of individuals with laterality disorders and associated congenital heart defects. <i>Journal of Medical Genetics</i> , 2022, 59, 691-696.	1.5	14
1232	Clinical and molecular characteristics of 168 probands and 65 relatives with a clinical presentation of classical Ehlers-Danlos syndrome. <i>Human Mutation</i> , 2021, 42, 1294-1306.	1.1	12
1233	Pathogenic noncoding variants in the neurofibromatosis and schwannomatosis predisposition genes. <i>Human Mutation</i> , 2021, 42, 1187-1207.	1.1	5
1234	Association of Coding Variants in Hydroxysteroid 17-beta Dehydrogenase 14 (HSD17B14) with Reduced Progression to End Stage Kidney Disease in Type 1 Diabetes. <i>Journal of the American Society of Nephrology: JASN</i> , 2021, 32, 2634-2651.	3.0	9
1235	A Novel Pathogenic NOD2 Variant in a Mother and Daughter with Blau Syndrome. <i>Ophthalmic Genetics</i> , 2021, 42, 1-12.	0.5	3
1236	Association Analysis of Candidate Variants in Admixed Brazilian Patients With Genetic Generalized Epilepsies. <i>Frontiers in Genetics</i> , 2021, 12, 672304.	1.1	1
1237	Secondary resistance to anti-EGFR therapy by transcriptional reprogramming in patient-derived colorectal cancer models. <i>Genome Medicine</i> , 2021, 13, 116.	3.6	10

#	ARTICLE	IF	CITATIONS
1238	Novel CLTC variants cause new brain and kidney phenotypes. <i>Journal of Human Genetics</i> , 2021, , .	1.1	4
1240	SMARCA4 and Other SWItch/Sucrose NonFermentable Family Genomic Alterations in NSCLC: Clinicopathologic Characteristics and Outcomes to Immune Checkpoint Inhibition. <i>Journal of Thoracic Oncology</i> , 2021, 16, 1176-1187.	0.5	49
1241	Two patients with chronic mucocutaneous candidiasis caused by TRAF3IP2 deficiency. <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 256-261.e2.	1.5	10
1242	Haplotype-resolved germline and somatic alterations in renal medullary carcinomas. <i>Genome Medicine</i> , 2021, 13, 114.	3.6	5
1243	Germline ATM variants predispose to melanoma: a joint analysis across the GenoMEL and MelaNostrum consortia. <i>Genetics in Medicine</i> , 2021, 23, 2087-2095.	1.1	19
1244	Coding and noncoding variants in EBF3 are involved in HADDs and simplex autism. <i>Human Genomics</i> , 2021, 15, 44.	1.4	16
1245	Transient developmental delays in infants with Duarte-2 variant galactosemia. <i>Molecular Genetics and Metabolism</i> , 2021, 134, 132-138.	0.5	8
1246	Common deletion variants causing protocadherin-1± deficiency contribute to the complex genetics of BAV and left-sided congenital heart disease. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100037.	1.0	7
1247	Phenotypic variability of a TREX1 variant in Aicardi-Goutieres type 1 patients from the Indian subcontinent. <i>European Journal of Medical Genetics</i> , 2021, 64, 104291.	0.7	1
1248	A Nationwide Study on the Impact of Routine Testing for EGFR Mutations in Advanced NSCLC Reveals Distinct Survival Patterns Based on EGFR Mutation Subclasses. <i>Cancers</i> , 2021, 13, 3641.	1.7	11
1249	African Global Representation in Biomedical Sciences. <i>Annual Review of Biomedical Data Science</i> , 2021, 4, 57-81.	2.8	3
1250	Characterization of Genetic Variants in the SLC5A5 Gene and Associations With Breast Milk Iodine Concentration in Lactating Women of African Descent: The NUPED Study. <i>Frontiers in Nutrition</i> , 2021, 8, 692504.	1.6	2
1252	Brain-wide Cas9-mediated cleavage of a gene causing familial Alzheimer's disease alleviates amyloid-related pathologies in mice. <i>Nature Biomedical Engineering</i> , 2022, 6, 168-180.	11.6	27
1253	Characterization of a novel variant in the HR1 domain of MFN2 in a patient with ataxia, optic atrophy and sensorineural hearing loss. <i>F1000Research</i> , 0, 10, 606.	0.8	3
1254	Rare Coding Variants Associated With Electrocardiographic Intervals Identify Monogenic Arrhythmia Susceptibility Genes: A Multi-Ancestry Analysis. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003300.	1.6	7
1255	Genetic Screening for TLR7 Variants in Young and Previously Healthy Men With Severe COVID-19. <i>Frontiers in Immunology</i> , 2021, 12, 719115.	2.2	76
1256	Mapping the human genetic architecture of COVID-19. <i>Nature</i> , 2021, 600, 472-477.	13.7	640
1257	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. <i>Nature Genetics</i> , 2021, 53, 1006-1021.	9.4	44

#	ARTICLE	IF	CITATIONS
1258	The PDE-Opathies: Diverse Phenotypes Produced by a Functionally Related Multigene Family. Trends in Genetics, 2021, 37, 669-681.	2.9	18
1260	Next-Generation Sequencing Enhances the Diagnosis Efficiency in Thyroid Nodules. Frontiers in Oncology, 2021, 11, 677892.	1.3	5
1261	Transcriptional Regulation of RUNX1: An Informatics Analysis. Genes, 2021, 12, 1175.	1.0	4
1262	TUMOSPEC: A Nation-Wide Study of Hereditary Breast and Ovarian Cancer Families with a Predicted Pathogenic Variant Identified through Multigene Panel Testing. Cancers, 2021, 13, 3659.	1.7	4
1263	A Reassessment of Copy Number Variations in Congenital Heart Defects: Picturing the Whole Genome. Genes, 2021, 12, 1048.	1.0	6
1264	Do monogenic inborn errors of immunity cause susceptibility to severe COVID-19?. Journal of Clinical Investigation, 2021, 131, .	3.9	3
1265	Summix: A method for detecting and adjusting for population structure in genetic summary data. American Journal of Human Genetics, 2021, 108, 1270-1282.	2.6	5
1268	GWAS of serum ALT and AST reveals an association of SLC30A10 Thr95Ile with hypermanganesemia symptoms. Nature Communications, 2021, 12, 4571.	5.8	26
1269	Biallelic loss of function variants in STAG3 result in primary ovarian insufficiency. Reproductive BioMedicine Online, 2021, 43, 899-902.	1.1	5
1270	Novel MFRP Mutation with Nanophthalmos, Optic Disc Drusen, and Peripheral Retinoschisis Imaged with Ultra-Widefield OCT. Retinal Cases and Brief Reports, 2021, Publish Ahead of Print, .	0.3	0
1271	G6PD distribution in sub-Saharan Africa and potential risks of using chloroquine/hydroxychloroquine based treatments for COVID-19. Pharmacogenomics Journal, 2021, 21, 649-656.	0.9	11
1272	Current gene panels account for nearly all homologous recombination repair-associated multiple-case breast cancer families. Npj Breast Cancer, 2021, 7, 109.	2.3	3
1273	Identification of a novel COL10A1 : c.1952 G>T variant in a family with Schmid metaphyseal chondrodysplasia and development of a noninvasive prenatal testing method. Molecular Genetics & Genomic Medicine, 2021, 9, e1758.	0.6	2
1274	Gene Alterations and Expression Spectrum of NANOS3 in Nonobstructive Azoospermia. Reproductive Sciences, 2021, , 1.	1.1	0
1275	Intrahepatic Cholestasis, Refractory Epilepsy, Skeletal Dysplasia, Endocrine Failure, and Dysmorphic Features in a Child With a Monoallelic 2q24-32.2 Deletion Encompassing ABCB11. Pediatric and Developmental Pathology, 2021, , 109352662110360.	0.5	1
1276	Prognostic Roles of BRAF, KIT, NRAS, IGF2R and SF3B1 Mutations in Mucosal Melanomas. Cells, 2021, 10, 2216.	1.8	8
1279	Exploring absent protein function in yeast: assaying post translational modification and human genetic variation. Microbial Cell, 2021, 8, 164-183.	1.4	10
1280	Lifetime risk of rheumatoid arthritis-associated interstitial lung disease in <i>MUC5B</i> mutation carriers. Annals of the Rheumatic Diseases, 2021, 80, 1530-1536.	0.5	25

#	ARTICLE	IF	CITATIONS
1281	Rates and Patterns of Clonal Oncogenic Mutations in the Normal Human Brain. <i>Cancer Discovery</i> , 2022, 12, 172-185.	7.7	19
1283	The HuaBiao project: whole-exome sequencing of 5000 Han Chinese individuals. <i>Journal of Genetics and Genomics</i> , 2021, 48, 1032-1035.	1.7	22
1284	An updated quantitative model to classify missense variants in the <i>TP53</i> gene: A novel multifactorial strategy. <i>Human Mutation</i> , 2021, 42, 1351-1361.	1.1	7
1285	Trigenic ADH5/ALDH2/ADGRV1 mutations in myelodysplasia with Usher syndrome. <i>Heliyon</i> , 2021, 7, e07804.	1.4	2
1286	Neutrophils require SKAP2 for reactive oxygen species production following C-type lectin and <i>Candida</i> stimulation. <i>IScience</i> , 2021, 24, 102871.	1.9	7
1287	Assessment of ANG variants in Parkinson's disease. <i>Neurobiology of Aging</i> , 2021, 104, 111.e1-111.e4.	1.5	1
1289	Wide Fontanels, Delayed Speech Development and Hoarse Voice as Useful Signs in the Diagnosis of KBC Syndrome: A Clinical Description of 23 Cases with Pathogenic Variants Involving the ANKRD11 Gene or Submicroscopic Chromosomal Rearrangements of 16q24.3. <i>Genes</i> , 2021, 12, 1257.	1.0	10
1291	Case Report: Identification of a Novel Homozygous Mutation in GPD1 Gene of a Chinese Child With Transient Infantile Hypertriglyceridemia. <i>Frontiers in Genetics</i> , 2021, 12, 726116.	1.1	4
1292	Human MLH1/3 variants causing aneuploidy, pregnancy loss, and premature reproductive aging. <i>Nature Communications</i> , 2021, 12, 5005.	5.8	13
1293	BAFF receptor polymorphisms and deficiency in humans. <i>Current Opinion in Immunology</i> , 2021, 71, 103-110.	2.4	7
1295	Exome sequencing of child-parent trios with bladder exstrophy: Findings in 26 children. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3028-3041.	0.7	4
1296	Undetectable NK Cells due to the FCGR3A Variant, L66H, Which May Not Be Directly Disease-Causing. <i>Journal of Clinical Immunology</i> , 2021, 41, 1957-1959.	2.0	1
1297	The genetic structure of the Turkish population reveals high levels of variation and admixture. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	42
1299	Biallelic <i>PI4KA</i> variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. <i>Brain</i> , 2021, 144, 2659-2669.	3.7	19
1300	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. <i>Genetics in Medicine</i> , 2021, 23, 2122-2137.	1.1	16
1301	Homozygous missense mutation in UQCRC2 associated with severe encephalomyopathy, mitochondrial complex III assembly defect and activation of mitochondrial protein quality control. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2021, 1867, 166147.	1.8	11
1302	Novel compound heterozygous TTN variants as a cause of severe neonatal congenital contracture syndrome without cardiac involvement diagnosed with rapid trio exome sequencing. <i>Neuromuscular Disorders</i> , 2021, 31, 783-787.	0.3	6
1303	Evaluation of low-pass genome sequencing in polygenic risk score calculation for Parkinson's disease. <i>Human Genomics</i> , 2021, 15, 58.	1.4	4

#	ARTICLE	IF	CITATIONS
1304	Ehlersâ€“Danlos syndrome type IV with a novel COL3A1 exon 14 skipping variation confirmed by Tohoku Medical Megabank Organization genomic database. <i>Journal of Dermatology</i> , 2021, 48, 1918-1922.	0.6	0
1305	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. <i>Human Genetics</i> , 2022, 141, 853-863.	1.8	7
1307	Strategies to Identify Genetic Variants Causing Infertility. <i>Trends in Molecular Medicine</i> , 2021, 27, 792-806.	3.5	9
1308	Towards population-specific pharmacogenomics in the era of next-generation sequencing. <i>Drug Discovery Today</i> , 2021, 26, 1776-1783.	3.2	2
1309	Human TBK1 deficiency leads to autoinflammation driven by TNF-induced cell death. <i>Cell</i> , 2021, 184, 4447-4463.e20.	13.5	64
1310	Exome sequencing in children with clinically suspected <scp>maturityâ€“onset</scp> diabetes of the young. <i>Pediatric Diabetes</i> , 2021, 22, 960-968.	1.2	6
1311	Machine learning-based identification and characterization of 15 novel pathogenic SUOX missense mutations. <i>Molecular Genetics and Metabolism</i> , 2021, 134, 188-194.	0.5	7
1312	Analytical Performance of NGS-Based Molecular Genetic Tests Used in the Diagnostic Workflow of Pheochromocytoma/Paraganglioma. <i>Cancers</i> , 2021, 13, 4219.	1.7	3
1313	Whole genome sequencing demonstrates substantial pathophysiological differences of MYC rearrangements in patients with plasma cell myeloma and B-cell lymphoma. <i>Leukemia and Lymphoma</i> , 2021, , 1-10.	0.6	6
1314	Haploinsufficiency of SF3B2 causes craniofacial microsomia. <i>Nature Communications</i> , 2021, 12, 4680.	5.8	43
1315	A patient with early-onset SMAX3 and a novel variant of ATP7A. <i>Brain and Development</i> , 2022, 44, 63-67.	0.6	1
1316	Understanding the impact of SNPs associated with autism spectrum disorder on biological pathways in the human fetal and adult cortex. <i>Scientific Reports</i> , 2021, 11, 15867.	1.6	15
1317	Structural dynamics bridge the gap between the genetic and functional levels of GPCRs. <i>Current Opinion in Structural Biology</i> , 2021, 69, 150-159.	2.6	6
1318	The value of primary transcripts to the clinical and nonâ€“clinical genomics community: Survey results and roadmap for improvements. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1786.	0.6	5
1320	Cost-Effective Mapping of Genetic Interactions in Mammalian Cells. <i>Frontiers in Genetics</i> , 2021, 12, 703738.	1.1	1
1321	Neuroimaging in Kabuki syndrome and another <scp><i>KMT2D</i></scp>-related disorder. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3770-3783.	0.7	7
1322	The Interferon-Induced Transmembrane Protein 3 -rs12252 Allele May Predict COVID-19 Severity Among Ethnic Minorities. <i>Frontiers in Genetics</i> , 2021, 12, 692254.	1.1	2
1323	Clinical description and mutational profile of a Moroccan series of patients with Rubinstein Taybi syndrome. <i>African Health Sciences</i> , 2021, 21, 960-967.	0.3	3

#	ARTICLE	IF	CITATIONS
1324	Characterization of Reference Materials with an Association for Molecular Pathology Pharmacogenetics Working Group Tier 2 Status: CYP2C9, CYP2C19, VKORC1, CYP2C Cluster Variant, and GGCX. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 952-958.	1.2	9
1325	Curating the gnomAD database: Report of novel variants in the thyroglobulin gene using in silico bioinformatics algorithms. <i>Molecular and Cellular Endocrinology</i> , 2021, 534, 111359.	1.6	7
1326	Frequent LPA KIV-2 Variants Lower Lipoprotein(a) Concentrations and Protect Against Coronary Artery Disease. <i>Journal of the American College of Cardiology</i> , 2021, 78, 437-449.	1.2	34
1327	Subcutaneous panniculitis-like T-cell lymphomas with homozygous inheritance of <i>HAVCR2</i> mutations in Vietnamese pedigrees. <i>Pediatric Blood and Cancer</i> , 2021, 68, e29292.	0.8	2
1328	Urine tumor DNA detection of minimal residual disease in muscle-invasive bladder cancer treated with curative-intent radical cystectomy: A cohort study. <i>PLoS Medicine</i> , 2021, 18, e1003732.	3.9	38
1329	Assigning function to SNPs: Considerations when interpreting genetic variation. <i>Seminars in Cell and Developmental Biology</i> , 2022, 121, 135-142.	2.3	13
1331	FIVEx: an interactive eQTL browser across public datasets. <i>Bioinformatics</i> , 2022, 38, 559-561.	1.8	14
1332	Multidimensional Approach Assessing the Role of Interleukin 1 Beta in Mesial Temporal Lobe Epilepsy. <i>Frontiers in Neurology</i> , 2021, 12, 690847.	1.1	2
1333	Late diagnoses of Dravet syndrome: How many individuals are we missing?. <i>Epilepsia Open</i> , 2021, 6, 770-776.	1.3	9
1334	TAC1 deficiency – a complex system out of balance. <i>Current Opinion in Immunology</i> , 2021, 71, 81-88.	2.4	21
1335	Human SMAD4 Genomic Variants Identified in Individuals with Heritable and Early-Onset Thoracic Aortic Disease. <i>Neurology International</i> , 2021, 11, 132-138.	0.2	1
1336	Assessing acute myeloid leukemia susceptibility in rearrangement-driven patients by <i>DNA</i> breakage at topoisomerase II and CCCTC-binding factor/cohesin binding sites. <i>Genes Chromosomes and Cancer</i> , 2021, 60, 808-821.	1.5	3
1337	RYR1 variant c.38T>G, p.Leu13Arg causes hypersensitivity of the ryanodine receptor-1 and is pathogenic for malignant hyperthermia. <i>British Journal of Anaesthesia</i> , 2021, 127, e63-e65.	1.5	4
1338	Targeted NGS Yields Plentiful Ultra-Rare Variants in Inborn Errors of Immunity Patients. <i>Genes</i> , 2021, 12, 1299.	1.0	8
1340	Mutational bias in spermatogonia impacts the anatomy of regulatory sites in the human genome. <i>Genome Research</i> , 2021, 31, 1994-2007.	2.4	4
1341	Evaluating the performance of a clinical genome sequencing program for diagnosis of rare genetic disease, seen through the lens of craniosynostosis. <i>Genetics in Medicine</i> , 2021, 23, 2360-2368.	1.1	13
1342	The PEMDAC phase 2 study of pembrolizumab and entinostat in patients with metastatic uveal melanoma. <i>Nature Communications</i> , 2021, 12, 5155.	5.8	85
1343	Hereditary sensory and autonomic neuropathy in a family of mixed breed dogs associated with a novel <i>RETREG1</i> variant. <i>Journal of Veterinary Internal Medicine</i> , 2021, 35, 2306-2314.	0.6	8

#	ARTICLE	IF	CITATIONS
1345	The evolution of hematopoietic cells under cancer therapy. <i>Nature Communications</i> , 2021, 12, 4803.	5.8	28
1346	RIC3 variants are not associated with Parkinson's disease in large European, Latin American, or East Asian cohorts. <i>Neurobiology of Aging</i> , 2022, 109, 264-268.	1.5	0
1347	Ankyrin repeats in context with human population variation. <i>PLoS Computational Biology</i> , 2021, 17, e1009335.	1.5	5
1348	Functional characterization of novel variants in <i>SMPD1</i> in Indian patients with acid sphingomyelinase deficiency. <i>Human Mutation</i> , 2021, 42, 1336-1350.	1.1	6
1349	Combined Effect of a Polygenic Risk Score and Rare Genetic Variants on Prostate Cancer Risk. <i>European Urology</i> , 2021, 80, 134-138.	0.9	39
1350	Discovery of structural deletions in breast cancer predisposition genes using whole genome sequencing data from >2000 women of African-ancestry. <i>Human Genetics</i> , 2021, 140, 1449-1457.	1.8	4
1351	Ancestral patterns of recessive dystrophic epidermolysis bullosa mutations in Hispanic populations suggest sephardic ancestry. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3390-3400.	0.7	1
1352	Metabolomics-Based Screening of Inborn Errors of Metabolism: Enhancing Clinical Application with a Robust Computational Pipeline. <i>Metabolites</i> , 2021, 11, 568.	1.3	11
1353	Effects of common mutations in the SARS-CoV-2 Spike RBD and its ligand, the human ACE2 receptor on binding affinity and kinetics. <i>ELife</i> , 2021, 10, .	2.8	267
1354	Development and Validation of StrataNGS, a Multiplex PCR, Semiconductor Sequencing-Based Comprehensive Genomic Profiling Test. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 1515-1533.	1.2	10
1356	c.-61G>A in <i>OVOL2</i> is a Pathogenic 5' UTR Untranslated Region Variant Causing Posterior Polymorphous Corneal Dystrophy 1. <i>Cornea</i> , 2022, 41, 89-94.	0.9	3
1358	Extended Phenotyping and Functional Validation Facilitate Diagnosis of a Complex Patient Harboring Genetic Variants in <i>MCCC1</i> and <i>GNB5</i> Causing Overlapping Phenotypes. <i>Genes</i> , 2021, 12, 1352.	1.0	3
1359	Prioritizing the Role of Major Lipoproteins and Subfractions as Risk Factors for Peripheral Artery Disease. <i>Circulation</i> , 2021, 144, 353-364.	1.6	47
1360	One Gene, Many Facets: Multiple Immune Pathway Dysregulation in <i>SOCS1</i> Haploinsufficiency. <i>Frontiers in Immunology</i> , 2021, 12, 680334.	2.2	11
1362	Utility of Gene Panels for the Diagnosis of Inborn Errors of Metabolism in a Metabolic Reference Center. <i>Genes</i> , 2021, 12, 1262.	1.0	6
1363	Rare variant <i>MX1</i> alleles increase human susceptibility to zoonotic H7N9 influenza virus. <i>Science</i> , 2021, 373, 918-922.	6.0	41
1364	A domain damage index to prioritizing the pathogenicity of missense variants. <i>Human Mutation</i> , 2021, 42, 1503-1517.	1.1	0
1365	Absent from DNA and protein: genomic characterization of nullomers and nullpeptides across functional categories and evolution. <i>Genome Biology</i> , 2021, 22, 245.	3.8	13

#	ARTICLE	IF	CITATIONS
1366	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. Orphanet Journal of Rare Diseases, 2021, 16, 365.	1.2	24
1367	Novel variants in the NARS2 gene as a cause of infantile-onset severe epilepsy leading to fatal refractory status epilepticus: case study and literature review. Neurogenetics, 2021, 22, 359-364.	0.7	13
1369	Genetic Analysis of MPO Variants in Four Psoriasis Subtypes in Patients from Germany. Journal of Investigative Dermatology, 2021, 141, 2079-2083.	0.3	3
1370	A comprehensive analysis of SNPs and CNVs identifies novel markers associated with disease outcomes in colorectal cancer. Molecular Oncology, 2021, 15, 3329-3347.	2.1	9
1371	Hereditary thrombocytosis: the genetic landscape. British Journal of Haematology, 2021, 194, 1098-1105.	1.2	6
1372	Alcohol use and cardiometabolic risk in the UK Biobank: A Mendelian randomization study. PLoS ONE, 2021, 16, e0255801.	1.1	24
1373	MSABrowser: dynamic and fast visualization of sequence alignments, variations and annotations. Bioinformatics Advances, 2021, 1, .	0.9	1
1374	Investigation of LINC00493/SMIM26 Gene Suggests Its Dual Functioning at mRNA and Protein Level. International Journal of Molecular Sciences, 2021, 22, 8477.	1.8	2
1375	Genome-wide association studies. Nature Reviews Methods Primers, 2021, 1, .	11.8	529
1376	Association of rs9939609-FTO with metabolic syndrome components among women from Mayan communities of Chiapas, Mexico. Journal of Physiological Anthropology, 2021, 40, 11.	1.0	2
1377	Bi-allelic loss of function variants in <sc>GOLGA2</sc> are associated with a complex neurological phenotype: Report of a second family. Clinical Genetics, 2021, 100, 748-751.	1.0	8
1378	Phenotypes and genotypes in non-consanguineous and consanguineous primary microcephaly: High incidence of epilepsy. Molecular Genetics & Genomic Medicine, 2021, 9, e1768.	0.6	6
1380	Common Genetic Aberrations Associated with Metabolic Interferences in Human Type-2 Diabetes and Acute Myeloid Leukemia: A Bioinformatics Approach. International Journal of Molecular Sciences, 2021, 22, 9322.	1.8	3
1381	HPDL deficiency causes a neuromuscular disease by impairing the mitochondrial respiration. Journal of Genetics and Genomics, 2021, 48, 727-736.	1.7	5
1382	Population sequencing data reveal a compendium of mutational processes in the human germ line. Science, 2021, 373, 1030-1035.	6.0	43
1383	Cancer Predisposition Sequencing Reporter (<sc>CPSR</sc>): A flexible variant report engine for high-throughput germline screening in cancer. International Journal of Cancer, 2021, 149, 1955-1960.	2.3	12
1384	Mutational patterns and clonal evolution from diagnosis to relapse in pediatric acute lymphoblastic leukemia. Scientific Reports, 2021, 11, 15988.	1.6	6
1385	A novel likely-pathogenic variant in a patient with Hermansky-Pudlak Syndrome. Journal of Physical Education and Sports Management, 2021, 7, mcs.a006110.	0.5	0

#	ARTICLE	IF	CITATIONS
1386	Whole genome sequencing of low input circulating cell-free DNA obtained from normal human subjects. <i>Physiological Reports</i> , 2021, 9, e14993.	0.7	0
1388	A Computational Approach to Evaluate the Combined Effect of SARS-CoV-2 RBD Mutations and ACE2 Receptor Genetic Variants on Infectivity: The COVID-19 Host-Pathogen Nexus. <i>Frontiers in Cellular and Infection Microbiology</i> , 2021, 11, 707194.	1.8	12
1391	Decoding disease: from genomes to networks to phenotypes. <i>Nature Reviews Genetics</i> , 2021, 22, 774-790.	7.7	46
1393	Neuropathological Findings in a Case of Parkinsonism and Developmental Delay Associated with a Monoallelic Variant in <i>PLXNA1</i> . <i>Movement Disorders</i> , 2021, 36, 2681-2687.	2.2	5
1394	Variants of Uncertain Significance: Twins With Identical Pathogenic Gene Mutations in Retinitis Punctata Albescens. <i>Ophthalmic Surgery Lasers and Imaging Retina</i> , 2021, 52, 450-453.	0.4	0
1395	ACO2 clinicobiological dataset with extensive phenotype ontology annotation. <i>Scientific Data</i> , 2021, 8, 205.	2.4	2
1396	Biallelic <i>GINS2</i> variant p.(Arg114Leu) causes Meier-Gorlin syndrome with craniosynostosis. <i>Journal of Medical Genetics</i> , 2022, 59, 776-780.	1.5	10
1397	An approach to gene-based testing accounting for dependence of tests among nearby genes. <i>Briefings in Bioinformatics</i> , 2021, 22, .	3.2	0
1398	Genomic profile – a possible diagnostic and prognostic marker in upper tract urothelial carcinoma. <i>BJU International</i> , 2022, 130, 92-101.	1.3	5
1400	Identification of 22 novel BTK gene variants in B cell deficiency with hypogammaglobulinemia. <i>Clinical Immunology</i> , 2021, 229, 108788.	1.4	2
1404	Targeted long-read sequencing identifies missing disease-causing variation. <i>American Journal of Human Genetics</i> , 2021, 108, 1436-1449.	2.6	105
1405	A Roadmap to Gene Discoveries and Novel Therapies in Monogenic Low and High Bone Mass Disorders. <i>Frontiers in Endocrinology</i> , 2021, 12, 709711.	1.5	13
1407	Linking genome variants to disease: scalable approaches to test the functional impact of human mutations. <i>Human Molecular Genetics</i> , 2021, 30, R187-R197.	1.4	27
1408	Rare variant contribution to human disease in 281,104 UK Biobank exomes. <i>Nature</i> , 2021, 597, 527-532.	13.7	224
1409	Filamin C missense variant associated with severe right atrial disease and skeletal myopathy. <i>Journal of Cardiovascular Electrophysiology</i> , 2021, 32, 2777-2780.	0.8	1
1410	Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation: A Novel DARS2 Mutation and Intra-familial Heterogeneity. <i>Movement Disorders Clinical Practice</i> , 2021, 8, 1116-1122.	0.8	2
1412	GWAS Identifies LINC01184/SLC12A2 as a Risk Locus for Skin and Soft Tissue Infections. <i>Journal of Investigative Dermatology</i> , 2021, 141, 2083-2086.e8.	0.3	4
1413	Examination of the predicted prevalence of Gitelman syndrome by ethnicity based on genome databases. <i>Scientific Reports</i> , 2021, 11, 16099.	1.6	13

#	ARTICLE	IF	CITATIONS
1414	Expanding the potential genes of inborn errors of immunity through protein interactions. <i>BMC Genomics</i> , 2021, 22, 618.	1.2	1
1415	Associations of Genetically Predicted Lp(a) (Lipoprotein [a]) Levels With Cardiovascular Traits in Individuals of European and African Ancestry. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003354.	1.6	21
1416	Cancer associated mutations in Sec61 β alter the permeability of the ER translocase. <i>PLoS Genetics</i> , 2021, 17, e1009780.	1.5	2
1417	The Estimated Prevalence of N-Linked Congenital Disorders of Glycosylation Across Various Populations Based on Allele Frequencies in General Population Databases. <i>Frontiers in Genetics</i> , 2021, 12, 719437.	1.1	9
1418	Protective Effects of Statin Therapy in Cirrhosis Are Limited by a Common SLCO1B1 Transporter Variant. <i>Hepatology Communications</i> , 2021, 5, 1755-1766.	2.0	3
1419	Further Delineation of the Clinical and Pathologic Features of HIKESHI-Related Hypomyelinating Leukodystrophy. <i>Pediatric Neurology</i> , 2021, 121, 11-19.	1.0	2
1421	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. <i>Clinical Epigenetics</i> , 2021, 13, 157.	1.8	22
1422	POSTAR3: an updated platform for exploring post-transcriptional regulation coordinated by RNA-binding proteins. <i>Nucleic Acids Research</i> , 2022, 50, D287-D294.	6.5	65
1423	Gene Panel Testing for Breast Cancer Reveals Differential Effect of Prior BRCA1/2 Probability. <i>Cancers</i> , 2021, 13, 4154.	1.7	5
1424	Evolutionary and structural constraints influencing apolipoprotein A β amyloid behavior. <i>Proteins: Structure, Function and Bioinformatics</i> , 2022, 90, 258-269.	1.5	4
1425	Biallelic SYNE2 Missense Mutations Leading to Nesprin-2 Giant Hypo-Expression Are Associated with Intellectual Disability and Autism. <i>Genes</i> , 2021, 12, 1294.	1.0	6
1426	Pathogenic variations in Germ Cell Nuclear Acidic Peptidase (GCNA) are associated with human male infertility. <i>European Journal of Human Genetics</i> , 2021, 29, 1781-1788.	1.4	10
1427	Nonsense-mediated decay is highly stable across individuals and tissues. <i>American Journal of Human Genetics</i> , 2021, 108, 1401-1408.	2.6	15
1428	<i>ZSWIM7</i> Is Associated With Human Female Meiosis and Familial Primary Ovarian Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, e254-e263.	1.8	13
1429	Cortical organoids model early brain development disrupted by 16p11.2 copy number variants in autism. <i>Molecular Psychiatry</i> , 2021, 26, 7560-7580.	4.1	61
1430	Bcl 2 -negative IGH λ BCL2 translocation-negative follicular lymphoma of the thyroid differs genetically and epigenetically from Bcl 2 -positive IGH λ BCL2 translocation-positive follicular lymphoma. <i>Histopathology</i> , 2021, 79, 521-532.	1.6	3
1431	Role of LRP10 in Parkinson's disease in a Taiwanese cohort. <i>Parkinsonism and Related Disorders</i> , 2021, 89, 79-83.	1.1	3
1432	<i>DIAPH1</i> Variants in Non-East Asian Patients With Sporadic Moyamoya Disease. <i>JAMA Neurology</i> , 2021, 78, 993.	4.5	33

#	ARTICLE	IF	CITATIONS
1433	The mutational landscape of human somatic and germline cells. <i>Nature</i> , 2021, 597, 381-386.	13.7	180
1434	From variant to function in human disease genetics. <i>Science</i> , 2021, 373, 1464-1468.	6.0	75
1435	Proteogenomic characterization of pancreatic ductal adenocarcinoma. <i>Cell</i> , 2021, 184, 5031-5052.e26.	13.5	236
1436	Pseudogene-Mediated Gene Conversion After CRISPR-Cas9 Editing Demonstrated by Partial <i>CD33</i> Conversion with <i>SIGLEC22P</i> . <i>CRISPR Journal</i> , 2021, 4, 699-709.	1.4	3
1437	Prevalence of filaggrin loss-of-function variants in Chilean population with and without atopic dermatitis. <i>International Journal of Dermatology</i> , 2022, 61, 310-315.	0.5	8
1438	Non-cancer-related pathogenic germline variants and expression consequences in ten-thousand cancer genomes. <i>Genome Medicine</i> , 2021, 13, 147.	3.6	4
1439	Cracking the Skin Barrier: Liquid-Liquid Phase Separation Shines under the Skin. <i>JID Innovations</i> , 2021, 1, 100036.	1.2	9
1440	Cloud-based genomics pipelines for ophthalmology: reviewed from research to clinical practice. <i>Modeling and Artificial Intelligence in Ophthalmology</i> , 2021, 3, 101-140.	0.1	1
1441	<i>De novo</i> missense variants in <i>FBXO11</i> alter its protein expression and subcellular localization. <i>Human Molecular Genetics</i> , 2022, 31, 440-454.	1.4	7
1443	Measuring Pharmacogene Variant Function at Scale Using Multiplexed Assays. <i>Annual Review of Pharmacology and Toxicology</i> , 2022, 62, 531-550.	4.2	9
1444	Charcot-Marie-Tooth disease type 2CC due to <i>NEFH</i> variants causes a progressive, non-length-dependent, motor-predominant phenotype. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2022, 93, 48-56.	0.9	9
1445	Mutational analysis of apoptotic genes in familial aggregation of hematological malignancies. <i>Bulletin Du Cancer</i> , 2021, 108, 798-805.	0.6	0
1446	Deciphering complex genome rearrangements in <i>C. elegans</i> using short-read whole genome sequencing. <i>Scientific Reports</i> , 2021, 11, 18258.	1.6	9
1447	Improved pathogenicity prediction for rare human missense variants. <i>American Journal of Human Genetics</i> , 2021, 108, 1891-1906.	2.6	51
1448	<i>RAD50</i> Loss of Function Variants in the Zinc Hook Domain Associated with Higher Risk of Familial Esophageal Squamous Cell Carcinoma. <i>Cancers</i> , 2021, 13, 4715.	1.7	1
1449	Five novel silenced <i>KEL</i> alleles. <i>Transfusion</i> , 2021, 61, E77-E79.	0.8	0
1451	Common and Unique Genetic Background between Attention-Deficit/Hyperactivity Disorder and Excessive Body Weight. <i>Genes</i> , 2021, 12, 1407.	1.0	7
1453	A case-control study of a combination of single nucleotide polymorphisms and clinical parameters to predict clinically relevant toxicity associated with fluoropyrimidine and platinum-based chemotherapy in gastric cancer. <i>BMC Cancer</i> , 2021, 21, 1030.	1.1	6

#	ARTICLE	IF	CITATIONS
1454	A novel de novo KDM5C variant in a female with global developmental delay and ataxia: a case report. <i>BMC Neurology</i> , 2021, 21, 358.	0.8	6
1455	The phenotypic spectrum of KCNT1: a new family with variable epilepsy syndromes including mild focal epilepsy. <i>Journal of Neurology</i> , 2022, 269, 2162-2171.	1.8	5
1456	Annotation of 1350 Common Genetic Variants of the 19 ALDH Multigene Family from Global Human Genome Aggregation Database (gnomAD). <i>Biomolecules</i> , 2021, 11, 1423.	1.8	12
1457	A Human Pleiotropic Multiorgan Condition Caused by Deficient Wnt Secretion. <i>New England Journal of Medicine</i> , 2021, 385, 1292-1301.	13.9	23
1458	Effect of a common UMOD variant on kidney function, blood pressure, cognitive and physical function in a community-based cohort of older adults. <i>Journal of Human Hypertension</i> , 2022, 36, 983-988.	1.0	1
1459	Heterozygous variants in <i>ZBTB7A</i> cause a neurodevelopmental disorder associated with symptomatic overgrowth of pharyngeal lymphoid tissue, macrocephaly, and elevated fetal hemoglobin. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 272-282.	0.7	4
1460	Clustered mutations in the GRIK2 kainate receptor subunit gene underlie diverse neurodevelopmental disorders. <i>American Journal of Human Genetics</i> , 2021, 108, 1692-1709.	2.6	18
1461	Expanding the phenotype of AFG3L2 mutations: Late-onset autosomal recessive spinocerebellar ataxia. <i>Journal of the Neurological Sciences</i> , 2021, 428, 117600.	0.3	5
1462	Biallelic variants of <i>ATP13A3</i> cause dose-dependent childhood-onset pulmonary arterial hypertension characterised by extreme morbidity and mortality. <i>Journal of Medical Genetics</i> , 2022, 59, 906-911.	1.5	22
1464	A novel gain-of-function sodium channel β 2 subunit mutation in idiopathic small fiber neuropathy. <i>Journal of Neurophysiology</i> , 2021, 126, 827-839.	0.9	5
1465	First characterization of congenital myasthenic syndrome type 5 in North Africa. <i>Molecular Biology Reports</i> , 2021, 48, 6999-7006.	1.0	4
1466	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. <i>American Journal of Human Genetics</i> , 2021, 108, 1710-1724.	2.6	18
1467	Comprehensive Genetic Analysis Reveals Complexity of Monogenic Urinary Stone Disease. <i>Kidney International Reports</i> , 2021, 6, 2862-2884.	0.4	15
1468	<i>GLI3</i> variants causing isolated polysyndactyly are not restricted to the protein's C-terminal third. <i>Clinical Genetics</i> , 2021, 100, 758-765.	1.0	4
1469	RNF170 mutation causes autosomal dominant sensory ataxia with variable pyramidal involvement. <i>European Journal of Neurology</i> , 2022, 29, 345-349.	1.7	2
1470	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. <i>Journal of Molecular Medicine</i> , 2021, 99, 1755-1768.	1.7	3
1471	The genome atlas: navigating a new era of reference genomes. <i>Trends in Genetics</i> , 2021, 37, 807-818.	2.9	8
1472	Phenotypic Expression and Outcomes in Individuals With Rare Genetic Variants of Hypertrophic Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2021, 78, 1097-1110.	1.2	55

#	ARTICLE	IF	CITATIONS
1474	Deep clinicopathological phenotyping identifies a previously unrecognized pathogenic <i>EMD</i> splice variant. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 2052-2058.	1.7	1
1475	Adrenal-permissive HSD3B1 genetic inheritance and risk of estrogen-driven postmenopausal breast cancer. <i>JCI Insight</i> , 2021, 6, .	2.3	13
1476	The genomic profiling and MAMLD1 expression in human and canines with Cushing's disease. <i>BMC Endocrine Disorders</i> , 2021, 21, 185.	0.9	1
1477	Clinicoradiographic and genetic features of cerebral small vessel disease indicate variability in mode of inheritance for monoallelic <i>HTRA1</i> variants. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1799.	0.6	4
1478	The prevalence of homologous recombination deficiency (HRD) in various solid tumors and the role of HRD as a single biomarker to immune checkpoint inhibitors. <i>Journal of Cancer Research and Clinical Oncology</i> , 2022, 148, 2427-2435.	1.2	5
1480	The Thai reference exome (T-REx) variant database. <i>Clinical Genetics</i> , 2021, 100, 703-712.	1.0	24
1481	A novel candidate gene in autosomal dominant facial pruritus. <i>Clinical and Experimental Dermatology</i> , 2022, 47, 184-186.	0.6	2
1482	Human RyR2 (Ryanodine Receptor 2) Loss-of-Function Mutations. <i>Circulation: Arrhythmia and Electrophysiology</i> , 2021, 14, e010013.	2.1	18
1483	Identifying dominant-negative actions of a dopamine transporter variant in patients with parkinsonism and neuropsychiatric disease. <i>JCI Insight</i> , 2021, 6, .	2.3	11
1484	Mutational landscape of TRPC6, WT1, LMX1B, APOL1, PTPRO, PMM2, LAMB2 and WT1 genes associated with Steroid resistant nephrotic syndrome. <i>Molecular Biology Reports</i> , 2021, 48, 7193-7201.	1.0	2
1485	PRDM12 Is Transcriptionally Active and Required for Nociceptor Function Throughout Life. <i>Frontiers in Molecular Neuroscience</i> , 2021, 14, 720973.	1.4	7
1487	Genetics of lipodystrophy syndromes. <i>Presse Medicale</i> , 2021, 50, 104074.	0.8	5
1488	Mesenteric cysts, lymphatic leak, and cerebral cavernous malformation in a proband with KRIT1-related disease. <i>American Journal of Medical Genetics, Part A</i> , 2021, .	0.7	0
1489	Mutations at a split codon in the GTPase-encoding domain of <i>OPA1</i> cause dominant optic atrophy through different molecular mechanisms. <i>Human Molecular Genetics</i> , 2022, 31, 761-774.	1.4	6
1491	Human genetic analyses of organelles highlight the nucleus in age-related trait heritability. <i>ELife</i> , 2021, 10, .	2.8	20
1492	Forensic proteomics. <i>Forensic Science International: Genetics</i> , 2021, 54, 102529.	1.6	24
1494	Genomic and evolutionary classification of lung cancer in never smokers. <i>Nature Genetics</i> , 2021, 53, 1348-1359.	9.4	81
1495	An insertion/deletion polymorphism in the <i>KLF1</i> gene resulting in an In(Lu) phenotype. <i>Transfusion</i> , 2021, 61, E73-E74.	0.8	0

#	ARTICLE	IF	CITATIONS
1497	Detection of mobile elements insertions for routine clinical diagnostics in targeted sequencing data. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1807.	0.6	6
1498	Recurrent Pregnancy Loss and Concealed Long QT Syndrome. <i>Journal of the American Heart Association</i> , 2021, 10, e021236.	1.6	3
1499	Mechanisms underlying genetic susceptibility to multisystem inflammatory syndrome in children (MIS-C). <i>Journal of Allergy and Clinical Immunology</i> , 2021, 148, 732-738.e1.	1.5	84
1501	CNV Detection from Exome Sequencing Data in Routine Diagnostics of Rare Genetic Disorders: Opportunities and Limitations. <i>Genes</i> , 2021, 12, 1427.	1.0	21
1502	Filamin C Cardiomyopathy Variants Cause Protein and Lysosome Accumulation. <i>Circulation Research</i> , 2021, 129, 751-766.	2.0	25
1503	Gene Mutations of the Three Ectodysplasin Pathway Key Players (EDA, EDAR, and EDARADD) Account for More than 60% of Egyptian Ectodermal Dysplasia: A Report of Seven Novel Mutations. <i>Genes</i> , 2021, 12, 1389.	1.0	4
1504	Real-World Evaluation of Universal Germline Screening for Cancer Treatment-Relevant Pharmacogenes. <i>Cancers</i> , 2021, 13, 4524.	1.7	6
1505	Deep mutational scanning of the plasminogen activator inhibitor-1 functional landscape. <i>Scientific Reports</i> , 2021, 11, 18827.	1.6	8
1508	Prioritizing de novo autism risk variants with calibrated gene- and variant-scoring models. <i>Human Genetics</i> , 2021, , 1.	1.8	1
1509	A pathogenic DMC1 frameshift mutation causes nonobstructive azoospermia but not primary ovarian insufficiency in humans. <i>Molecular Human Reproduction</i> , 2021, 27, .	1.3	9
1510	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. <i>American Journal of Human Genetics</i> , 2021, 108, 1981-2005.	2.6	38
1511	Genetic landscape of 125 pharmacogenes in Chinese from the Chinese Millionome Database. <i>Scientific Reports</i> , 2021, 11, 19222.	1.6	3
1512	Incessant atrial and ventricular tachycardias associated with an SCN5A mutation. <i>HeartRhythm Case Reports</i> , 2021, 7, 806-811.	0.2	0
1513	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in <i>Drosophila</i> . <i>American Journal of Human Genetics</i> , 2021, 108, 1669-1691.	2.6	23
1514	Biochemically deleterious human <i>NFKB1</i> variants underlie an autosomal dominant form of common variable immunodeficiency. <i>Journal of Experimental Medicine</i> , 2021, 218, .	4.2	32
1515	Association of Monogenic and Polygenic Risk With the Prevalence of Open-Angle Glaucoma. <i>JAMA Ophthalmology</i> , 2021, 139, 1023.	1.4	15
1516	Discovery and implications of polygenicity of common diseases. <i>Science</i> , 2021, 373, 1468-1473.	6.0	80
1517	Protein-coding repeat polymorphisms strongly shape diverse human phenotypes. <i>Science</i> , 2021, 373, 1499-1505.	6.0	96

#	ARTICLE	IF	CITATIONS
1518	A murine model of the human CREBRFR457Q obesity-risk variant does not influence energy or glucose homeostasis in response to nutritional stress. <i>PLoS ONE</i> , 2021, 16, e0251895.	1.1	3
1519	Organic Anion Transporting Polypeptide 2B1 (OATP2B1) Genetic Variants: In Vitro Functional Characterization and Association With Circulating Concentrations of Endogenous Substrates. <i>Frontiers in Pharmacology</i> , 2021, 12, 713567.	1.6	10
1520	Sweat-gland carcinoma with neuroendocrine differentiation (SCAND): a clinicopathologic study of 13 cases with genetic analysis. <i>Modern Pathology</i> , 2022, 35, 33-43.	2.9	10
1521	“Please See this Man with a 69-Year History of Hypoglycaemia” <i>Journal of the Royal College of Physicians of Edinburgh, The</i> , 2021, 51, 266-268.	0.2	2
1523	A cross-population atlas of genetic associations for 220 human phenotypes. <i>Nature Genetics</i> , 2021, 53, 1415-1424.	9.4	560
1524	A novel homozygous variant of the thrombomodulin gene causes a hereditary bleeding disorder. <i>Blood Advances</i> , 2021, 5, 3830-3838.	2.5	6
1525	Developmental and temporal characteristics of clonal sperm mosaicism. <i>Cell</i> , 2021, 184, 4772-4783.e15.	13.5	27
1526	Identifying susceptibility genes for primary ovarian insufficiency on the high-risk genetic background of a fragile X premutation. <i>Fertility and Sterility</i> , 2021, 116, 843-854.	0.5	5
1527	Waardenburg syndrome type 2A in a large Iranian family with a novel MITF gene mutation. <i>BMC Medical Genomics</i> , 2021, 14, 230.	0.7	0
1528	Phenotypic heterogeneity and mosaicism in Xia-Gibbs syndrome: Five Danish patients with novel variants in AHDC1. <i>European Journal of Medical Genetics</i> , 2021, 64, 104280.	0.7	3
1529	Analysis of SARS-CoV-2 nucleocapsid phosphoprotein N variations in the binding site to human 14-3-3 proteins. <i>Biochemical and Biophysical Research Communications</i> , 2021, 569, 154-160.	1.0	11
1530	Precision Medicine Approaches for Infantile-Onset Developmental and Epileptic Encephalopathies. <i>Annual Review of Pharmacology and Toxicology</i> , 2022, 62, 641-662.	4.2	10
1531	The Opportunities and Challenges of Integrating Population Histories Into Genetic Studies for Diverse Populations: A Motivating Example From Native Hawaiians. <i>Frontiers in Genetics</i> , 2021, 12, 643883.	1.1	5
1532	ABHD16A deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. <i>American Journal of Human Genetics</i> , 2021, 108, 2017-2023.	2.6	9
1533	Contribution of rare variant associations to neurodegenerative disease presentation. <i>Npj Genomic Medicine</i> , 2021, 6, 80.	1.7	14
1534	Adapting the ACMG/AMP variant classification framework: A perspective from the ClinGen Hemoglobinopathy Variant Curation Expert Panel. <i>Human Mutation</i> , 2022, 43, 1089-1096.	1.1	20
1535	Noncoding sequence variants define a novel regulatory element in the first intron of the <i>acetylglutamate synthase</i> gene. <i>Human Mutation</i> , 2021, 42, 1624-1636.	1.1	3
1536	Molecular landscape of <i>DYSF</i> mutations in dysferlinopathy: From a Chinese multicenter analysis to a worldwide perspective. <i>Human Mutation</i> , 2021, 42, 1615-1623.	1.1	6

#	ARTICLE	IF	CITATIONS
1537	PIGG defines the Emm blood group system. <i>Scientific Reports</i> , 2021, 11, 18545.	1.6	7
1538	DDX3X and DDX3Y are redundant in protein synthesis. <i>Rna</i> , 2021, 27, 1577-1588.	1.6	38
1539	Mild phenotype of knockouts of the major apurinic/aprimidinic endonuclease APEX1 in a non-cancer human cell line. <i>PLoS ONE</i> , 2021, 16, e0257473.	1.1	4
1541	Schizophrenia, autism spectrum disorders and developmental disorders share specific disruptive coding mutations. <i>Nature Communications</i> , 2021, 12, 5353.	5.8	44
1542	Whole exome sequencing in 17 consanguineous Iranian pedigrees expands the mutational spectrum of inherited retinal dystrophies. <i>Scientific Reports</i> , 2021, 11, 19332.	1.6	2
1543	<i>COL3A1</i> Missense Variant in a Patient Presenting With Hemoptysis. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003386.	1.6	0
1544	Genomic Profiling of Lung Adenocarcinoma in Never-Smokers. <i>Journal of Clinical Oncology</i> , 2021, 39, 3747-3758.	0.8	38
1545	Local adaptation and archaic introgression shape global diversity at human structural variant loci. <i>ELife</i> , 2021, 10, .	2.8	33
1546	A practical guide to genetic testing in endocrinology. <i>Clinical Endocrinology</i> , 2022, 97, 388-399.	1.2	3
1547	Coexistence of Two Rare Genetic Variants in Canonical and Non-canonical Exons of <i>SCN5A</i> : A Potential Source of Misinterpretation. <i>Frontiers in Genetics</i> , 2021, 12, 722291.	1.1	1
1548	Two novel alleles on Fucosyltransferase 2 from northern Thai para-Bombay family and computational prediction on mutation effect. <i>Transfusion</i> , 2021, 61, 3247-3257.	0.8	1
1551	Novel Mutation of the TGF- β 3 Protein (Loeys-Dietz Type 5) Associated With Aortic and Carotid Dissections. <i>Neurology: Genetics</i> , 2021, 7, e625.	0.9	5
1552	Genetic analysis of TRIM family genes for early-onset Parkinson's disease in Chinese population. <i>Parkinsonism and Related Disorders</i> , 2021, 90, 105-113.	1.1	2
1553	Targeted whole exome sequencing and Drosophila modelling to unveil the molecular basis of primary ovarian insufficiency. <i>Human Reproduction</i> , 2021, 36, 2975-2991.	0.4	9
1554	Exome Sequencing of 5 Families with Severe Early-Onset Periodontitis. <i>Journal of Dental Research</i> , 2022, 101, 151-157.	2.5	5
1555	Common host variation drives malaria parasite fitness in healthy human red cells. <i>ELife</i> , 2021, 10, .	2.8	17
1557	Genetic and Clinical Characterization of Patients with Maturity-Onset of Diabetes of the Young (MODY): Identification of Novel Variations. , 2021, 38, 272-277.		4
1558	A report from the European Hyperoxaluria Consortium (OxalEurope) Registry on a large cohort of patients with primary hyperoxaluria type 3. <i>Kidney International</i> , 2021, 100, 621-635.	2.6	26

#	ARTICLE	IF	CITATIONS
1559	Congenital myopathy and epidermolysis bullosa due to PLEC variant. <i>Neuromuscular Disorders</i> , 2021, 31, 1212-1217.	0.3	4
1560	Non-functional alternative splicing caused by a Latino pathogenic variant in a case of PMM2-CDG. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 28, 100781.	0.4	1
1562	Frequency and management of medically actionable incidental findings from genome and exome sequencing data: a systematic review. <i>Physiological Genomics</i> , 2021, 53, 373-384.	1.0	11
1563	Differential genetic diagnoses of adult post-lingual hearing loss according to the audiogram pattern and novel candidate gene evaluation. <i>Human Genetics</i> , 2022, 141, 915-927.	1.8	9
1564	HIF-1 α Pulmonary Phenotype Wide Association Study Unveils a Link to Inflammatory Airway Conditions. <i>Frontiers in Genetics</i> , 2021, 12, 756645.	1.1	6
1565	Whole genome sequence analysis of platelet traits in the NHLBI Trans-Omics for Precision Medicine (TOPMed) initiative. <i>Human Molecular Genetics</i> , 2022, 31, 347-361.	1.4	9
1566	Rare germline variants in individuals diagnosed with schizophrenia within multiplex families. <i>Psychiatry Research</i> , 2021, 303, 114038.	1.7	6
1567	An international virtual hackathon to build tools for the analysis of structural variants within a species ranging from coronaviruses to vertebrates. <i>F1000Research</i> , 2021, 10, 246.	0.8	2
1568	Signet ring cell/histiocytoid carcinoma of the axilla: a clinicopathological and genetic analysis of 11 cases, review of the literature, and comparison with potentially related tumours. <i>Histopathology</i> , 2021, 79, 926-939.	1.6	4
1569	Use of Treatment-Focused Tumor Sequencing to Screen for Germline Cancer Predisposition. <i>Journal of Molecular Diagnostics</i> , 2021, 23, 1145-1158.	1.2	2
1570	BEND4 as a Candidate Gene for an Infection-Induced Acute Encephalopathy Characterized by a Cyst and Calcification of the Pons and Cerebellar Atrophy. <i>Molecular Syndromology</i> , 2022, 13, 12-22.	0.3	0
1571	VannoPortal: multiscale functional annotation of human genetic variants for interrogating molecular mechanism of traits and diseases. <i>Nucleic Acids Research</i> , 2022, 50, D1408-D1416.	6.5	31
1572	Molecular Alterations in Meningioangiomas Causing Epilepsy. <i>Journal of Neuropathology and Experimental Neurology</i> , 2021, 80, 1043-1051.	0.9	3
1573	Mutational patterns and their correlation to CHIP-related mutations and age in hematological malignancies. <i>Blood Advances</i> , 2021, 5, 4426-4434.	2.5	30
1574	Low frequency of treatable pediatric disease alleles in gnomAD: An opportunity for future genomic screening of newborns. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100059.	1.0	3
1575	VIPaHL: Semi-automated ACMG/AMP variant interpretation platform for genetic hearing loss. <i>Human Mutation</i> , 2021, 42, 1567-1575.	1.1	10
1576	Position effects at the FGF8 locus are associated with femoral hypoplasia. <i>American Journal of Human Genetics</i> , 2021, 108, 1725-1734.	2.6	4
1577	Manifestation of epilepsy in a patient with <i>EED</i> -related overgrowth (<i>Cohen-Gibson</i> syndrome). <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 292-297.	0.7	3

#	ARTICLE	IF	CITATIONS
1579	Massively parallel characterization of CYP2C9 variant enzyme activity and abundance. <i>American Journal of Human Genetics</i> , 2021, 108, 1735-1751.	2.6	53
1580	A scalable <i>Drosophila</i> assay for clinical interpretation of human PTEN variants in suppression of PI3K/AKT induced cellular proliferation. <i>PLoS Genetics</i> , 2021, 17, e1009774.	1.5	4
1581	ChÃ©diakâ€“Higashi syndrome presenting as a hereditary spastic paraplegia. <i>Journal of Human Genetics</i> , 2022, 67, 119-121.	1.1	2
1582	Case Report: Two New Cases of Chromosome 12q14 Deletions and Review of the Literature. <i>Frontiers in Genetics</i> , 2021, 12, 716874.	1.1	3
1583	GWAS for genetics of complex quantitative traits: Genome to pangenome and SNPs to SVs and <i>mers</i> . <i>BioEssays</i> , 2021, 43, e2100109.	1.2	15
1584	Low-viscosity matrix suspension culture enables scalable analysis of patient-derived organoids and tumoroids from the large intestine. <i>Communications Biology</i> , 2021, 4, 1067.	2.0	15
1585	Pathogenic variants in nucleoporin <i>TPR</i> (translocated promoter region, nuclear basket protein) cause severe intellectual disability in humans. <i>Human Molecular Genetics</i> , 2022, 31, 362-375.	1.4	6
1586	Distinct functional classes of <i>PDGFRB</i> pathogenic variants in primary familial brain calcification. <i>Human Molecular Genetics</i> , 2022, 31, 399-409.	1.4	8
1588	Molecular genetic evaluation of pediatric renovascular hypertension due to renal artery stenosis and abdominal aortic coarctation in neurofibromatosis type 1. <i>Human Molecular Genetics</i> , 2022, 31, 334-346.	1.4	2
1590	PD-L1P146R is prognostic and a negative predictor of response to immunotherapy in gastric cancer. <i>Molecular Therapy</i> , 2022, 30, 621-631.	3.7	17
1591	SmProt: A Reliable Repository with Comprehensive Annotation of Small Proteins Identified from Ribosome Profiling. <i>Genomics, Proteomics and Bioinformatics</i> , 2021, 19, 602-610.	3.0	28
1592	Biallelic variants in <i>YRDC</i> cause a developmental disorder with progeroid features. <i>Human Genetics</i> , 2021, 140, 1679-1693.	1.8	3
1593	High-throughput imaging of <i>ATG9A</i> distribution as a diagnostic functional assay for adaptor protein complex 4-associated hereditary spastic paraplegia. <i>Brain Communications</i> , 2021, 3, fcab221.	1.5	11
1594	Chromatin and gene-regulatory dynamics of the developing human cerebral cortex at single-cell resolution. <i>Cell</i> , 2021, 184, 5053-5069.e23.	13.5	209
1596	Multi-phenotype genome-wide association studies of the Norfolk Island isolate implicate pleiotropic loci involved in chronic kidney disease. <i>Scientific Reports</i> , 2021, 11, 19425.	1.6	1
1597	Regional <i>TMPRSS2</i> V197M Allele Frequencies Are Correlated with COVID-19 Case Fatality Rates. <i>Molecules and Cells</i> , 2021, 44, 680-687.	1.0	12
1598	<i>AHDC1</i> missense mutations in Xia-Gibbs syndrome. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100049.	1.0	5
1599	DNA variant classificationâ€“reconsidering â€œallele rarityâ€œand â€œphenotypeâ€œcriteria in ACMG/AMP guidelines. <i>European Journal of Medical Genetics</i> , 2021, 64, 104312.	0.7	11

#	ARTICLE	IF	CITATIONS
1600	Common Statin Intolerance Variants in ABCB1 and LILRB5 Show Synergistic Effects on Statin Response: An Observational Study Using Electronic Health Records. <i>Frontiers in Genetics</i> , 2021, 12, 713181.	1.1	12
1601	Rare, Damaging DNA Variants in <i>CORIN</i> and Risk of Coronary Artery Disease: Insights From Functional Genomics and Large-Scale Sequencing Analyses. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003399.	1.6	10
1602	A folding insulator defines cryptic domains in tropomyosin. <i>Journal of Molecular Biology</i> , 2021, 433, 167281.	2.0	0
1603	A clinical, molecular genetics and pathological study of a FTDP-17 family with a heterozygous splicing variant c.823-10G>T at the intron 9/exon 10 of the MAPT gene. <i>Neurobiology of Aging</i> , 2021, 106, 343.e1-343.e8.	1.5	5
1604	Urine organic acid as the first clue towards aromatic L-amino acid decarboxylase (AADC) deficiency in a high prevalence area. <i>Clinica Chimica Acta</i> , 2021, 521, 40-44.	0.5	1
1605	Distinct gene-set burden patterns underlie common generalized and focal epilepsies. <i>EBioMedicine</i> , 2021, 72, 103588.	2.7	7
1606	Molecular and genetic characterization of a large Brazilian cohort presenting hearing loss. <i>Human Genetics</i> , 2022, 141, 519-538.	1.8	6
1607	Evolutionary conservation in noncoding genomic regions. <i>Trends in Genetics</i> , 2021, 37, 903-918.	2.9	13
1608	COQ2 V393A confers high risk susceptibility for multiple system atrophy in East Asian population. <i>Journal of the Neurological Sciences</i> , 2021, 429, 117623.	0.3	17
1610	Extended phenotypes of PIEZO1-related lymphatic dysplasia caused by two novel compound heterozygous variants. <i>European Journal of Medical Genetics</i> , 2021, 64, 104295.	0.7	8
1611	Age-dependent impact of the major common genetic risk factor for COVID-19 on severity and mortality. <i>Journal of Clinical Investigation</i> , 2021, 131, .	3.9	72
1612	Biallelic <i>AOPEP</i> Loss of Function Variants Cause Progressive Dystonia with Prominent Limb Involvement. <i>Movement Disorders</i> , 2022, 37, 137-147.	2.2	14
1613	Long-read technologies identify a hidden inverted duplication in a family with choroideremia. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100046.	1.0	4
1614	Potential role of <i>CMPK1</i> , <i>SLC29A1</i> , and <i>TLE4</i> polymorphisms in gemcitabine-based chemotherapy in HER2-negative metastatic breast cancer patients: pharmacogenetic study results from the prospective randomized phase II study of eribulin plus gemcitabine versus paclitaxel plus gemcitabine (KCSG-BR-13-11). <i>ESMO Open</i> , 2021, 6, 100236.	2.0	0
1615	Impaired Cx43 gap junction endocytosis causes morphological and functional defects in zebrafish. <i>Molecular Biology of the Cell</i> , 2021, 32, ar13.	0.9	6
1616	The ethnogeographic variability of genetic factors underlying G6PD deficiency. <i>Pharmacological Research</i> , 2021, 173, 105904.	3.1	14
1617	Study of rare genetic variants in <i>TM4SF20</i> , <i>NFXL1</i> , <i>CNTNAP2</i> , and <i>ATP2C2</i> in Pakistani probands and families with language impairment. <i>Meta Gene</i> , 2021, 30, 100966.	0.3	6
1618	CABE-RY: A PAM-flexible dual-mutation base editor for reliable modeling of multi-nucleotide variants. <i>Molecular Therapy - Nucleic Acids</i> , 2021, 26, 114-121.	2.3	8

#	ARTICLE	IF	CITATIONS
1619	OUP accepted manuscript. Human Molecular Genetics, 2021, , .	1.4	1
1620	Genetic and Epigenetic Regulation of Organic Cation Transporters. Handbook of Experimental Pharmacology, 2021, 266, 81-100.	0.9	8
1622	Novel exon-skipping variant disrupting the basic domain of HCFC1 causes intellectual disability without metabolic abnormalities in both male and female patients. Journal of Human Genetics, 2021, 66, 717-724.	1.1	10
1623	Differential intolerance to loss of function and missense mutations in genes that encode human matricellular proteins. Journal of Cell Communication and Signaling, 2021, 15, 93-105.	1.8	2
1624	Statistical approaches to rare disease analyses. , 2021, , 205-213.		1
1625	OUP accepted manuscript. Human Molecular Genetics, 2021, 30, 2068-2081.	1.4	7
1626	æ”æ—çš,,ã,ªãfÿã,¬ã,¹è§£æžã«ã,^ã,«ç—¾æ,£ç—...æ...«è§£æ~žã”ã,²ãfžãfã€ã”ãCE—ãCE»ç™, Journal of JCS Cardiologists, 2021, 30, 20-28		
1627	CNV Analysis of the Correlation between Preoperative Lymph Node Metastasis and Prognosis of Early Tongue Cancer. Journal of Cancer, 2021, 12, 6135-6144.	1.2	1
1628	Update of genetic variants in <i>CEP120</i> and <i>CC2D2A</i>”With an emphasis on genotype-phenotype correlations, tissue specific transcripts and exploring mutation specific exon skipping therapies. Molecular Genetics & Genomic Medicine, 2021, 9, e1603.	0.6	8
1629	A likelihood ratio approach for identifying three-quarter siblings in genetic databases. Heredity, 2021, 126, 537-547.	1.2	5
1630	Targeted High-Throughput Sequencing Analysis Results of Osteogenesis Imperfecta Patients from Different Regions of Turkey. Genetic Testing and Molecular Biomarkers, 2021, 25, 59-67.	0.3	2
1631	Tractor uses local ancestry to enable the inclusion of admixed individuals in GWAS and to boost power. Nature Genetics, 2021, 53, 195-204.	9.4	125
1632	<i>Sarm1</i> Haploinsufficiency and Low Expression Levels after Antisense Oligonucleotides Delays Programmed Axon Degeneration. SSRN Electronic Journal, 0, , .	0.4	2
1633	HIF1A: A Putative Modifier of Hemochromatosis. International Journal of Molecular Sciences, 2021, 22, 1245.	1.8	5
1634	Somatic data usage for classification of germ line variants. , 2021, , 169-192.		0
1635	Rare cause of xanthinuria: a pediatric case of molybdenum cofactor deficiency B. CEN Case Reports, 2021, 10, 378-382.	0.5	7
1636	Clinically relevant variants in a large cohort of Indian patients with Marfan syndrome and related disorders identified by next-generation sequencing. Scientific Reports, 2021, 11, 764.	1.6	7
1637	DALIA- a comprehensive resource of Disease Alleles in Arab population. PLoS ONE, 2021, 16, e0244567.	1.1	3

#	ARTICLE	IF	CITATIONS
1638	Protective Effects of <i>APOE</i> ϵ 2 Genotype on Cognition in Older Breast Cancer Survivors: The Thinking and Living With Cancer Study. <i>JNCI Cancer Spectrum</i> , 2021, 5, pkab013.	1.4	6
1639	Two Missense CACNA1A Variants in a Single Family with Variable Neurobehavioral, Cerebellar, Epileptic, and Oculomotor Features. <i>Neuropediatrics</i> , 2021, 52, 186-191.	0.3	4
1640	Spectrum of Germline Mutations Within Fanconi Anemia-Associated Genes Across Populations of Varying Ancestry. <i>JNCI Cancer Spectrum</i> , 2021, 5, .	1.4	3
1642	Sleep phenotype of individuals with autism spectrum disorder bearing mutations in the <i>PER2</i> circadian rhythm gene. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1120-1130.	0.7	17
1643	The fructose-1,6-bisphosphatase deficiency and the p.(Lys204ArgfsTer72) variant. <i>Genetics and Molecular Biology</i> , 2021, 44, e20200281.	0.6	2
1644	Genomic sequencing of rare diseases. , 2021, , 61-95.		6
1645	Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. <i>Science Advances</i> , 2021, 7, .	4.7	25
1647	The contribution of X-linked coding variation to severe developmental disorders. <i>Nature Communications</i> , 2021, 12, 627.	5.8	33
1648	Integrative modeling of transmitted and <i>de novo</i> variants identifies novel risk genes for congenital heart disease. <i>Quantitative Biology</i> , 2021, 9, 216-227.	0.3	4
1649	Discovering Genotype Variants in an Infant with VACTERL through Clinical Exome Sequencing: A Support for Personalized Risk Assessment and Disease Prevention. <i>Pediatric Reports</i> , 2021, 13, 45-56.	0.5	3
1650	A population scale analysis of rare SNCA variation in the UK Biobank. <i>Neurobiology of Disease</i> , 2021, 148, 105182.	2.1	5
1651	Investigating rare pathogenic/likely pathogenic exonic variation in bipolar disorder. <i>Molecular Psychiatry</i> , 2021, 26, 5239-5250.	4.1	15
1652	Mammalian cell proliferation requires noncatalytic functions of O-GlcNAc transferase. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	48
1653	Alagille syndrome and risk for hepatocellular carcinoma: Need for increased surveillance in adults with mild liver phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 719-731.	0.7	12
1654	Biallelic loss of <i>OTUD7A</i> causes severe muscular hypotonia, intellectual disability, and seizures. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1182-1186.	0.7	14
1655	<i>PD-1</i> blockade using pembrolizumab in adolescent and young adult patients with advanced bone and soft tissue sarcoma. <i>Cancer Reports</i> , 2021, 4, e1327.	0.6	8
1656	Bi-allelic Pathogenic Variants in HS2ST1 Cause a Syndrome Characterized by Developmental Delay and Corpus Callosum, Skeletal, and Renal Abnormalities. <i>American Journal of Human Genetics</i> , 2020, 107, 1044-1061.	2.6	11
1657	De Novo VPS4A Mutations Cause Multisystem Disease with Abnormal Neurodevelopment. <i>American Journal of Human Genetics</i> , 2020, 107, 1129-1148.	2.6	38

#	ARTICLE	IF	CITATIONS
1658	A Global Map of G Protein Signaling Regulation by RGS Proteins. <i>Cell</i> , 2020, 183, 503-521.e19.	13.5	82
1659	Comparison of Pathogenicity Prediction Tools on Somatic Variants. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 1383-1392.	1.2	19
1660	Thousands of human sequences provide deep insight into single genomes. <i>Nature</i> , 2020, 581, 385-386.	13.7	1
1661	Deubiquitylases in developmental ubiquitin signaling and congenital diseases. <i>Cell Death and Differentiation</i> , 2021, 28, 538-556.	5.0	27
1662	Genetic mechanisms of critical illness in COVID-19. <i>Nature</i> , 2021, 591, 92-98.	13.7	1,014
1663	The V369M Gcgr knock-in mice are a precision medicine model of mild Mahvash disease. <i>Biochemical Journal</i> , 2020, 477, 2873-2874.	1.7	3
1664	Cross-subunit interactions that stabilize open states mediate gating in NMDA receptors. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	10
1665	The impact of structural bioinformatics tools and resources on SARS-CoV-2 research and therapeutic strategies. <i>Briefings in Bioinformatics</i> , 2021, 22, 742-768.	3.2	29
1666	Accurate, scalable cohort variant calls using DeepVariant and GLnexus. <i>Bioinformatics</i> , 2021, 36, 5582-5589.	1.8	86
1667	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. <i>Brain</i> , 2020, 143, 3589-3602.	3.7	39
1668	Genomic analysis of patients in a South Indian Community with autosomal dominant cortical tremor, myoclonus and epilepsy suggests a founder repeat expansion mutation in the SAMD12 gene. <i>Brain Communications</i> , 2021, 3, fcaa214.	1.5	7
1669	NKCC1: Newly Found as a Human Disease-Causing Ion Transporter. <i>Function</i> , 2020, 2, zqaa028.	1.1	29
1670	<i>De Novo</i> variants in <i>EEF2</i> cause a neurodevelopmental disorder with benign external hydrocephalus. <i>Human Molecular Genetics</i> , 2021, 29, 3892-3899.	1.4	11
1671	Germline Cancer Predisposition Variants inâ€“, Pediatric Rhabdomyosarcoma: A Report From the Childrenâ€™s Oncology Group. <i>Journal of the National Cancer Institute</i> , 2021, 113, 875-883.	3.0	55
1672	jMorp updates in 2020: large enhancement of multi-omics data resources on the general Japanese population. <i>Nucleic Acids Research</i> , 2021, 49, D536-D544.	6.5	107
1673	AI-Driver: an ensemble method for identifying driver mutations in personal cancer genomes. <i>NAR Genomics and Bioinformatics</i> , 2020, 2, lqaa084.	1.5	19
1674	Cholestasis Due to USP53 Deficiency. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2021, 72, 667-673.	0.9	18
1736	Implementing cell-free DNA of pancreatic cancer patientâ€™ derived organoids for personalized oncology. <i>JCI Insight</i> , 2020, 5, .	2.3	30

#	ARTICLE	IF	CITATIONS
1737	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. <i>Journal of Clinical Investigation</i> , 2020, 130, 4423-4439.	3.9	43
1738	Human NK cell deficiency as a result of biallelic mutations in MCM10. <i>Journal of Clinical Investigation</i> , 2020, 130, 5272-5286.	3.9	44
1739	YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress. <i>Journal of Clinical Investigation</i> , 2020, 130, 6338-6353.	3.9	58
1740	Genetic variation in the Middle East – an opportunity to advance the human genetics field. <i>Genome Medicine</i> , 2020, 12, 116.	3.6	27
1741	Multiple Endocrine Tumors Associated with Germline <i>MAX</i> Mutations: Multiple Endocrine Neoplasia Type 5?. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e1163-e1182.	1.8	43
1742	<i>EIF2AK2</i> -related Neurodevelopmental Disorder With Leukoencephalopathy, Developmental Delay, and Episodic Neurologic Regression Mimics Pelizaeus-Merzbacher Disease. <i>Neurology: Genetics</i> , 2021, 7, e539.	0.9	9
1743	Identification of <i>MYOM2</i> as a candidate gene in hypertrophic cardiomyopathy and tetralogy of fallot and its functional evaluation in the <i>Drosophila</i> heart. <i>DMM Disease Models and Mechanisms</i> , 2020, 13, .	1.2	16
1744	Characterization of circulating breast cancer cells with tumorigenic and metastatic capacity. <i>EMBO Molecular Medicine</i> , 2020, 12, e11908.	3.3	77
1745	<i>ALX</i> -related frontonasal dysplasia results from defective neural crest cell development and migration. <i>EMBO Molecular Medicine</i> , 2020, 12, e12013.	3.3	15
1746	The mutational landscape of the <i>SCAN</i> – real-world primary breast cancer transcriptome. <i>EMBO Molecular Medicine</i> , 2020, 12, e12118.	3.3	36
1747	Identification of a <i>TMEM127</i> variant in a patient with paraganglioma and acromegaly. <i>Endocrinology, Diabetes and Metabolism Case Reports</i> , 2020, 2020, .	0.2	4
1749	Whole-Exome Sequencing Identified Novel <i>CLMP</i> Mutations in a Family With Congenital Short Bowel Syndrome Presenting Differently in Two Proband. <i>Frontiers in Genetics</i> , 2020, 11, 574943.	1.1	4
1750	Breast Cancer Organoids Model Patient-Specific Response to Drug Treatment. <i>Cancers</i> , 2020, 12, 3869.	1.7	43
1751	Pharmacogenomics at the Point of Care: A Community Pharmacy Project in British Columbia. <i>Journal of Personalized Medicine</i> , 2021, 11, 11.	1.1	14
1752	Genotype-phenotype analysis in Mowat-Wilson syndrome associated with two novel and two recurrent <i>ZEB2</i> variants. <i>Experimental and Therapeutic Medicine</i> , 2020, 20, 1-1.	0.8	4
1753	<i>KDM5A</i> mutations identified in autism spectrum disorder using forward genetics. <i>ELife</i> , 2020, 9, .	2.8	27
1754	The cryo-EM structure of the human uromodulin filament core reveals a unique assembly mechanism. <i>ELife</i> , 2020, 9, .	2.8	26
1755	<i>SON</i> and <i>SRRM2</i> are essential for nuclear speckle formation. <i>ELife</i> , 2020, 9, .	2.8	122

#	ARTICLE	IF	CITATIONS
1756	The Impact of Variants at Branchpoint Splicing Elements in Cancer Genes. SSRN Electronic Journal, 0, , .	0.4	0
1757	The UCSC Genome Browser database: 2022 update. Nucleic Acids Research, 2022, 50, D1115-D1122.	6.5	175
1758	Bi-allelic variants in DNA mismatch repair proteins MutS Homolog <i>MSH4</i> and <i>MSH5</i> cause infertility in both sexes. Human Reproduction, 2021, 37, 178-189.	0.4	18
1759	CircleBase: an integrated resource and analysis platform for human eccDNAs. Nucleic Acids Research, 2022, 50, D72-D82.	6.5	20
1760	ConVarT: a search engine for matching human genetic variants with variants from non-human species. Nucleic Acids Research, 2022, 50, D1172-D1178.	6.5	16
1761	The TKFC Ala185Thr Variant, Reported as "Null" for Fructose Metabolism, is Fully Active as Triokinase. SSRN Electronic Journal, 0, , .	0.4	0
1762	Different Fumarate Hydratase Gene Variants Are Associated With Distinct Cancer Phenotypes. JCO Precision Oncology, 2021, 5, 1568-1578.	1.5	2
1763	Dominant Optic Atrophy: How to Determine the Pathogenicity of Novel Variants?. Journal of Neuro-Ophthalmology, 2022, 42, 149-153.	0.4	0
1764	Clinico-genetic findings in 509 frontotemporal dementia patients. Molecular Psychiatry, 2021, 26, 5824-5832.	4.1	23
1765	A cis-acting structural variation at the ZNF558 locus controls a gene regulatory network in human brain development. Cell Stem Cell, 2022, 29, 52-69.e8.	5.2	37
1766	Pathogenic BCL11A variants provide insights into the mechanisms of human fetal hemoglobin silencing. PLoS Genetics, 2021, 17, e1009835.	1.5	10
1767	<i>DLG2</i> Mutations in the Etiology of Pubertal Delay and Idiopathic Hypogonadotropic Hypogonadism. Hormone Research in Paediatrics, 2021, 94, 364-368.	0.8	2
1768	Biallelic <i>FRA10AC1</i> variants cause a neurodevelopmental disorder with growth retardation. Brain, 2022, 145, 1551-1563.	3.7	9
1769	Therapeutically expanded human regulatory T-cells are super-suppressive due to HIF1A induced expression of CD73. Communications Biology, 2021, 4, 1186.	2.0	19
1770	A Novel Variant of <sc><i>ATP5MC3</i></sc> Associated with Both Dystonia and Spastic Paraplegia. Movement Disorders, 2022, 37, 375-383.	2.2	10
1772	In Silico Analysis of a De Novo OTC Variant as a Cause of Ornithine Transcarbamylase Deficiency. Applied Immunohistochemistry and Molecular Morphology, 2021, Publish Ahead of Print, .	0.6	0
1773	Rare variant association study of veteran twin whole-genomes links severe depression with a nonsynonymous change in the neuronal gene <i>BHLHE22</i>. World Journal of Biological Psychiatry, 2022, 23, 295-306.	1.3	1
1776	Absence of the RNF213 p.R4810K variant may indicate a severe form of pediatric moyamoya disease in Japanese patients. Journal of Neurosurgery: Pediatrics, 2022, 29, 48-56.	0.8	10

#	ARTICLE	IF	CITATIONS
1777	Loss of Function Glucose-Dependent Insulinotropic Polypeptide Receptor Variants Are Associated With Alterations in BMI, Bone Strength and Cardiovascular Outcomes. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 749607.	1.8	12
1778	A massive effort links protein-coding gene variants to health. <i>Nature</i> , 2021, 599, 561-563.	13.7	1
1782	Novel disease-causing variants and phenotypic features of X-linked megalocornea. <i>Acta Ophthalmologica</i> , 2021, , .	0.6	1
1783	Maternal mosaicism for a missense variant in the <i>SMS</i> gene that causes Snyder-Robinson syndrome. <i>Journal of Physical Education and Sports Management</i> , 2021, 7, a006122.	0.5	0
1785	Dibifunctional protein deficiency caused by splicing variants in a neonate with severe peroxisomal dysfunction and persistent hypoglycemia. <i>American Journal of Medical Genetics, Part A</i> , 2021, , .	0.7	2
1786	Homozygous <i>MEFV</i> Gene Variant and Pyrin-Associated Autoinflammation With Neutrophilic Dermatitis. <i>JAMA Dermatology</i> , 2021, 157, 1466.	2.0	10
1787	Profile of esophageal squamous cell carcinoma mutations in Brazilian patients. <i>Scientific Reports</i> , 2021, 11, 20596.	1.6	2
1788	Privacy-preserving storage of sequenced genomic data. <i>BMC Genomics</i> , 2021, 22, 712.	1.2	2
1791	Rare Variants in RPPH1 qPCR Control Assay Binding Sites Result in Incorrect Copy Number Calls. <i>Journal of Molecular Diagnostics</i> , 2021, , .	1.2	3
1792	Beyond GWAS: from simple associations to functional insights. <i>Seminars in Immunopathology</i> , 2022, 44, 3-14.	2.8	13
1795	Further delineation of the clinical spectrum of White-Sutton syndrome: 12 new individuals and a review of the literature. <i>European Journal of Human Genetics</i> , 2022, 30, 95-100.	1.4	8
1797	Comprehensive landscape and interference of clonal haematopoiesis mutations for liquid biopsy: A Chinese pan-cancer cohort. <i>Journal of Cellular and Molecular Medicine</i> , 2021, 25, 10279-10290.	1.6	4
1799	Detecting cryptic clinically relevant structural variation in exome-sequencing data increases diagnostic yield for developmental disorders. <i>American Journal of Human Genetics</i> , 2021, 108, 2186-2194.	2.6	12
1800	Combinatorial patterns of gene expression changes contribute to variable expressivity of the developmental delay-associated 16p12.1 deletion. <i>Genome Medicine</i> , 2021, 13, 163.	3.6	5
1801	The dihydropyrimidine dehydrogenase gene contributes to heritable differences in sleep in mice. <i>Current Biology</i> , 2021, 31, 5238-5248.e7.	1.8	5
1802	Calibrated rare variant genetic risk scores for complex disease prediction using large exome sequence repositories. <i>Nature Communications</i> , 2021, 12, 5852.	5.8	19
1803	Persistent sodium currents in <i>SCN1A</i> developmental and degenerative epileptic dyskinetic encephalopathy. <i>Brain Communications</i> , 2021, 3, fcab235.	1.5	12
1805	Targeted treatment of immune thrombocytopenia in CTLA4 insufficiency: a case report. <i>British Journal of Haematology</i> , 2021, , .	1.2	0

#	ARTICLE	IF	CITATIONS
1806	Clinical Characteristics and Mutation Spectrum of Neurofibromatosis Type 1 in 27 Turkish Families. , 2021, 38, 365-373.		4
1808	Association of Inherited Mutations in DNA Repair Genes with Localized Prostate Cancer. European Urology, 2022, 81, 559-567.	0.9	17
1809	TIGER: The gene expression regulatory variation landscape of human pancreatic islets. Cell Reports, 2021, 37, 109807.	2.9	45
1811	The Effect of Single Nucleotide Variations in the Transmembrane Domain of OATP1B1 on in vitro Functionality. Pharmaceutical Research, 2021, 38, 1663-1675.	1.7	5
1812	Somatic Mosaicism in Biology and Disease. Annual Review of Physiology, 2022, 84, 113-133.	5.6	5
1813	Clinical and genetic characterization of adult-onset leukoencephalopathy caused by <i>CSF1R</i> mutations. Annals of Clinical and Translational Neurology, 2021, 8, 2121-2131.	1.7	9
1814	Active site variants in STT3A cause a dominant type I congenital disorder of glycosylation with neuromusculoskeletal findings. American Journal of Human Genetics, 2021, 108, 2130-2144.	2.6	5
1815	Semaphorin-Plexin Signaling: From Axonal Guidance to a New X-Linked Intellectual Disability Syndrome. Pediatric Neurology, 2022, 126, 65-73.	1.0	8
1816	Association of Genetic Variants With Outcomes in Patients With Nonischemic Dilated Cardiomyopathy. Journal of the American College of Cardiology, 2021, 78, 1682-1699.	1.2	55
1817	Comprehensive multi-omics integration identifies differentially active enhancers during human brain development with clinical relevance. Genome Medicine, 2021, 13, 162.	3.6	9
1818	Sequence complementarity between human noncoding RNAs and SARS-CoV-2 genes: What are the implications for human health?. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2022, 1868, 166291.	1.8	5
1819	Analysis of whole exome sequencing in severe mental illness hints at selection of brain development and immune related genes. Scientific Reports, 2021, 11, 21088.	1.6	1
1820	Dystonia-specific mutations in THAP1 alter transcription of genes associated with neurodevelopment and myelin. American Journal of Human Genetics, 2021, 108, 2145-2158.	2.6	13
1822	Which Is the Best In Silico Program for the Missense Variations in IDUA Gene? A Comparison of 33 Programs Plus a Conservation Score and Evaluation of 586 Missense Variants. Frontiers in Molecular Biosciences, 2021, 8, 752797.	1.6	5
1823	Wilson Disease Prevalence. Journal of Pediatric Gastroenterology and Nutrition, 2022, 74, 192-199.	0.9	7
1825	Targeted Sequencing of 242 Clinically Important Genes in the Russian Population From the Ivanovo Region. Frontiers in Genetics, 2021, 12, 709419.	1.1	19
1827	Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. Genome Medicine, 2021, 13, 153.	3.6	53
1828	Thrombomodulin in patients with mild to moderate bleeding tendency. Haemophilia, 2021, 27, 1028-1036.	1.0	8

#	ARTICLE	IF	CITATIONS
1830	Integrating thousands of PTEN variant activity and abundance measurements reveals variant subgroups and new dominant negatives in cancers. <i>Genome Medicine</i> , 2021, 13, 165.	3.6	14
1831	Distinction of lymphoid and myeloid clonal hematopoiesis. <i>Nature Medicine</i> , 2021, 27, 1921-1927.	15.2	130
1832	Progress towards completing the mutant mouse null resource. <i>Mammalian Genome</i> , 2022, 33, 123-134.	1.0	15
1835	Familial cleft tongue caused by a unique translation initiation codon variant in TP63. <i>European Journal of Human Genetics</i> , 2021, , .	1.4	7
1838	Oncomineâ„¢ Comprehensive Assay v3 vs. Oncomineâ„¢ Comprehensive Assay Plus. <i>Cancers</i> , 2021, 13, 5230.	1.7	19
1839	Endâ€Truncated <sc>LAMB1</sc> Causes a Hippocampal Memory Defect and a Leukoencephalopathy. <i>Annals of Neurology</i> , 2021, 90, 962-975.	2.8	5
1840	Proteotranscriptomic classification and characterization of pancreatic neuroendocrine neoplasms. <i>Cell Reports</i> , 2021, 37, 109817.	2.9	14
1841	NuMorph: Tools for cortical cellular phenotyping in tissue-cleared whole-brain images. <i>Cell Reports</i> , 2021, 37, 109802.	2.9	8
1842	Exome sequencing and analysis of 454,787 UK Biobank participants. <i>Nature</i> , 2021, 599, 628-634.	13.7	377
1843	Polygenic basis and biomedical consequences of telomere length variation. <i>Nature Genetics</i> , 2021, 53, 1425-1433.	9.4	145
1844	Case Report: Novel Compound-Heterozygous Variants of SKIV2L Gene that Cause Trichohepatoenteric Syndrome 2. <i>Frontiers in Genetics</i> , 2021, 12, 756451.	1.1	2
1846	Genomic sequencing to inform therapy in advanced pancreatic cancer: A systematic review and meta-analysis of prospective studies. <i>Cancer Treatment Reviews</i> , 2021, 101, 102310.	3.4	2
1847	Leveraging health systems data to characterize a large effect variant conferring risk for liver disease in Puerto Ricans. <i>American Journal of Human Genetics</i> , 2021, 108, 2099-2111.	2.6	4
1849	Congenital adrenal hyperplasia caused by compound heterozygosity of two novel CYP11B1 gene variants. <i>Hormones</i> , 2022, 21, 155-161.	0.9	2
1850	Identification of novel missense mutation in a patient with an asymptomatic para-aortic paraganglioma. <i>BMJ Case Reports</i> , 2021, 14, e245427.	0.2	1
1851	Variable clinical severity in <sc>TANGO2</sc> deficiency: Case series and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 473-487.	0.7	17
1854	Toward a methodology for evaluating DNA variants in nuclear families. <i>PLoS ONE</i> , 2021, 16, e0258375.	1.1	0
1855	A p.Arg127Gln variant in GPIIb/3 LRR5 allosterically enhances affinity for VWF: a novel form of platelet-type VWD. <i>Blood Advances</i> , 2021, , .	2.5	4

#	ARTICLE	IF	CITATIONS
1857	WDR37 syndrome: identification of a distinct new cluster of disease-associated variants and functional analyses of mutant proteins. <i>Human Genetics</i> , 2021, 140, 1775-1789.	1.8	4
1861	Current Tools, Databases, and Resources for Phenotype and Variant Analysis of Clinical Exome Sequencing. <i>Advances in Molecular Pathology</i> , 2021, 4, 1-15.	0.2	0
1862	Profound inhibition of CD73-dependent formation of anti-inflammatory adenosine in B cells of SLE patients. <i>EBioMedicine</i> , 2021, 73, 103616.	2.7	14
1863	First report: Rare RNF213 variant associated with familial moyamoya disease in an African American family. <i>Journal of Stroke and Cerebrovascular Diseases</i> , 2021, 30, 106123.	0.7	0
1864	Maximizing insights from monogenic immune disorders. <i>Current Opinion in Immunology</i> , 2021, 73, 50-57.	2.4	2
1865	Ancestry may confound genetic machine learning: Candidate-gene prediction of opioid use disorder as an example. <i>Drug and Alcohol Dependence</i> , 2021, 229, 109115.	1.6	6
1885	A comprehensive analysis of RHOA mutation positive and negative angioimmunoblastic T-cell lymphomas by targeted deep sequencing, expression profiling and single cell digital image analysis. <i>International Journal of Molecular Medicine</i> , 2020, 46, 1466-1476.	1.8	9
1902	Detection of PKD1 and PKD2 Somatic Variants in Autosomal Dominant Polycystic Kidney Cyst Epithelial Cells by Whole-Genome Sequencing. <i>Journal of the American Society of Nephrology: JASN</i> , 2021, 32, 3114-3129.	3.0	13
1903	Massive parallel sequencing in individuals with multiple primary tumours reveals the benefit of re-analysis. <i>Hereditary Cancer in Clinical Practice</i> , 2021, 19, 46.	0.6	3
1905	Causal and Candidate Gene Variants in a Large Cohort of Women With Primary Ovarian Insufficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 685-714.	1.8	13
1906	Bi-allelic variants in the mitochondrial RNase P subunit PRORP cause mitochondrial tRNA processing defects and pleiotropic multisystem presentations. <i>American Journal of Human Genetics</i> , 2021, 108, 2195-2204.	2.6	26
1909	An open approach to systematically prioritize causal variants and genes at all published human GWAS trait-associated loci. <i>Nature Genetics</i> , 2021, 53, 1527-1533.	9.4	208
1911	Integrative assessment of CIP2A overexpression and mutational effects in human malignancies identifies possible deleterious variants. <i>Computers in Biology and Medicine</i> , 2021, 139, 104986.	3.9	6
1912	Somatic variation as an incidental finding in the pediatric next-generation sequencing era. <i>Journal of Physical Education and Sports Management</i> , 2021, 7, a006135.	0.5	3
1913	Pathogenic Variants in Selenoproteins and Selenocysteine Biosynthesis Machinery. <i>International Journal of Molecular Sciences</i> , 2021, 22, 11593.	1.8	10
1915	Exome-Wide Pan-Cancer Analysis of Germline Variants in 8,719 Individuals Finds Little Evidence of Rare Variant Associations. <i>Human Heredity</i> , 2021, 86, 34-44.	0.4	1
1916	Autosomal recessive SLC30A9 variants in a Proband with a Cerebro-Renal Syndrome and No Parental Consanguinity. <i>Journal of Physical Education and Sports Management</i> , 2021, , mcs.a006137.	0.5	4
1917	<sc><i>PUS3</i></sc>-related disorder: Report of a novel patient and delineation of the phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 635-641.	0.7	5

#	ARTICLE	IF	CITATIONS
1918	Disease variant prediction with deep generative models of evolutionary data. <i>Nature</i> , 2021, 599, 91-95.	13.7	306
1921	Monoallelic loss of <i>YTHDF3</i> and neurodevelopmental disorder: clinical features of four individuals with 8q12.3 deletions. <i>Clinical Genetics</i> , 2022, 101, 208-213.	1.0	2
1922	Breast cancer in West Africa: molecular analysis of BRCA genes in early-onset breast cancer patients in Burkina Faso. <i>Human Genomics</i> , 2021, 15, 65.	1.4	9
1923	Fumarate hydratase variant prevalence and manifestations among individuals receiving germline testing. <i>Cancer</i> , 2022, 128, 675-684.	2.0	11
1924	Analysis of 272 Genetic Variants in the Upgraded Interactive FXI Web Database Reveals New Insights into FXI Deficiency. <i>TH Open</i> , 2021, 05, e543-e556.	0.7	8
1926	Embryonic lethal genetic variants and chromosomally normal pregnancy loss. <i>Fertility and Sterility</i> , 2021, 116, 1351-1358.	0.5	5
1927	Urine concentrating defect as presenting sign of progressive renal failure in Bardet-Biedl syndrome patients. <i>CKJ: Clinical Kidney Journal</i> , 2021, 14, 1545-1551.	1.4	8
1928	Phenylketonuria Diagnosis by Massive Parallel Sequencing and Genotype-Phenotype Association in Brazilian Patients. <i>Genes</i> , 2021, 12, 20.	1.0	5
1933	Wiedemann-Steiner Syndrome as a Differential Diagnosis of Cornelia de Lange Syndrome Using Targeted Next-Generation Sequencing: A Case Report. <i>Molecular Syndromology</i> , 2021, 12, 46-51.	0.3	11
1934	Mutation in histone deacetylase HDA-3 leads to shortened locomotor healthspan in <i>Caenorhabditis elegans</i> . <i>Aging</i> , 2020, 12, 23525-23547.	1.4	4
1935	The Effect of Synonymous Single-Nucleotide Polymorphisms on an Atypical Cystic Fibrosis Clinical Presentation. <i>Life</i> , 2021, 11, 14.	1.1	5
1937	Autozygosity mapping and time-to-spontaneous delivery in Norwegian parent-offspring trios. <i>Human Molecular Genetics</i> , 2021, 29, 3845-3858.	1.4	1
1938	Identification of New, Functionally Relevant Mutations in the Coding Regions of the Human Fos and Jun Proto-Oncogenes in Rheumatoid Arthritis Synovial Tissue. <i>Life</i> , 2021, 11, 5.	1.1	14
1939	Does Virus-Receptor Interplay Influence Human Coronaviruses Infection Outcome?. <i>Medical Science Monitor</i> , 2020, 26, e928572.	0.5	1
1941	A 6.7 kb deletion in the <i>COL2A1</i> gene in a Holstein calf with achondrogenesis type II and perosomus elumbis. <i>Animal Genetics</i> , 2021, 52, 244-245.	0.6	2
1944	Machine learning suggests polygenic risk for cognitive dysfunction in amyotrophic lateral sclerosis. <i>EMBO Molecular Medicine</i> , 2021, 13, e12595.	3.3	13
1945	De novo <i>FZR1</i> loss-of-function variants cause developmental and epileptic encephalopathies. <i>Brain</i> , 2022, 145, 1684-1697.	3.7	5
1946	Leveraging Northern European population history: novel low-frequency variants for polycystic ovary syndrome. <i>Human Reproduction</i> , 2022, 37, 352-365.	0.4	25

#	ARTICLE	IF	CITATIONS
1947	Uniparental Disomy Leading to a Rare Juvenile Form of ALS. <i>Journal of Pediatrics Perinatology and Child Health</i> , 2020, 04, 107-110.	0.0	1
1948	Mutational analysis of T315I in patients with chronic myeloid leukemia who did not respond to second-generation tyrosine kinase inhibitors. <i>Iraqi Journal of Hematology</i> , 2020, 9, 131.	0.0	0
1951	Pharmacogenomics Based Study for Liraglutide And Metformin (PCOS Drugs) Efficacy In Populations Across The Globe. , 2021, , .		0
1952	A Monoallelic Variant in REST Is Associated with Non-Syndromic Autosomal Dominant Hearing Impairment in a South African Family. <i>Genes</i> , 2021, 12, 1765.	1.0	5
1954	M-DATA: A statistical approach to jointly analyzing de novo mutations for multiple traits. <i>PLoS Genetics</i> , 2021, 17, e1009849.	1.5	4
1955	A population-specific reference panel for improved genotype imputation in African Americans. <i>Communications Biology</i> , 2021, 4, 1269.	2.0	15
1956	Frequency of allele variations in the CFTR gene in a Mexican population. <i>BMC Medical Genomics</i> , 2021, 14, 262.	0.7	0
1957	Network analysis reveals rare disease signatures across multiple levels of biological organization. <i>Nature Communications</i> , 2021, 12, 6306.	5.8	36
1958	The context-specific role of germline pathogenicity in tumorigenesis. <i>Nature Genetics</i> , 2021, 53, 1577-1585.	9.4	44
1959	Whole exome sequencing reveals potentially pathogenic variants in a small subset of premenopausal women with idiopathic osteoporosis. <i>Bone</i> , 2022, 154, 116253.	1.4	12
1961	De Novo Variants in the DYNC1H1 Gene Associated With Infantile Spasms. <i>Frontiers in Neurology</i> , 2021, 12, 733178.	1.1	2
1963	CSC software corrects off-target mediated gRNA depletion in CRISPR-Cas9 essentiality screens. <i>Nature Communications</i> , 2021, 12, 6461.	5.8	4
1964	The Impact of Rare Human Variants on Barrier-To-Auto-Integration Factor 1 (Banf1) Structure and Function. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 775441.	1.8	8
1965	Gene-level analysis of rare variants in 379,066 whole exome sequences identifies an association of GIGYF1 loss of function with type 2 diabetes. <i>Scientific Reports</i> , 2021, 11, 21565.	1.6	25
1966	Genetic and Functional Analysis of Glycosyltransferase 8 Domain-Containing Protein 1 in Taiwanese Patients With Amyotrophic Lateral Sclerosis. <i>Neurology: Genetics</i> , 2021, 7, e627.	0.9	2
1967	MYT1L-associated neurodevelopmental disorder: description of 40 new cases and literature review of clinical and molecular aspects. <i>Human Genetics</i> , 2022, 141, 65-80.	1.8	14
1968	Association between Paraoxonase-1 p.Q192R Polymorphism and Coronary Artery Disease susceptibility in the Colombian Population. <i>Vascular Health and Risk Management</i> , 2021, Volume 17, 689-699.	1.0	5
1969	Albino mice with the point mutation at the tyrosinase locus show high cholesterol diet-induced NASH susceptibility. <i>Scientific Reports</i> , 2021, 11, 21827.	1.6	2

#	ARTICLE	IF	CITATIONS
1972	SARS-CoV-2 susceptibility and COVID-19 disease severity are associated with genetic variants affecting gene expression in a variety of tissues. <i>Cell Reports</i> , 2021, 37, 110020.	2.9	25
1974	Pairwise effects between lipid GWAS genes modulate lipid plasma levels and cellular uptake. <i>Nature Communications</i> , 2021, 12, 6411.	5.8	6
1976	A Hidden Structural Variation in a Known IRD Gene: A Cautionary Tale of Two New Disease Candidate Genes. <i>Journal of Physical Education and Sports Management</i> , 2021, , mcs.a006131.	0.5	0
1977	Nâ€methylâ€dâ€aspartate (NMDA) receptor genetics: The power of paralog homology and protein dynamics in defining dominant genetic variants. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 556-568.	0.7	2
1979	Predicting novel candidate human obesity genes and their site of action by systematic functional screening in <i>Drosophila</i> . <i>PLoS Biology</i> , 2021, 19, e3001255.	2.6	7
1980	Assessing the effects of PMM2 variants on protein stability. <i>Molecular Genetics and Metabolism</i> , 2021, 134, 344-352.	0.5	2
1981	Laniakea@ReCaS: exploring the potential of customisable Galaxy on-demand instances as a cloud-based service. <i>BMC Bioinformatics</i> , 2021, 22, 544.	1.2	4
1982	One Genetic Defect and Two Related Entities in Monozygotic Twins: Otosclerosis and Superior Semicircular Canal Near Dehiscence Syndrome. <i>Journal of Audiology and Otology</i> , 2021, , .	0.2	0
1984	A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder. <i>Npj Genomic Medicine</i> , 2021, 6, 91.	1.7	9
1985	Myoclonic status epilepticus and cerebellar hypoplasia associated with a novel variant in the GRIA3 gene. <i>Neurogenetics</i> , 2021, , 1.	0.7	2
1986	Severe Congenital Thrombocytopenia Characterized by Decreased Platelet Sialylation and Moderate Complement Activation Caused by Novel Compound Heterozygous Variants in GNE. <i>Frontiers in Immunology</i> , 2021, 12, 777402.	2.2	7
1987	Deficiency in X-linked inhibitor of apoptosis protein promotes susceptibility to microbial triggers of intestinal inflammation. <i>Science Immunology</i> , 2021, 6, eabf7473.	5.6	15
1988	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. <i>BMJ, The</i> , 2021, 375, e066288.	3.0	42
1989	Returning actionable genomic results in a research biobank: Analytic validity, clinical implementation, and resource utilization. <i>American Journal of Human Genetics</i> , 2021, 108, 2224-2237.	2.6	34
1990	Clinical and Functional Characterization of Ryanodine Receptor 2 Variants Implicated in Calcium-Release Deficiency Syndrome. <i>JAMA Cardiology</i> , 2022, 7, 84.	3.0	28
1991	Clinical Spectrum and Geographic Distribution of Keratitis Fugax Hereditaria Caused by the Pathogenic Variant c.61G>C in NLRP3. <i>American Journal of Ophthalmology</i> , 2022, 236, 309-318.	1.7	5
1992	Autism-associated mutations in K _V 7 channels induce gating pore current. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	12
1998	Biallelic Mutation of SETX and Additional Likely â€œIn Cisâ€•SETX Sequence Change in Ataxia with Oculomotor Apraxia Type 2. <i>Journal of Pediatric Genetics</i> , 2021, 10, 311-314.	0.3	0

#	ARTICLE	IF	CITATIONS
2014	Interactive Web-Based Resource for Annotation of Genetic Variants Causing Hereditary Angioedema (HADA): Database Development, Implementation, and Validation. <i>Journal of Medical Internet Research</i> , 2020, 22, e19040.	2.1	4
2025	Digenic Variants in the FGF21 Signaling Pathway Associated with Severe Insulin Resistance and Pseudoacromegaly. <i>Journal of the Endocrine Society</i> , 2020, 4, bvaa138.	0.1	6
2026	Familial non-autoimmune hyperthyroidism in family members across four generations due to a novel disease-causing variant in the thyrotropin receptor gene. <i>Balkan Journal of Medical Genetics</i> , 2021, 23, 87-92.	0.5	4
2030	Germline variants are associated with increased primary melanoma tumor thickness at diagnosis. <i>Human Molecular Genetics</i> , 2021, 29, 3578-3587.	1.4	3
2032	Neuronal Transcriptome from Repeat Expanded Human Tissue is Associated with Loss of C9orf72 Function. <i>Free Neuropathology</i> , 2020, 1, .	2.4	1
2033	A resource-conserving serologic and highthroughput molecular approach to screen for blood donors with an IN:5 phenotype. <i>Immunohematology</i> , 2020, 36, 129-132.	0.2	1
2034	Drug Response Pharmacogenetics for 200,000 UK Biobank Participants. <i>Pacific Symposium on Biocomputing Pacific Symposium on Biocomputing</i> , 2021, 26, 184-195.	0.7	6
2035	Genotype phenotype analysis in a family carrying truncating mutations in the titin gene. <i>Acta Myologica</i> , 2021, 40, 61-65.	1.5	0
2036	Translating Pharmacogenomic Research to Therapeutic Potentials (Bench to Bedside). , 2021, , .		0
2037	The Balance Between Self-Renewal and Differentiation of Human Neural Stem Cells Requires the Amyloid Precursor Protein. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0
2038	Tag Variants of LGALS-3 Containing Haplotype Block in Advanced Carotid Atherosclerosis. <i>Journal of Stroke and Cerebrovascular Diseases</i> , 2022, 31, 106212.	0.7	1
2039	Decoding the cellular effects of genetic variation through interaction proteomics. <i>Current Opinion in Chemical Biology</i> , 2022, 66, 102100.	2.8	6
2040	NyuWa Genome resource: A deep whole-genome sequencing-based variation profile and reference panel for the Chinese population. <i>Cell Reports</i> , 2021, 37, 110017.	2.9	49
2041	Whole exome sequencing identifies monogenic forms of nephritis in a previously unsolved cohort of children with steroid-resistant nephrotic syndrome and hematuria. <i>Pediatric Nephrology</i> , 2021, , 1.	0.9	0
2042	Complete CFTR gene sequencing in 5,058 individuals with cystic fibrosis informs variant-specific treatment. <i>Journal of Cystic Fibrosis</i> , 2022, 21, 463-470.	0.3	13
2043	Mutation saturation for fitness effects at human CpG sites. <i>ELife</i> , 2021, 10, .	2.8	23
2044	Differential Regulation of Human Surfactant Protein A Genes, SFTPA1 and SFTPA2, and Their Corresponding Variants. <i>Frontiers in Immunology</i> , 2021, 12, 766719.	2.2	9
2045	The importance of automation in genetic diagnosis: Lessons from analyzing an inherited retinal degeneration cohort with the Mendelian Analysis Toolkit (MATK). <i>Genetics in Medicine</i> , 2022, 24, 332-343.	1.1	7

#	ARTICLE	IF	CITATIONS
2046	Sporadic and Lynch syndrome-associated mismatch repair-deficient brain tumors. <i>Laboratory Investigation</i> , 2022, 102, 160-171.	1.7	21
2048	Whole-exome analysis of adolescents with low VWF and heavy menstrual bleeding identifies novel genetic associations. <i>Blood Advances</i> , 2022, 6, 420-428.	2.5	4
2049	The global carrier frequency and genetic prevalence of Upshaw-Schulman syndrome. <i>BMC Genomic Data</i> , 2021, 22, 50.	0.7	8
2050	Homozygous duplication identified by whole genome sequencing causes LRBA deficiency. <i>Npj Genomic Medicine</i> , 2021, 6, 96.	1.7	3
2052	A novel insertion mutation in atlastin 1 is associated with spastic quadriplegia, increased membrane tethering, and aberrant conformational switching. <i>Journal of Biological Chemistry</i> , 2022, 298, 101438.	1.6	3
2053	Carrier Screening in the Mexican Jewish Community using a Pan-Ethnic Expanded Carrier Screening NGS Panel. <i>Genetics in Medicine</i> , 2021, , .	1.1	3
2054	The Mutational Landscape of PTK7 in Congenital Scoliosis and Adolescent Idiopathic Scoliosis. <i>Genes</i> , 2021, 12, 1791.	1.0	5
2055	Novel genetic variants in <i>MAPT</i> and alterations in tau phosphorylation in amyotrophic lateral sclerosis post-mortem motor cortex and cerebrospinal fluid. <i>Brain Pathology</i> , 2022, 32, e13035.	2.1	15
2057	Deep learning enables genetic analysis of the human thoracic aorta. <i>Nature Genetics</i> , 2022, 54, 40-51.	9.4	90
2058	Immunogenicity of Anti-SARS-CoV-2 Vaccines in Common Variable Immunodeficiency. <i>Journal of Clinical Immunology</i> , 2022, 42, 240-252.	2.0	48
2059	An Integrated Taxonomy for Monogenic Inflammatory Bowel Disease. <i>Gastroenterology</i> , 2022, 162, 859-876.	0.6	37
2060	<i>PHIP</i> gene variants with protein modeling, interactions, and clinical phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 579-589.	0.7	3
2061	Potential added value of combined DPYD/DPD genotyping and phenotyping to prevent severe toxicity in patients with a DPYD variant and decreased dihydropyrimidine dehydrogenase enzyme activity. <i>Journal of Oncology Pharmacy Practice</i> , 2021, , 107815522110491.	0.5	3
2062	Comprehensive characterization of copy number variation (CNV) called from array, long- and short-read data. <i>BMC Genomics</i> , 2021, 22, 826.	1.2	7
2063	Pathogenic in-Frame Variants in SCN8A: Expanding the Genetic Landscape of SCN8A-Associated Disease. <i>Frontiers in Pharmacology</i> , 2021, 12, 748415.	1.6	1
2065	Constructing germline research cohorts from the discarded reads of clinical tumor sequences. <i>Genome Medicine</i> , 2021, 13, 179.	3.6	25
2067	Genomic expansion of <i>Aldh1a1</i> protects beavers against high metabolic aldehydes from lipid oxidation. <i>Cell Reports</i> , 2021, 37, 109965.	2.9	7
2068	Assessing Foveal Structure in Individuals with TYR R402Q and S192Y Hypomorphic Alleles. <i>Ophthalmology Science</i> , 2021, 1, 100077.	1.0	2

#	ARTICLE	IF	CITATIONS
2069	Landscape of Driver Gene Events, Biomarkers and Druggable Targets Identified by Whole Genome Sequencing of Glioblastomas. <i>Neuro-Oncology Advances</i> , 2022, 4, vdab177.	0.4	3
2071	Lung transplantation for idiopathic pulmonary fibrosis enriches for individuals with telomere-mediated disease. <i>Journal of Heart and Lung Transplantation</i> , 2022, 41, 654-663.	0.3	19
2072	Genetic Polymorphisms Affecting Ranibizumab Response in High Myopia Patients. <i>Pharmaceutics</i> , 2021, 13, 1973.	2.0	2
2073	A novel <i>de novo</i> hemizygous <i>ARHGEF9</i> mutation associated with severe intellectual disability and epilepsy: a case report. <i>Journal of International Medical Research</i> , 2021, 49, 030006052110583.	0.4	1
2074	<i>NR2F1</i> database: 112 variants and 84 patients support refining the clinical synopsis of Boschâ€“Boonstraâ€“Schaaf optic atrophy syndrome. <i>Human Mutation</i> , 2022, 43, 128-142.	1.1	12
2075	A standardised hERG phenotyping pipeline to evaluate <i>KCNH2</i> genetic variant pathogenicity. <i>Clinical and Translational Medicine</i> , 2021, 11, e609.	1.7	7
2076	Loss-of-function mutation in <i>IKZF2</i> leads to immunodeficiency with dysregulated germinal center reactions and reduction of MAIT cells. <i>Science Immunology</i> , 2021, 6, eabe3454.	5.6	30
2078	A Novel <i>WT1</i> Mutation Identified in a 46,XX Testicular/Ovotesticular DSD Patient Results in the Retention of Intron 9. <i>Biology</i> , 2021, 10, 1248.	1.3	8
2079	Clinical and molecular characterization of a large cohort of childhood onset hereditary spastic paraplegias. <i>Scientific Reports</i> , 2021, 11, 22248.	1.6	8
2080	<i>BET1</i> variants establish impaired vesicular transport as a cause for muscular dystrophy with epilepsy. <i>EMBO Molecular Medicine</i> , 2021, 13, e13787.	3.3	9
2081	Clinical and Genetic Study of X-Linked Juvenile Retinoschisis in the Czech Population. <i>Genes</i> , 2021, 12, 1816.	1.0	4
2082	A catalog of curated breast cancer genes. <i>Breast Cancer Research and Treatment</i> , 2022, 191, 431-441.	1.1	3
2083	Further insight into the global variability of the <i>OCA2-HERC2</i> locus for human pigmentation from multiallelic markers. <i>Scientific Reports</i> , 2021, 11, 22530.	1.6	2
2084	A multi-enhancer <i>RET</i> regulatory code is disrupted in Hirschsprung disease. <i>Genome Research</i> , 2021, 31, 2199-2208.	2.4	10
2085	Rare germline variants in the <i>AXIN2</i> gene in families with colonic polyposis and colorectal cancer. <i>Familial Cancer</i> , 2021, , 1.	0.9	5
2086	The ACMG SF v3.0 gene list increases returnable variant detection by 22% when compared with v2.0 in the ClinSeq cohort. <i>Genetics in Medicine</i> , 2022, 24, 736-743.	1.1	7
2087	Whole Genome Sequence Analysis of the Plasma Proteome in Black Adults Provides Novel Insights Into Cardiovascular Disease. <i>Circulation</i> , 2022, 145, 357-370.	1.6	39
2088	Diagnostic Yield of Targeted Hearing Loss Gene Panel Sequencing in a Large German Cohort With a Balanced Age Distribution from a Single Diagnostic Center: An Eight-year Study. <i>Ear and Hearing</i> , 2022, 43, 1049-1066.	1.0	13

#	ARTICLE	IF	CITATIONS
2089	Identification of Pathogenic CNVs in Unexplained Developmental Disabilities Using Exome Sequencing: A Family Trio Study. Russian Journal of Genetics, 2021, 57, 1351-1355.	0.2	2
2090	Comprehensive molecular-genetic analysis of mid-frequency sensorineural hearing loss. Scientific Reports, 2021, 11, 22488.	1.6	2
2091	Whole genome sequencing and in vitro splice assays reveal genetic causes for inherited retinal diseases. Npj Genomic Medicine, 2021, 6, 97.	1.7	27
2094	Genomic Alterations in Human Papillomavirus-Positive and -Negative Conjunctival Squamous Cell Carcinomas. , 2021, 62, 11.		4
2095	Genetic Evidence for Congenital Vascular Disorders in Patients with VACTERL Association. European Journal of Pediatric Surgery, 2022, 32, 061-066.	0.7	0
2096	Analysis of 180 Genetic Variants in a New Interactive FX Variant Database Reveals Novel Insights into FX Deficiency. TH Open, 2021, 05, e557-e569.	0.7	3
2097	The MUC5B Promoter Polymorphism Associates With Severe COVID-19 in the European Population. Frontiers in Medicine, 2021, 8, 668024.	1.2	18
2098	Variant Selection and Interpretation: An Example of Modified VarSome Classifier of ACMG Guidelines in the Diagnostic Setting. Genes, 2021, 12, 1885.	1.0	4
2099	A 3D structural SARS-CoV-2-human interactome to explore genetic and drug perturbations. Nature Methods, 2021, 18, 1477-1488.	9.0	17
2100	Phase 1/dose expansion trial of brentuximab vedotin and lenalidomide in relapsed or refractory diffuse large B-cell lymphoma. Blood, 2022, 139, 1999-2010.	0.6	17
2103	Prediction and impact of personalized donation intervals. Vox Sanguinis, 2022, 117, 504-512.	0.7	9
2104	An infant with congenital respiratory insufficiency and diaphragmatic paralysis: A novel <i>BICD2</i> phenotype?. American Journal of Medical Genetics, Part A, 2022, 188, 926-930.	0.7	3
2107	Artificial intelligence (AI)-assisted exome reanalysis greatly aids in the identification of new positive cases and reduces analysis time in a clinical diagnostic laboratory. Genetics in Medicine, 2022, 24, 192-200.	1.1	19
2108	The splicing effect of variants at branchpoint elements in cancer genes. Genetics in Medicine, 2022, 24, 398-409.	1.1	9
2110	Estimated prevalence of Niemann-Pick type C disease in Quebec. Scientific Reports, 2021, 11, 22621.	1.6	5
2111	Can gene therapy be used to prevent cancer? Gene therapy for aldehyde dehydrogenase 2 deficiency. Cancer Gene Therapy, 2022, 29, 889-896.	2.2	1
2113	MutTMPredictor: Robust and accurate cascade XGBoost classifier for prediction of mutations in transmembrane proteins. Computational and Structural Biotechnology Journal, 2021, 19, 6400-6416.	1.9	16
2114	De novo <i>PBX1</i> variant in a patient with glaucoma, kidney anomalies, and developmental delay: An expansion of the <i>CAKUTHED</i> phenotype. American Journal of Medical Genetics, Part A, 2022, 188, 919-925.	0.7	6

#	ARTICLE	IF	CITATIONS
2115	Arginase 2 and Polyamines in Human Pancreatic Beta Cells: Possible Role in the Pathogenesis of Type 2 Diabetes. <i>International Journal of Molecular Sciences</i> , 2021, 22, 12099.	1.8	5
2116	Comprehensive Genetic Analysis Results of TSC1/TSC2 Genes in Patients with Clinical Suspicion of Tuberous Sclerosis Complex and Definition of 3 Novel Variants. , 2021, 38, 341-347.		1
2118	Decoding the effects of synonymous variants. <i>Nucleic Acids Research</i> , 2021, 49, 12673-12691.	6.5	17
2119	<i>RACGAP1</i> variants in a sporadic case of CDA III implicate the dysfunction of centralspindlin as the basis of the disease. <i>Blood</i> , 2022, 139, 1413-1418.	0.6	9
2120	Familial Hypomagnesemia With Secondary Hypocalcemia: A Case Report. <i>Cureus</i> , 2021, 13, e19847.	0.2	0
2121	In silico analysis of <i>GATA4</i> variants demonstrates main contribution to congenital heart disease. <i>Journal of Cardiovascular and Thoracic Research</i> , 2021, 13, 336-354.	0.3	2
2122	RNA assay identifies a previous misclassification of BARD1 c.1977A>G variant. <i>Scientific Reports</i> , 2021, 11, 22948.	1.6	0
2124	Quantitative retrospective natural history modeling of <i>WDR45</i> -related developmental and epileptic encephalopathy â€” a systematic cross-sectional analysis of 160 published cases. <i>Autophagy</i> , 2022, 18, 1715-1727.	4.3	5
2125	Monoallelic deleterious <i>MUTYH</i> germline variants as a driver for tumorigenesis. <i>Journal of Pathology</i> , 2022, 256, 214-222.	2.1	12
2127	Breast and prostate cancer risk: The interplay of polygenic risk, rare pathogenic germline variants, and family history. <i>Genetics in Medicine</i> , 2022, 24, 576-585.	1.1	22
2128	Case Report: Infantile Urticaria as a Herald of Neonatal Onset Multisystem Inflammatory Disease With a Novel Mutation in NLRP3. <i>Frontiers in Immunology</i> , 2021, 12, 775140.	2.2	2
2129	Concurrent Newborn Hearing and Genetic Screening in a Multi-Ethnic Population in South China. <i>Frontiers in Pediatrics</i> , 2021, 9, 734300.	0.9	5
2130	Genetic diversity of â€”Very Important Pharmacogenesâ€”™ in two South-Asian populations. <i>PeerJ</i> , 2021, 9, e12294.	0.9	2
2131	Endoplasmic stressâ€”inducing variants in <i>CPB1</i> and <i>CPA1</i> and risk of pancreatic cancer: A caseâ€”control study and metaâ€”analysis. <i>International Journal of Cancer</i> , 2022, 150, 1123-1133.	2.3	11
2133	Molecular and in vivo phenotyping of missense variants of the human glucagon receptor. <i>Journal of Biological Chemistry</i> , 2022, 298, 101413.	1.6	8
2134	Nature of spontaneously arising single base substitutions in normal cells. <i>Genome Instability & Disease</i> , 2021, 2, 339.	0.5	0
2135	Pediatric cardiomyopathy and the PCM Genes study: A summary with insights on genetic testing, variant interpretation, race and ethnicity. <i>Progress in Pediatric Cardiology</i> , 2021, 63, 101468.	0.2	0
2137	Should Patients with Kearns-Sayre Syndrome and Corneal Endothelial Failure Be Genotyped for a TCF4 Trinucleotide Repeat, Commonly Associated with Fuchs Endothelial Corneal Dystrophy?. <i>Genes</i> , 2021, 12, 1918.	1.0	0

#	ARTICLE	IF	CITATIONS
2138	Poking COVID-19: Insights on Genomic Constraints among Immune-Related Genes between Qatari and Italian Populations. <i>Genes</i> , 2021, 12, 1842.	1.0	1
2139	Quantifying evidence toward pathogenicity for rare phenotypes: The case of succinate dehydrogenase genes, SDHB and SDHD. <i>Genetics in Medicine</i> , 2022, 24, 41-50.	1.1	5
2140	Accurate assignment of disease liability to genetic variants using only population data. <i>Genetics in Medicine</i> , 2022, 24, 87-99.	1.1	4
2141	CCR5 and Biological Complexity: The Need for Data Integration and Educational Materials to Address Genetic/Biological Reductionism at the Interface of Ethical, Legal, and Social Implications. <i>Frontiers in Immunology</i> , 2021, 12, 790041.	2.2	5
2142	Bi-Allelic c.1746G>T; p.Leu582= Variants in &TUBGCP4& in a Boy with Autism: Clinical Data and Literature Review. <i>Molecular Syndromology</i> , 2022, 13, 165-170.	0.3	2
2143	Identification of discriminative gene-level and protein-level features associated with pathogenic gain-of-function and loss-of-function variants. <i>American Journal of Human Genetics</i> , 2021, 108, 2301-2318.	2.6	21
2144	Four decades in the making: Collagen III and mechanisms of vascular Ehlers Danlos Syndrome. <i>Matrix Biology Plus</i> , 2021, 12, 100090.	1.9	15
2145	A rare case of a male child with post-zygotic de novo mosaic variant c.538C&T in MECP2 gene: a case report of Rett syndrome. <i>BMC Neurology</i> , 2021, 21, 469.	0.8	4
2147	Closing the gap: Systematic integration of multiplexed functional data resolves variants of uncertain significance in BRCA1, TP53, and PTEN. <i>American Journal of Human Genetics</i> , 2021, 108, 2248-2258.	2.6	42
2150	Trellis for efficient data and task management in the VA Million Veteran Program. <i>Scientific Reports</i> , 2021, 11, 23229.	1.6	2
2151	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100074.	1.0	14
2152	Genomic characterization of non-schistosomiasis-related squamous cell carcinoma of the urinary bladder: A retrospective exploratory study. <i>PLoS ONE</i> , 2021, 16, e0259272.	1.1	4
2153	Most myopathic lamin variants aggregate: a functional genomics approach for assessing variants of uncertain significance. <i>Npj Genomic Medicine</i> , 2021, 6, 103.	1.7	12
2154	Compendium of human transcription factor effector domains. <i>Molecular Cell</i> , 2022, 82, 514-526.	4.5	46
2155	Variant interpretation using population databases: Lessons from gnomAD. <i>Human Mutation</i> , 2022, 43, 1012-1030.	1.1	184
2157	<i>APOE</i> Missense Variant R145C is Associated with Increased Alzheimer's Disease Risk in African Ancestry Individuals with the <i>APOE</i> Î·3/Î·4 Genotype. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0
2158	Collagen VI Muscle Disorders: Mutation Types, Pathogenic Mechanisms and Approaches to Therapy. <i>Advances in Experimental Medicine and Biology</i> , 2021, 1348, 311-323.	0.8	4
2159	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. <i>Human Molecular Genetics</i> , 2022, 31, 2307-2316.	1.4	8

#	ARTICLE	IF	CITATIONS
2160	High Coverage Whole Genome Sequencing of the Expanded 1000 Genomes Project Cohort Including 602 Trios. SSRN Electronic Journal, 0, , .	0.4	10
2161	A detection method for the capture of genomic signatures: From disease diagnosis to genome editing. Methods in Enzymology, 2021, 661, 251-282.	0.4	2
2162	Gene-mapping study of extremes of cerebral small vessel disease reveals TRIM47 as a strong candidate. Brain, 2022, 145, 1992-2007.	3.7	6
2163	Computational Methods and Approaches in Pharmacogenomic Research. , 2022, , 53-83.		1
2164	OUP accepted manuscript. Brain, 2022, , .	3.7	1
2165	Technical Performance of a 455-Gene Preventative Genomics Assay to Identify Multiple Variant Types Associated with Adult-Onset Monogenic Conditions, Susceptibility Loci, and Pharmacogenetic Insights. SSRN Electronic Journal, 0, , .	0.4	0
2166	NDUFV1 mutations in complex I deficiency: Case reports and review of symptoms. Genetics and Molecular Biology, 2021, 44, e20210149.	0.6	4
2167	Transthyretin Gene Variants and Associated Phenotypes in Danish Patients with Amyloid Cardiomyopathy. Neurology International, 2022, 12, 1-11.	0.2	1
2168	Clinical, pathological and molecular spectrum of patients with glycogen storage diseases in Pakistan. Journal of Pediatric Endocrinology and Metabolism, 2022, .	0.4	1
2169	Autoinflammatory Keratinization Disease With Hepatitis and Autism Reveals Roles for JAK1 Kinase Hyperactivity in Autoinflammation. Frontiers in Immunology, 2021, 12, 737747.	2.2	11
2170	Mutations in the ribosome biogenesis factor gene <i>LTV1</i> are linked to LIPHAK syndrome, a novel poikiloderma-like disorder. Human Molecular Genetics, 2022, 31, 1970-1978.	1.4	4
2171	DSP missense variant in a Scottish Highland calf with congenital ichthyosis, alopecia, acantholysis of the tongue and corneal defects. BMC Veterinary Research, 2022, 18, 20.	0.7	0
2173	Genome-wide analysis of copy number variants and normal facial variation in a large cohort of Bantu Africans. Human Genetics and Genomics Advances, 2022, 3, 100082.	1.0	1
2174	A mixed-ethnicity myoclonus-dystonia patient with a novel SGCE nonsense mutation: a case report. BMC Neurology, 2022, 22, 11.	0.8	0
2175	Clinical validation of genomic functional screen data: Analysis of observed BRCA1 variants in an unselected population cohort. Human Genetics and Genomics Advances, 2022, 3, 100086.	1.0	3
2176	Overcoming constraints on the detection of recessive selection in human genes from population frequency data. American Journal of Human Genetics, 2022, 109, 33-49.	2.6	5
2177	Clin.iobio: A Collaborative Diagnostic Workflow to Enable Team-Based Precision Genomics. Journal of Personalized Medicine, 2022, 12, 73.	1.1	1
2178	The role of junctophilin proteins in cellular function. Physiological Reviews, 2022, 102, 1211-1261.	13.1	25

#	ARTICLE	IF	CITATIONS
2179	Cellular Physiology and Pathophysiology of EAAT Anion Channels. <i>Frontiers in Cellular Neuroscience</i> , 2021, 15, 815279.	1.8	14
2182	The GG genotype of the serotonin 4 receptor genetic polymorphism, rs1345697, is associated with lower remission rates after antidepressant treatment: Findings from the METADAP cohort. <i>Journal of Affective Disorders</i> , 2022, 299, 335-343.	2.0	0
2183	The rs6942067 genotype is associated with a worse overall survival in young or non-smoking HPV-negative patients with positive nodal status in head and neck squamous cell carcinoma. <i>Oral Oncology</i> , 2022, 125, 105696.	0.8	0
2184	Leu226Trp CACNA1A variant associated with juvenile myoclonic epilepsy with and without intellectual disability. <i>Clinical Neurology and Neurosurgery</i> , 2022, 213, 107108.	0.6	2
2185	Rare coding variants in DNA damage repair genes associated with timing of natural menopause. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100079.	1.0	4
2186	Drug Response Pharmacogenetics for 200,000 UK Biobank Participants. , 2020, , .		7
2189	[CASE REPORT] Homozygous N-terminal missense variant in PLEKHG5 associated with intermediate CMT: a case report. <i>Journal of Neuromuscular Diseases</i> , 2021, , 1-5.	1.1	1
2190	First genome-wide association study of esophageal atresia identifies three genetic risk loci at CTNNA3, FOXF1/FOXC2/FOXL1, and HNF1B. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100093.	1.0	4
2191	Population-based screening in children for early diagnosis and treatment of familial hypercholesterolemia: design of the VRONI study. <i>European Journal of Public Health</i> , 2022, 32, 422-428.	0.1	11
2192	Phenotypic Heterogeneity among GBA p.R202X Carriers in Lewy Body Spectrum Disorders. <i>Biomedicines</i> , 2022, 10, 160.	1.4	0
2193	Inverting the model of genomics data sharing with the NHGRI Genomic Data Science Analysis, Visualization, and Informatics Lab-space. <i>Cell Genomics</i> , 2022, 2, 100085.	3.0	59
2194	Variant interpretation: UCSC Genome Browser Recommended Track Sets. <i>Human Mutation</i> , 2022, , .	1.1	2
2195	Computational Genomics in the Era of Precision Medicine: Applications to Variant Analysis and Gene Therapy. <i>Journal of Personalized Medicine</i> , 2022, 12, 175.	1.1	4
2196	Structure and Function of the ABCD1 Variant Database: 20 Years, 940 Pathogenic Variants, and 3400 Cases of Adrenoleukodystrophy. <i>Cells</i> , 2022, 11, 283.	1.8	23
2197	<sc><i>AOPEP</i></sc> Homozygous Loss-of-Function Variant in an Indian Patient with Early-onset Generalized Dystonia. <i>Movement Disorders</i> , 2022, 37, 874-875.	2.2	4
2198	Analysis of histone variant constraint and tissue expression suggests five potential novel human disease genes: H2AFY2, H2AFZ, H2AFY, H2AFV, H1FO. <i>Human Genetics</i> , 2022, 141, 1409-1421.	1.8	3
2199	Human INHBB Gene Variant (c.1079T>C;p.Met360Thr) Alters Testis Germ Cell Content, but Does Not Impact Fertility in Mice. <i>Endocrinology</i> , 2022, 163, .	1.4	2
2204	Genetic Susceptibility Toward Nausea and Vomiting in Surgical Patients. <i>Frontiers in Genetics</i> , 2021, 12, 816908.	1.1	1

#	ARTICLE	IF	CITATIONS
2205	Targeted Genotyping in Clinical Pharmacogenomics. <i>Journal of Molecular Diagnostics</i> , 2022, 24, 253-261.	1.2	13
2206	Comparative assessment of genes driving cancer and somatic evolution in non-cancer tissues: an update of the Network of Cancer Genes (NCC) resource. <i>Genome Biology</i> , 2022, 23, 35.	3.8	38
2207	Variant pathogenic prediction by locus variability: the importance of the current picture of evolution. <i>European Journal of Human Genetics</i> , 2022, 30, 555-559.	1.4	3
2209	Individualized Treatment for Patients With Familial Hypercholesterolemia. <i>Journal of Lipid and Atherosclerosis</i> , 2022, 11, 39.	1.1	4
2210	Whole-exome sequencing in syndromic craniosynostosis increases diagnostic yield and identifies candidate genes in osteogenic signaling pathways. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1464-1475.	0.7	7
2211	Mutations affecting the N-terminal domains of SHANK3 point to different pathomechanisms in neurodevelopmental disorders. <i>Scientific Reports</i> , 2022, 12, 902.	1.6	9
2212	Prevalence of Pathogenic and Potentially Pathogenic Inborn Error of Immunity Associated Variants in Children with Severe Sepsis. <i>Journal of Clinical Immunology</i> , 2022, 42, 350-364.	2.0	8
2213	Review of the Forensic Applicability of Biostatistical Methods for Inferring Ancestry from Autosomal Genetic Markers. <i>Genes</i> , 2022, 13, 141.	1.0	5
2214	The genetic architecture of pediatric cardiomyopathy. <i>American Journal of Human Genetics</i> , 2022, 109, 282-298.	2.6	21
2215	Novel biallelic variants expand the SLC5A6-related phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2022, 30, 439-449.	1.4	10
2216	Major Contribution of GREB1L Alterations to Severe Inner Ear Malformation Largely in a Non-mendelian Fashion. <i>Clinical and Experimental Otorhinolaryngology</i> , 2022, 15, 115-118.	1.1	5
2217	Interplay of Dinner Timing and <i>MTNR1B</i> Type 2 Diabetes Risk Variant on Glucose Tolerance and Insulin Secretion: A Randomized Crossover Trial. <i>Diabetes Care</i> , 2022, 45, 512-519.	4.3	26
2218	Rare variants in previously identified linkage regions associated with carotid plaque in Dominican Republic families. <i>PLoS ONE</i> , 2022, 17, e0250799.	1.1	1
2219	Association between 9p21-23 Locus and Frailty in a Community-Dwelling Greek Population: Results from the Hellenic Longitudinal Investigation of Ageing and Diet. <i>Journal of Prevention of Alzheimer's Disease</i> , 2022, 9, 1-9.	1.5	2
2220	Analysis of coding variants in the human FTO gene from the gnomAD database. <i>PLoS ONE</i> , 2022, 17, e0248610.	1.1	1
2221	Genomic Screening of Chronic Migraine Patients Identified Genes Linked to Drug and Endogenous Substances Metabolism. <i>BioNanoScience</i> , 2022, 12, 154-159.	1.5	1
2222	Novel RCBTB1 variants causing later-onset non-syndromic retinal dystrophy with macular chorioretinal atrophy. <i>Ophthalmic Genetics</i> , 2022, , 1-8.	0.5	2
2223	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. <i>American Journal of Human Genetics</i> , 2022, 109, 361-372.	2.6	6

#	ARTICLE	IF	CITATIONS
2224	Steroid-sensitive nephrotic syndrome candidate gene CLVS1 regulates podocyte oxidative stress and endocytosis. <i>JCI Insight</i> , 2022, 7, .	2.3	12
2225	Whole exome sequencing identifies potential candidate genes for spina bifida derived from mouse models. <i>American Journal of Medical Genetics, Part A</i> , 2022, , .	0.7	2
2226	A Novel Homozygous &i>KLHL3</i> Mutation as a Cause of Autosomal Recessive Pseudohypaldosteronism Type II Diagnosed Late in Life. <i>Nephron</i> , 2022, 146, 418-428.	0.9	4
2227	Exome-based mutation screening in South African children with primary congenital glaucoma. <i>Eye</i> , 2023, 37, 362-368.	1.1	2
2228	A Machine Learning Approach to Identifying Causal Monogenic Variants in Inflammatory Bowel Disease. , 2022, 1, 171-179.		0
2229	Genome-wide identification of the genetic basis of amyotrophic lateral sclerosis. <i>Neuron</i> , 2022, 110, 992-1008.e11.	3.8	51
2230	Case series of congenital pseudarthrosis of the tibia unfulfilling neurofibromatosis type 1 diagnosis: 21% with somatic NF1 haploinsufficiency in the periosteum. <i>Human Genetics</i> , 2022, 141, 1371-1383.	1.8	4
2231	Whole-Exome Sequencing Implicates Neuronal Calcium Channel with Familial Atrial Fibrillation. <i>Frontiers in Genetics</i> , 2022, 13, 806429.	1.1	1
2232	Developmental disorders caused by haploinsufficiency of transcriptional regulators: a perspective based on cell fate determination. <i>Biology Open</i> , 2022, 11, .	0.6	16
2233	Variants in Mitochondrial <sc>ATP</sc> Synthase Cause Variable Neurologic Phenotypes. <i>Annals of Neurology</i> , 2022, 91, 225-237.	2.8	12
2234	<sc>LRFN5</sc> locus structure is associated with autism and influenced by the sex of the individual and locus conversions. <i>Autism Research</i> , 2022, 15, 421-433.	2.1	9
2236	An evaluation of pipelines for DNA variant detection can guide a reanalysis protocol to increase the diagnostic ratio of genetic diseases. <i>Npj Genomic Medicine</i> , 2022, 7, 7.	1.7	8
2237	From karyotypes to precision genomics in 9p deletion and duplication syndromes. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100081.	1.0	9
2239	Association of ultra-rare coding variants with genetic generalized epilepsy: A case-control whole exome sequencing study. <i>Epilepsia</i> , 2022, 63, 723-735.	2.6	8
2240	Congenital anemia reveals distinct targeting mechanisms for master transcription factor GATA1. <i>Blood</i> , 2022, 139, 2534-2546.	0.6	14
2242	Recent advancements in understanding the genetic involvement of alpha-1 antitrypsin deficiency associated lung disease: a look at future precision medicine approaches. <i>Expert Review of Respiratory Medicine</i> , 2022, 16, 173-182.	1.0	1
2243	A somatic <i>UBA2</i> variant preceded <i>ETV6-RUNX1</i> in the concordant BCP-ALL of monozygotic twins. <i>Blood Advances</i> , 2022, 6, 2275-2289.	2.5	5
2244	Monoallelic MUTYH pathogenic variants ascertained via multi-gene hereditary cancer panels are not associated with colorectal, endometrial, or breast cancer. <i>Familial Cancer</i> , 2022, 21, 415-422.	0.9	7

#	ARTICLE	IF	CITATIONS
2245	<scp>Ehlersâ€Danlos</scp>/myopathy overlap syndrome caused by a large de novo deletion in <scp><i>COL12A1</i></scp>. American Journal of Medical Genetics, Part A, 2022, 188, 1556-1561.	0.7	3
2247	A de novo paradigm for male infertility. Nature Communications, 2022, 13, 154.	5.8	38
2248	Clinical, Genetic and Functional Characterization of a Novel AVPR2 Missense Mutation in a Woman with X-Linked Recessive Nephrogenic Diabetes Insipidus. Journal of Personalized Medicine, 2022, 12, 118.	1.1	2
2249	Identification of Survival-Specific Genes in Clear Cell Renal Cell Carcinoma Using a Customized Next-Generation Sequencing Gene Panel. Journal of Personalized Medicine, 2022, 12, 113.	1.1	2
2250	Establishing gene regulatory networks from Parkinsonâ€™s disease risk loci. Brain, 2022, 145, 2422-2435.	3.7	10
2252	Human population genomics approach in food metabolism. , 2022, , 433-449.		0
2253	Retinal cadherins and the retinal cadherinopathies: Current concepts and future directions. Progress in Retinal and Eye Research, 2022, 90, 101038.	7.3	11
2254	Case of Metastatic Pheochromocytoma and Meningiomas in a Patient With Lynch Syndrome. JCO Precision Oncology, 2022, 6, e2100251.	1.5	1
2255	StrVCTVRE: A supervised learning method to predict the pathogenicity of human genome structural variants. American Journal of Human Genetics, 2022, 109, 195-209.	2.6	29
2256	Mitochondrial DNA variation across 56,434 individuals in gnomAD. Genome Research, 2022, 32, 569-582.	2.4	59
2257	The p.<scp>Thr395Met</scp> missense variant of <scp><i>NFIA</i></scp> found in a patient with intellectual disability is a defective variant. American Journal of Medical Genetics, Part A, 2022, 188, 1184-1192.	0.7	5
2259	Genome-wide association study on 13â€™167 individuals identifies regulators of blood CD34+cell levels. Blood, 2022, 139, 1659-1669.	0.6	4
2260	Phenotyping Zebrafish Mutant Models to Assess Candidate Genes Associated with Aortic Aneurysm. Genes, 2022, 13, 123.	1.0	7
2262	Identification and characterization of novel <scp><i>MPC1</i></scp> gene variants causing mitochondrial pyruvate carrier deficiency. Journal of Inherited Metabolic Disease, 2022, 45, 264-277.	1.7	7
2263	Universal annotation of the human genome through integration of over a thousand epigenomic datasets. Genome Biology, 2022, 23, 9.	3.8	39
2264	Delineation of a novel neurodevelopmental syndrome associated with <i>PAX5</i> haploinsufficiency. Human Mutation, 2022, 43, 461-470.	1.1	5
2265	The global prevalence and ethnic heterogeneity of primary ciliary dyskinesia gene variants: a genetic database analysis. Lancet Respiratory Medicine,the, 2022, 10, 459-468.	5.2	63
2268	Revisiting the malaria hypothesis: accounting for polygenicity and pleiotropy. Trends in Parasitology, 2022, 38, 290-301.	1.5	5

#	ARTICLE	IF	CITATIONS
2269	Genotype-Phenotype Comparison in POGZ-Related Neurodevelopmental Disorders by Using Clinical Scoring. <i>Genes</i> , 2022, 13, 154.	1.0	6
2270	OUP accepted manuscript. <i>Briefings in Bioinformatics</i> , 2022, 23, .	3.2	5
2271	The Spectrum of Genetic Variants Associated with the Development of Monogenic Obesity in Qatar. <i>Obesity Facts</i> , 2022, 15, 357-365.	1.6	8
2272	An <i>in-silico</i> analysis to identify structural, functional and regulatory role of SNPs in <i>hMRE11</i> . <i>Journal of Biomolecular Structure and Dynamics</i> , 2023, 41, 2160-2174.	2.0	2
2273	Molecular Diagnostic Outcomes from 700 Cases. <i>Journal of Molecular Diagnostics</i> , 2022, 24, 274-286.	1.2	7
2274	Sequencing of a Chinese tetralogy of Fallot cohort reveals clustering mutations in myogenic heart progenitors. <i>JCI Insight</i> , 2022, 7, .	2.3	9
2275	NDUFA1 p.Gly32Arg variant in early-onset dementia. <i>Neurobiology of Aging</i> , 2022, 114, 113-116.	1.5	6
2276	Whole Genome Sequencing Unravels New Genetic Determinants of Early-Onset Familial Osteoporosis and Low BMD in Malta. <i>Genes</i> , 2022, 13, 204.	1.0	2
2277	Effect of Ions and Sequence Variants on the Antagonist Binding Properties of the Histamine H1 Receptor. <i>International Journal of Molecular Sciences</i> , 2022, 23, 1420.	1.8	3
2278	Evidence for a Dual-Pathway, 2-Hit Genetic Model for Focal Cortical Dysplasia and Epilepsy. <i>Neurology: Genetics</i> , 2022, 8, e652.	0.9	14
2279	The Scope of Pathogenic ABCA4 Mutations Targetable by CRISPR DNA Base Editing Systems – A Systematic Review. <i>Frontiers in Genetics</i> , 2021, 12, 814131.	1.1	4
2280	Inherited and de novo variants extend the etiology of TAOK1-associated neurodevelopmental disorder. <i>Journal of Physical Education and Sports Management</i> , 2022, , mcs.a006180.	0.5	6
2281	Understanding the impact of <i>ZBTB18</i> missense variation on transcription factor function in neurodevelopment and disease. <i>Journal of Neurochemistry</i> , 2022, 161, 219-235.	2.1	5
2282	Variant interpretation in molecular autopsy: a useful dilemma. <i>International Journal of Legal Medicine</i> , 2022, 136, 475-482.	1.2	9
2284	Functional variants in cytochrome b5 type A (CYB5A) are enriched in Southwest American Indian individuals and associate with obesity. <i>Obesity</i> , 2022, 30, 546-552.	1.5	2
2285	The breast pre-cancer atlas illustrates the molecular and micro-environmental diversity of ductal carcinoma in situ. <i>Npj Breast Cancer</i> , 2022, 8, 6.	2.3	13
2286	Redefining tissue specificity of genetic regulation of gene expression in the presence of allelic heterogeneity. <i>American Journal of Human Genetics</i> , 2022, 109, 223-239.	2.6	26
2287	Bi-allelic SMO variants in hypothalamic hamartoma: a recessive cause of Pallister-Hall syndrome. <i>European Journal of Human Genetics</i> , 2022, 30, 384-388.	1.4	6

#	ARTICLE	IF	CITATIONS
2288	Premature Termination Codon in 5' Region of Desmoplakin and Plakoglobin Genes May Escape Nonsense-Mediated Decay through the Reinitiation of Translation. <i>International Journal of Molecular Sciences</i> , 2022, 23, 656.	1.8	2
2289	SCHOOL: Software for Clinical Health in Oncology for Omics Laboratories. <i>Journal of Pathology Informatics</i> , 2022, 13, 100163.	0.8	4
2290	Diagnostic and management considerations in pseudohypoaldosteronism type 1b. <i>BMJ Case Reports</i> , 2022, 15, e246538.	0.2	0
2291	Increased incidence of pathogenic variants in ATM in the context of testing for breast and ovarian cancer predisposition. <i>Journal of Human Genetics</i> , 2022, , .	1.1	3
2292	Comprehensive Genetic Analysis of RASopathy in the Era of Next-Generation Sequencing and Definition of a Novel Likely Pathogenic KRAS Variation. <i>Molecular Syndromology</i> , 2022, 13, 150-160.	0.3	1
2294	Genetic and phenotypic variability in adult patients with Niemann Pick type C from Serbia: single-center experience. <i>Journal of Neurology</i> , 2022, , 1.	1.8	1
2297	PDZD8 Disruption Causes Cognitive Impairment in Humans, Mice, and Fruit Flies. <i>Biological Psychiatry</i> , 2022, 92, 323-334.	0.7	14
2298	Functional characterisation of the amyotrophic lateral sclerosis risk locus GPX3/TNIP1. <i>Genome Medicine</i> , 2022, 14, 7.	3.6	12
2299	Loss-of-function mutations in the co-chaperone protein BAG5 cause dilated cardiomyopathy requiring heart transplantation. <i>Science Translational Medicine</i> , 2022, 14, eabf3274.	5.8	16
2300	The characteristics of early-stage research into human genes are substantially different from subsequent research. <i>PLoS Biology</i> , 2022, 20, e3001520.	2.6	5
2301	Rare coding variants in 35 genes associate with circulating lipid levels—A multi-ancestry analysis of 170,000 exomes. <i>American Journal of Human Genetics</i> , 2022, 109, 81-96.	2.6	24
2302	Clinico-radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. <i>Human Mutation</i> , 2022, 43, 403-419.	1.1	9
2303	Systematic analysis of naturally occurring insertions and deletions that alter transcription factor spacing identifies tolerant and sensitive transcription factor pairs. <i>ELife</i> , 2022, 11, .	2.8	5
2304	Impaired catabolism of free oligosaccharides due to MAN2C1 variants causes a neurodevelopmental disorder. <i>American Journal of Human Genetics</i> , 2022, 109, 345-360.	2.6	4
2305	A Compound Heterozygous Mutation in Calpain 1 Identifies a New Genetic Cause for Spinal Muscular Atrophy Type 4 (SMA4). <i>Frontiers in Genetics</i> , 2021, 12, 801253.	1.1	1
2306	Mast Syndrome Outside the Amish Community: SPC21 in Europe. <i>Frontiers in Neurology</i> , 2021, 12, 799953.	1.1	0
2307	Genome sequencing and RNA sequencing of urinary cells reveal an intronic FBN1 variant causing aberrant splicing. <i>Journal of Human Genetics</i> , 2022, 67, 387-392.	1.1	7
2308	Pharmacogenomics of Vincristine-Induced Peripheral Neuropathy in Children with Cancer: A Systematic Review and Meta-Analysis. <i>Cancers</i> , 2022, 14, 612.	1.7	10

#	ARTICLE	IF	CITATIONS
2309	Recessive <i>GCH1</i> Deficiency Causing DOPA-Responsive Dystonia Diagnosed by Reported Negative Exome. <i>Pediatrics</i> , 2022, 149, .	1.0	1
2310	Amelioration of a neurodevelopmental disorder by carbamazepine in a case having a gain-of-function <i>GRIA3</i> variant. <i>Human Genetics</i> , 2022, 141, 283-293.	1.8	6
2311	Linkage disequilibrium between rare mutations. <i>Genetics</i> , 2022, 220, .	1.2	15
2312	Common disease-associated gene variants in a Saudi Arabian population. <i>Annals of Saudi Medicine</i> , 2022, 42, 29-35.	0.5	9
2313	Expanding the genotype and phenotype spectrum of <i>SYT1</i> -associated neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, 24, 880-893.	1.1	14
2315	Evaluation of Evidence for Pathogenicity Demonstrates That <i>BLK</i> , <i>KLF11</i> , and <i>PAX4</i> Should Not Be Included in Diagnostic Testing for MODY. <i>Diabetes</i> , 2022, 71, 1128-1136.	0.3	27
2316	A clinician's guide to omics resources in dermatology. <i>Clinical and Experimental Dermatology</i> , 2022, , .	0.6	1
2317	Cardiovascular Genetics. <i>Medical Clinics of North America</i> , 2022, 106, 313-324.	1.1	1
2318	ANSWER: Annotation Software for Electronic Reporting. <i>JCO Clinical Cancer Informatics</i> , 2022, 6, e2100113.	1.0	3
2319	Genetic variants in eleven central and peripheral chemoreceptor genes in sudden infant death syndrome. <i>Pediatric Research</i> , 2022, 92, 1026-1033.	1.1	4
2320	Mosaicism in <i>PTEN</i> —new case and comment on the literature. <i>European Journal of Human Genetics</i> , 2022, 30, 641-644.	1.4	6
2321	Qatar genome: Insights on genomics from the Middle East. <i>Human Mutation</i> , 2022, 43, 499-510.	1.1	29
2322	Investigating the interference of single nucleotide polymorphisms with miRNA mediated gene regulation in pancreatic ductal adenocarcinoma: An in silico approach. <i>Gene</i> , 2022, 819, 146259.	1.0	1
2323	Application of Whole Exome Sequencing and Functional Annotations to Identify Genetic Variants Associated with Marfan Syndrome. <i>Journal of Personalized Medicine</i> , 2022, 12, 198.	1.1	2
2324	Recessive variants in <i>COL25A1</i> gene as novel cause of arthrogryposis multiplex congenita with ocular congenital cranial dysinnervation disorder. <i>Human Mutation</i> , 2022, 43, 487-498.	1.1	8
2327	High-throughput evaluation of epilepsy-associated <i>KCNQ2</i> variants reveals functional and pharmacological heterogeneity. <i>JCI Insight</i> , 2022, 7, .	2.3	27
2329	Genome Sequencing for Genetics Diagnosis of Patients With Intellectual Disability: The DEFIDIAG Study. <i>Frontiers in Genetics</i> , 2021, 12, 766964.	1.1	7
2330	Probing the functional consequence and clinical relevance of <i>CD320</i> p.E88del, a variant in the transcobalamin receptor gene. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1124-1141.	0.7	2

#	ARTICLE	IF	CITATIONS
2331	A Framework of Critical Considerations in Clinical Exome Reanalyses by Clinical and Laboratory Standards Institute. <i>Journal of Molecular Diagnostics</i> , 2022, 24, 177-188.	1.2	4
2332	Evaluation of cytosine base editing and adenine base editing as a potential treatment for alpha-1 antitrypsin deficiency. <i>Molecular Therapy</i> , 2022, 30, 1396-1406.	3.7	13
2333	Can leaky splicing and evasion of premature termination codon surveillance contribute to the phenotypic variability in Alkuraya-Kucinkas syndrome?. <i>European Journal of Medical Genetics</i> , 2022, 65, 104427.	0.7	3
2334	Identifying patients and assessing variant pathogenicity for an autosomal dominant disease-driving gene. <i>STAR Protocols</i> , 2022, 3, 101150.	0.5	4
2335	Sodium channel expression and transcript variation in the developing brain of human, Rhesus monkey, and mouse. <i>Neurobiology of Disease</i> , 2022, 164, 105622.	2.1	6
2336	HKG: an open genetic variant database of 205 Hong Kong cantonese exomes. <i>NAR Genomics and Bioinformatics</i> , 2022, 4, lqac005.	1.5	2
2337	Dynein axonemal heavy chain 9 M4374I variation may have an effect on imatinib mesylate resistance in CML. <i>Medicine International</i> , 2022, 2, .	0.2	0
2338	Machine learning methods for prediction of cancer driver genes: a survey paper. <i>Briefings in Bioinformatics</i> , 2022, 23, .	3.2	15
2339	Novel Cranial Imaging Findings and a Splice-Site Variant in a Patient with Tyrosinemia Type III, and a Summary of Published Cases. <i>Molecular Syndromology</i> , 2022, 13, 193-199.	0.3	1
2340	Characterizing mobile element insertions in 5675 genomes. <i>Nucleic Acids Research</i> , 2022, 50, 2493-2508.	6.5	16
2341	A dominant negative variant of <i>RAB5B</i> disrupts maturation of surfactant protein B and surfactant protein C. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	9
2343	Genome-Wide De Novo Variants in Congenital Heart Disease Are Not Associated With Maternal Diabetes or Obesity. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, CIRCGEN121003500.	1.6	8
2344	The earliest events in <i>BRAF</i> mutant colorectal cancer: exome sequencing of sessile serrated lesions with a tiny focus dysplasia or cancer reveals recurring mutations in two distinct progression pathways. <i>Journal of Pathology</i> , 2022, 257, 239-249.	2.1	5
2345	Centers for Mendelian Genomics: A decade of facilitating gene discovery. <i>Genetics in Medicine</i> , 2022, 24, 784-797.	1.1	44
2346	<i>CELSR1</i> Risk Alleles in Familial Bicuspid Aortic Valve and Hypoplastic Left Heart Syndrome. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, CIRCGEN121003523.	1.6	11
2347	CCR8-targeted specific depletion of clonally expanded Treg cells in tumor tissues evokes potent tumor immunity with long-lasting memory. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	68
2348	Known allosteric proteins have central roles in genetic disease. <i>PLoS Computational Biology</i> , 2022, 18, e1009806.	1.5	2
2349	Genome-Wide Characterization of a Highly Penetrant Form of Hyperlipoprotein(a)emia Associated With Genetically Elevated Cardiovascular Risk. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, CIRCGEN121003489.	1.6	5

#	ARTICLE	IF	CITATIONS
2350	Channelopathy Genes in Pulmonary Arterial Hypertension. <i>Biomolecules</i> , 2022, 12, 265.	1.8	6
2351	The Past and Future of Rare Skin Disease Research and Therapy. <i>Journal of Investigative Dermatology</i> , 2022, 142, 1010-1014.	0.3	1
2352	A roadmap to increase diversity in genomic studies. <i>Nature Medicine</i> , 2022, 28, 243-250.	15.2	195
2353	Case Report: Two Families With HPDL Related Neurodegeneration. <i>Frontiers in Genetics</i> , 2022, 13, 780764.	1.1	4
2354	BnVIR: bridging the genotype-phenotype gap to accelerate mining of candidate variations underlying agronomic traits in <i>Brassica napus</i> . <i>Molecular Plant</i> , 2022, 15, 779-782.	3.9	13
2355	Pathogenic variants in the human m6A reader YTHDC2 are associated with primary ovarian insufficiency. <i>JCI Insight</i> , 2022, 7, .	2.3	8
2356	Case Report of Gastric Bypass Complicated with Genetic-Associated Thrombosis. <i>BioNanoScience</i> , 0, , 1.	1.5	0
2357	Mouse Model of a Human STAT4 Point Mutation That Predisposes to Disseminated Coccidiomycosis. <i>ImmunoHorizons</i> , 2022, 6, 130-143.	0.8	9
2358	Activating <i>RAC1</i> variants in the switch II region cause a developmental syndrome and alter neuronal morphology. <i>Brain</i> , 2022, 145, 4232-4245.	3.7	6
2359	A Founder Mutation in EHD1 Presents with Tubular Proteinuria and Deafness. <i>Journal of the American Society of Nephrology: JASN</i> , 2022, 33, 732-745.	3.0	7
2360	Analysis of missense variants in the human genome reveals widespread gene-specific clustering and improves prediction of pathogenicity. <i>American Journal of Human Genetics</i> , 2022, 109, 457-470.	2.6	29
2361	Combined effects of host genetics and diet on human gut microbiota and incident disease in a single population cohort. <i>Nature Genetics</i> , 2022, 54, 134-142.	9.4	164
2362	Progranulin mutations in clinical and neuropathological Alzheimer's disease. <i>Alzheimer's and Dementia</i> , 2022, 18, 2458-2467.	0.4	12
2363	Somatic mutation analyses of stem-like cells in gingivobuccal oral squamous cell carcinoma reveals DNA damage response genes. <i>Genomics</i> , 2022, 114, 110308.	1.3	1
2364	Tumour mutational burden: an overview for pathologists. <i>Pathology</i> , 2022, 54, 249-253.	0.3	7
2365	Population-level deficit of homozygosity unveils CPSF3 as an intellectual disability syndrome gene. <i>Nature Communications</i> , 2022, 13, 705.	5.8	7
2366	Deleterious variants in <i>CRLS1</i> lead to cardiolipin deficiency and cause an autosomal recessive multi-system mitochondrial disease. <i>Human Molecular Genetics</i> , 2022, 31, 3597-3612.	1.4	11
2367	Polygenic risk impacts <i>PDGFRA</i> mutation penetrance in non-syndromic cleft lip and palate. <i>Human Molecular Genetics</i> , 2022, 31, 2348-2357.	1.4	7

#	ARTICLE	IF	CITATIONS
2368	Genome sequencing in a genetically elusive multigenerational long QT syndrome pedigree identifies a novel LQT2-causative deeply intronic KCNH2 variant. <i>Heart Rhythm</i> , 2022, 19, 998-1007.	0.3	10
2369	The Diverse Phenotype of Intestinal Dysmotility Secondary to ACTG2-related Disorders. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2022, 74, 575-581.	0.9	4
2372	De Novo and Dominantly Inherited <i>SPTAN1</i> Mutations Cause Spastic Paraplegia and Cerebellar Ataxia. <i>Movement Disorders</i> , 2022, 37, 1175-1186.	2.2	9
2374	Prioritization of putatively detrimental variants in euploid miscarriages. <i>Scientific Reports</i> , 2022, 12, 1997.	1.6	3
2375	The 90 plus: longevity and COVID-19 survival. <i>Molecular Psychiatry</i> , 2022, , .	4.1	2
2376	The TKFC Ala185Thr variant, reported as "null" for fructose metabolism, is fully active as triokinase. <i>FEBS Letters</i> , 2022, , .	1.3	1
2377	Whole exome sequencing in Alopecia Areata identifies rare variants in KRT82. <i>Nature Communications</i> , 2022, 13, 800.	5.8	10
2378	Autosomal recessive nonsyndromic hearing impairment in two Finnish families due to the population enriched CABP2 c.637+1G>T variant. <i>Molecular Genetics & Genomic Medicine</i> , 2022, , e1866.	0.6	1
2379	A postzygotic de novo NCDN mutation identified in a sporadic FTLN patient results in neurochondrin haploinsufficiency and altered FUS granule dynamics. <i>Acta Neuropathologica Communications</i> , 2022, 10, 20.	2.4	5
2382	Identification of recurrent genetic patterns from targeted sequencing panels with advanced data science: a case-study on sporadic and genetic neurodegenerative diseases. <i>BMC Medical Genomics</i> , 2022, 15, 26.	0.7	4
2383	Signatures of TOP1 transcription-associated mutagenesis in cancer and germline. <i>Nature</i> , 2022, 602, 623-631.	13.7	38
2384	DECIPHER: Supporting the interpretation and sharing of rare disease phenotype-linked variant data to advance diagnosis and research. <i>Human Mutation</i> , 2022, , .	1.1	10
2385	Variant Identification in <i>BARD1</i> , <i>PRDM9</i> , <i>RCC1</i> , and <i>RECQL</i> in Patients with Ovarian Cancer by Targeted Next-generation Sequencing of DNA Pools. <i>Cancer Prevention Research</i> , 2022, 15, 151-160.	0.7	2
2387	Sarm1 haploinsufficiency or low expression levels after antisense oligonucleotides delay programmed axon degeneration. <i>Cell Reports</i> , 2021, 37, 110108.	2.9	15
2388	Genomic Features of Muscle-invasive Bladder Cancer Arising After Prostate Radiotherapy. <i>European Urology</i> , 2022, 81, 466-473.	0.9	12
2389	Loss of Sucrase-Isomaltase Function Increases Acetate Levels and Improves Metabolic Health in Greenlandic Cohorts. <i>Gastroenterology</i> , 2022, 162, 1171-1182.e3.	0.6	9
2390	A reference-quality, fully annotated genome from a Puerto Rican individual. <i>Genetics</i> , 2022, 220, .	1.2	7
2394	Embryonic osteocalcin signaling determines lifelong adrenal steroidogenesis and homeostasis in the mouse. <i>Journal of Clinical Investigation</i> , 2022, 132, .	3.9	16

#	ARTICLE	IF	CITATIONS
2395	Impaired SorLA maturation and trafficking as a new mechanism for SORL1 missense variants in Alzheimer disease. <i>Acta Neuropathologica Communications</i> , 2021, 9, 196.	2.4	17
2396	COBRE for Computational Biology of Human Disease at Brown University: Progress and Prospects. <i>Rhode Island Medical Journal (2013)</i> , 2021, 104, 54-59.	0.2	0
2397	Identification of heritable rare variants associated with early-stage lung adenocarcinoma risk. <i>Translational Lung Cancer Research</i> , 2022, 11, 509-522.	1.3	5
2398	Scaling up oligogenic diseases research with OLIDA: the Oligogenic Diseases Database. <i>Database: the Journal of Biological Databases and Curation</i> , 2022, 2022, .	1.4	10
2399	Computational Resources for the Interpretation of Variations in Cancer. <i>Advances in Experimental Medicine and Biology</i> , 2022, 1361, 177-198.	0.8	2
2400	Somatic variants in diverse genes leads to a spectrum of focal cortical malformations. <i>Brain</i> , 2022, 145, 2704-2720.	3.7	33
2401	Characterization of the 3'UTR of the BTD gene and identification of regulatory elements and microRNAs. <i>Genetics and Molecular Biology</i> , 2022, 45, e20200432.	0.6	2
2402	Lamina Propria Phagocyte Profiling Reveals Targetable Signaling Pathways in Refractory Inflammatory Bowel Disease. , 2022, 1, 380-392.		5
2403	Somatic and Germline Variant Calling from Next-Generation Sequencing Data. <i>Advances in Experimental Medicine and Biology</i> , 2022, 1361, 37-54.	0.8	2
2404	Alterations in synaptonemal complex coding genes and human infertility. <i>International Journal of Biological Sciences</i> , 2022, 18, 1933-1943.	2.6	7
2406	De novo loss-of-function variant in <i>PTDSS1</i> is associated with developmental delay. <i>American Journal of Medical Genetics, Part A</i> , 2022, , .	0.7	0
2407	Clonal hematopoiesis as a pitfall in germline variant interpretation in the context of Mendelian disorders. <i>Human Molecular Genetics</i> , 2022, 31, 2386-2395.	1.4	7
2408	A guide for the diagnosis of rare and undiagnosed disease: beyond the exome. <i>Genome Medicine</i> , 2022, 14, 23.	3.6	101
2409	New insights on familial colorectal cancer type X syndrome. <i>Scientific Reports</i> , 2022, 12, 2846.	1.6	10
2410	The RDConnect Genome-Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. <i>Human Mutation</i> , 2022, , .	1.1	18
2411	CHEK2p.I157T Mutation Is Associated with Increased Risk of Adult-Type Ovarian Granulosa Cell Tumors. <i>Cancers</i> , 2022, 14, 1208.	1.7	0
2412	Clinical pharmacogenetic analysis in 5,001 individuals with diagnostic Exome Sequencing data. <i>Npj Genomic Medicine</i> , 2022, 7, 12.	1.7	10
2413	Bi-allelic variants in <i>CHKA</i> cause a neurodevelopmental disorder with epilepsy and microcephaly. <i>Brain</i> , 2022, 145, 1916-1923.	3.7	3

#	ARTICLE	IF	CITATIONS
2414	Integrated Genomic Analysis Identifies <i>UBTF</i> Tandem Duplications as a Recurrent Lesion in Pediatric Acute Myeloid Leukemia. <i>Blood Cancer Discovery</i> , 2022, 3, 194-207.	2.6	38
2415	Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. <i>Kidney International</i> , 2022, 101, 1039-1053.	2.6	8
2418	<i>PRUNE1</i> c.933G>A synonymous variant induces exon 7 skipping, disrupts the <i>DHHA2</i> domain, and leads to an atypical <i>NMIHBA</i> syndrome presentation: Case report and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1868-1874.	0.7	2
2419	Exploring the relevance of NUP93 variants in steroid-resistant nephrotic syndrome using next generation sequencing and a fly kidney model. <i>Pediatric Nephrology</i> , 2022, 37, 2643-2656.	0.9	5
2422	TNNT1 myopathy with novel compound heterozygous mutations. <i>Neuromuscular Disorders</i> , 2022, 32, 176-184.	0.3	5
2423	Searching for Constitutive Androstane Receptor Modulators. <i>Drug Metabolism and Disposition</i> , 2022, 50, 1002-1009.	1.7	5
2424	The Molecular Tumor Board Portal supports clinical decisions and automated reporting for precision oncology. <i>Nature Cancer</i> , 2022, 3, 251-261.	5.7	44
2425	Diagnostic yield of multi-gene panel for muscular dystrophies and other hereditary myopathies. <i>Neurological Sciences</i> , 2022, 43, 4473-4481.	0.9	4
2426	Placental methylome reveals a 22q13.33 brain regulatory gene locus associated with autism. <i>Genome Biology</i> , 2022, 23, 46.	3.8	22
2427	TDP-43 loss and ALS-risk SNPs drive mis-splicing and depletion of <i>UNC13A</i> . <i>Nature</i> , 2022, 603, 131-137.	13.7	188
2428	Usher syndrome type IV: clinically and molecularly confirmed by novel <i>ARSG</i> variants. <i>Human Genetics</i> , 2022, 141, 1723-1738.	1.8	16
2429	Variant Enrichment Analysis to Explore Pathways Functionality in Complex Autoinflammatory Skin Disorders through Whole Exome Sequencing Analysis. <i>International Journal of Molecular Sciences</i> , 2022, 23, 2278.	1.8	12
2430	Systematic benchmark of state-of-the-art variant calling pipelines identifies major factors affecting accuracy of coding sequence variant discovery. <i>BMC Genomics</i> , 2022, 23, 155.	1.2	23
2431	Rare catastrophes and evolutionary legacies: human germline gene variants in <i>MLKL</i> and the necroptosis signalling pathway. <i>Biochemical Society Transactions</i> , 2022, 50, 529-539.	1.6	5
2433	Rare disorders have many faces: in silico characterization of rare disorder spectrum. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 76.	1.2	6
2434	Epilepsy Genetics and Precision Medicine in Adults: A New Landscape for Developmental and Epileptic Encephalopathies. <i>Frontiers in Neurology</i> , 2022, 13, 777115.	1.1	21
2435	Implementation of CYP2D6 copy-number imputation panel and frequency of key pharmacogenetic variants in Finnish individuals with a psychotic disorder. <i>Pharmacogenomics Journal</i> , 2022, 22, 166-172.	0.9	6
2436	<i>HEATR3</i> variants impair nuclear import of uL18 (RPL5) and drive Diamond-Blackfan anemia. <i>Blood</i> , 2022, 139, 3111-3126.	0.6	15

#	ARTICLE	IF	CITATIONS
2437	<i>PMS2</i> variant results in loss of <sc>ATPase</sc> activity without compromising mismatch repair. <i>Molecular Genetics & Genomic Medicine</i> , 2022, , e1908.	0.6	1
2438	Spatiotemporal dynamics of clonal selection and diversification in normal endometrial epithelium. <i>Nature Communications</i> , 2022, 13, 943.	5.8	24
2442	A Novel, Apparently Silent Variant in MFSD8 Causes Neuronal Ceroid Lipofuscinosis with Marked Intrafamilial Variability. <i>International Journal of Molecular Sciences</i> , 2022, 23, 2271.	1.8	3
2443	Genotype-phenotype correlations for COL4A3-COL4A5 variants resulting in Gly substitutions in Alport syndrome. <i>Scientific Reports</i> , 2022, 12, 2722.	1.6	21
2444	A retrospective two centre study of Birt-Hogg-DubÃ© syndrome reveals a pathogenic founder mutation in FLCN in the Swedish population. <i>PLoS ONE</i> , 2022, 17, e0264056.	1.1	5
2446	Large Phenotypic Variation of Individuals from a Family with a Novel ASPM Mutation Associated with Microcephaly, Epilepsy, and Behavioral and Cognitive Deficits. <i>Genes</i> , 2022, 13, 429.	1.0	1
2448	Genetic resiliency associated with dominant lethal TPM1 mutation causing atrial septal defect with high heritability. <i>Cell Reports Medicine</i> , 2022, 3, 100501.	3.3	0
2450	<i>NCOR2</i> is a novel candidate gene for migraine-epilepsy phenotype. <i>Cephalalgia</i> , 2022, 42, 631-644.	1.8	6
2452	The R369 Myosin Residue within Loop 4 Is Critical for Actin Binding and Muscle Function in <i>Drosophila</i> . <i>International Journal of Molecular Sciences</i> , 2022, 23, 2533.	1.8	1
2454	Precision targeting tumor cells using cancer-specific InDel mutations with CRISPR-Cas9. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	15
2455	<i>PIGN</i> encephalopathy: Characterizing the epileptology. <i>Epilepsia</i> , 2022, 63, 974-991.	2.6	4
2456	IL-11 Is Elevated and Drives the Profibrotic Phenotype Transition of Orbital Fibroblasts in Thyroid-Associated Ophthalmopathy. <i>Frontiers in Endocrinology</i> , 2022, 13, 846106.	1.5	12
2457	Transcriptome-wide association study of coronary artery disease identifies novel susceptibility genes. <i>Basic Research in Cardiology</i> , 2022, 117, 6.	2.5	22
2458	Monogenic Versus Multifactorial Inheritance in the Development of Isolated Cleft Palate: A Whole Genome Sequencing Study. <i>Frontiers in Genetics</i> , 2022, 13, 828534.	1.1	5
2459	m.3685T>C is a Novel Mitochondrial DNA Variant That Causes Leigh Syndrome. <i>Journal of Physical Education and Sports Management</i> , 2022, , mcs.a006136.	0.5	0
2460	Genetic associations of protein-coding variants in human disease. <i>Nature</i> , 2022, 603, 95-102.	13.7	67
2461	Low Mutational Burden of Extranodal Marginal Zone Lymphoma of Mucosa-Associated Lymphoid Tissue in Patients with Primary Sjogren-™s Syndrome. <i>Cancers</i> , 2022, 14, 1010.	1.7	5
2462	Genetic Epidemiology of Medication Safety and Efficacy Related Variants in the Central Han Chinese Population With Whole Genome Sequencing. <i>Frontiers in Pharmacology</i> , 2021, 12, 790832.	1.6	3

#	ARTICLE	IF	CITATIONS
2463	Analysis of rare genetic variation underlying cardiometabolic diseases and traits among 200,000 individuals in the UK Biobank. <i>Nature Genetics</i> , 2022, 54, 240-250.	9.4	68
2464	Accuracy and applications of sequencing and genotyping approaches for CYP2A6 and homologous genes. <i>Pharmacogenetics and Genomics</i> , 2022, Publish Ahead of Print, .	0.7	1
2465	Exome Sequencing Identifies a Novel <i>SIN3A</i> Variant in a Patient with Witteveen-Kolk Syndrome. <i>Molecular Syndromology</i> , 0, , 1-6.	0.3	0
2466	Analysis of recent shared ancestry in a familial cohort identifies coding and noncoding autism spectrum disorder variants. <i>Npj Genomic Medicine</i> , 2022, 7, 13.	1.7	18
2467	An Evaluation for the Causes of Reduced Hb A ₂ and the Molecular Characterization of <i>HBD</i> Variants in Hong Kong. <i>Hemoglobin</i> , 2022, , 1-5.	0.4	2
2469	Evaluation of the genetic risk for COVID-19 outcomes in COPD and differences among worldwide populations. <i>PLoS ONE</i> , 2022, 17, e0264009.	1.1	11
2470	An omic and multidimensional spatial atlas from serial biopsies of an evolving metastatic breast cancer. <i>Cell Reports Medicine</i> , 2022, 3, 100525.	3.3	22
2471	Discovery of an unusually high number of de novo mutations in sperm of older men using duplex sequencing. <i>Genome Research</i> , 2022, 32, 499-511.	2.4	9
2472	A human breast cancer-derived xenograft and organoid platform for drug discovery and precision oncology. <i>Nature Cancer</i> , 2022, 3, 232-250.	5.7	133
2473	No preferential mode of inheritance for highly constrained genes. <i>Intractable and Rare Diseases Research</i> , 2022, 11, 25-28.	0.3	4
2474	Patterns of molar agenesis associated with p.P20L and p.R77Q variants in <i>PAX9</i> . <i>European Journal of Oral Sciences</i> , 2022, 130, e12855.	0.7	3
2475	<i>Drosophila</i> carrying epilepsy-associated variants in the vitamin B6 metabolism gene <i>PNPO</i> display allele- and diet-dependent phenotypes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	6
2477	Clinical exome sequencing—Mistakes and caveats. <i>Human Mutation</i> , 2022, 43, 1041-1055.	1.1	20
2478	P2X2 receptor subunit interfaces are missense variant hotspots, where mutations tend to increase apparent ATP affinity. <i>British Journal of Pharmacology</i> , 2022, 179, 3859-3874.	2.7	1
2479	Mutations in <i>RNU7-1</i> Weaken Secondary RNA Structure, Induce MCP-1 and CXCL10 in CSF, and Result in Aicardi-Goutières Syndrome with Severe End-Organ Involvement. <i>Journal of Clinical Immunology</i> , 2022, 42, 962-974.	2.0	8
2482	Lifetime risk of autosomal recessive neurodegeneration with brain iron accumulation (NBIA) disorders calculated from genetic databases. <i>EBioMedicine</i> , 2022, 77, 103869.	2.7	11
2483	Structural mapping of <i>GABRB3</i> variants reveals genotype–phenotype correlations. <i>Genetics in Medicine</i> , 2022, 24, 681-693.	1.1	10
2484	Pharmacogenomic landscape of Indian population using whole genomes. <i>Clinical and Translational Science</i> , 2022, 15, 866-877.	1.5	8

#	ARTICLE	IF	CITATIONS
2487	3q27.1 microdeletion causes prenatal and postnatal growth restriction and neurodevelopmental abnormalities. <i>Molecular Cytogenetics</i> , 2022, 15, 7.	0.4	1
2489	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1283-1296.	1.1	9
2491	Genomic answers for children: Dynamic analyses of >1000 pediatric rare disease genomes. <i>Genetics in Medicine</i> , 2022, 24, 1336-1348.	1.1	37
2494	A Brief Review of Machine Learning-Based Bioactive Compound Research. <i>Applied Sciences (Switzerland)</i> , 2022, 12, 2906.	1.3	3
2495	Design of a targeted next-generation DNA sequencing panel for pediatric T-cell lymphoblastic lymphoma to unravel biology and optimize treatment. <i>Genes Chromosomes and Cancer</i> , 2022, 61, 459-470.	1.5	2
2496	Pre- and Postnatal Characterization of Autosomal Recessive <i>KIDINS220</i>-Associated Ventriculomegaly. <i>Molecular Syndromology</i> , 2022, 13, 419-424.	0.3	1
2497	Arrhythmia Variant Associations and Reclassifications in the eMERGE-III Sequencing Study. <i>Circulation</i> , 2022, 145, 877-891.	1.6	18
2498	Constrained human genes under scrutiny. <i>Nature</i> , 2022, 603, 799-801.	13.7	0
2499	The implication of holocytochrome c synthase mutation in Korean familial hypoplastic amelogenesis imperfecta. <i>Clinical Oral Investigations</i> , 2022, 26, 4487-4498.	1.4	1
2500	A review of the genetic spectrum of hereditary spastic paraplegias, inherited neuropathies and spinal muscular atrophies in Africans. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 133.	1.2	4
2501	Novel genes bearing mutations in rare cases of early-onset ataxia with cerebellar hypoplasia. <i>European Journal of Human Genetics</i> , 2022, 30, 703-711.	1.4	3
2502	A general framework for identifying oligogenic combinations of rare variants in complex disorders. <i>Genome Research</i> , 2022, , .	2.4	7
2503	A novel synonymous <sc><i>KMT2B</i></sc> variant in a patient with dystonia causes aberrant splicing. <i>Molecular Genetics & Genomic Medicine</i> , 2022, , e1923.	0.6	2
2504	Genetic Evaluation of Late-Onset Hypertrophic Cardiomyopathy: An Autobiographical Case Report. <i>Cureus</i> , 2022, 14, e23349.	0.2	1
2505	Monocyte secretory profiling in a clinical and MEFV genotype-characterized cohort of Danish familial Mediterranean fever patients: diagnostic potential of CCL1 and CXCL1.. <i>Scandinavian Journal of Rheumatology</i> , 2023, 52, 181-189.	0.6	1
2506	Mammalian HEMK1 methylates glutamine residue of the GGQ motif of mitochondrial release factors. <i>Scientific Reports</i> , 2022, 12, 4104.	1.6	2
2507	<sc>Elâ€Hattabâ€Alkuraya</sc> syndrome caused by biallelic <sc><i>WDR45B</i></sc> pathogenic variants: Further delineation of the phenotype and genotype. <i>Clinical Genetics</i> , 2022, 101, 530-540.	1.0	7
2508	Collision of germline POLE and PMS2 variants in a young patient treated with immune checkpoint inhibitors. <i>Npj Precision Oncology</i> , 2022, 6, 15.	2.3	11

#	ARTICLE	IF	CITATIONS
2509	Whole-exome sequencing in a Japanese multiplex family identifies new susceptibility genes for intracranial aneurysms. <i>PLoS ONE</i> , 2022, 17, e0265359.	1.1	1
2511	Transcript levels in plasma contribute substantial predictive value as potential Alzheimer's disease biomarkers in African Americans. <i>EBioMedicine</i> , 2022, , 103929.	2.7	2
2512	Identification of three novel homozygous variants in COL9A3 causing autosomal recessive Stickler syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 97.	1.2	3
2513	Spectrum and frequency of CHEK2 variants in breast cancer affected and general population in the Baltic states region, initial results and literature review. <i>European Journal of Medical Genetics</i> , 2022, 65, 104477.	0.7	5
2514	Expanding the phenotypic spectrum of ARCN1-related syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1227-1237.	1.1	5
2515	Characterization of intrinsically disordered regions in proteins informed by human genetic diversity. <i>PLoS Computational Biology</i> , 2022, 18, e1009911.	1.5	13
2516	GENESIS: Gene-Specific Machine Learning Models for Variants of Uncertain Significance Found in Catecholaminergic Polymorphic Ventricular Tachycardia and Long QT Syndrome-Associated Genes. <i>Circulation: Arrhythmia and Electrophysiology</i> , 2022, 15, 101161CIRCEP121010326.	2.1	17
2517	High-throughput characterization of the role of non-B DNA motifs on promoter function. <i>Cell Genomics</i> , 2022, 2, 100111.	3.0	17
2518	Cross-ethnic analysis of common gene variants in hemostasis show lopsided representation of global populations in genetic databases. <i>BMC Medical Genomics</i> , 2022, 15, 69.	0.7	0
2519	Identification of a novel MAGT1 mutation supports a diagnosis of XMEN disease. <i>Genes and Immunity</i> , 2022, 23, 66-72.	2.2	8
2521	Clinical and Genetic Characteristics of Finnish Patients with Autosomal Recessive and Dominant Non-Syndromic Hearing Loss Due to Pathogenic TMC1 Variants. <i>Journal of Clinical Medicine</i> , 2022, 11, 1837.	1.0	2
2522	TADA—a machine learning tool for functional annotation-based prioritisation of pathogenic CNVs. <i>Genome Biology</i> , 2022, 23, 67.	3.8	4
2523	Synonymous mutation in adenosine triphosphatase copper-transporting beta causes enhanced exon skipping in Wilson disease. <i>Hepatology Communications</i> , 2022, 6, 1611-1619.	2.0	6
2524	Arginase 1 Deficiency: using genetic databases as a tool to establish global prevalence. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 94.	1.2	7
2525	Pervasive occurrence of splice-site-creating mutations and their possible involvement in genetic disorders. <i>Npj Genomic Medicine</i> , 2022, 7, 22.	1.7	3
2526	PLIN1 Haploinsufficiency Causes a Favorable Metabolic Profile. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, e2318-e2323.	1.8	7
2527	Significant sparse polygenic risk scores across 813 traits in UK Biobank. <i>PLoS Genetics</i> , 2022, 18, e1010105.	1.5	40
2528	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. <i>Cell Reports</i> , 2022, 38, 110517.	2.9	24

#	ARTICLE	IF	CITATIONS
2529	Genome-wide linkage analysis combined with genome sequencing in large families with intracranial aneurysms. <i>European Journal of Human Genetics</i> , 2022, 30, 833-840.	1.4	2
2530	RevUP: an online scoring system for regulatory variants implicated in rare diseases. <i>Bioinformatics</i> , 2022, 38, 2664-2666.	1.8	0
2531	Comprehensive RNA dataset of tissue and plasma from patients with esophageal cancer or precursor lesions. <i>Scientific Data</i> , 2022, 9, 86.	2.4	1
2532	Reduced reproductive success is associated with selective constraint on human genes. <i>Nature</i> , 2022, 603, 858-863.	13.7	29
2533	Associations of common genetic risk variants of the muscarinic acetylcholine receptor M2 with cardiac autonomic dysfunction in patients with schizophrenia. <i>World Journal of Biological Psychiatry</i> , 2022, , 1-11.	1.3	1
2535	Genetic Influence on Neurodevelopment in Nonsyndromic Craniosynostosis. <i>Plastic and Reconstructive Surgery</i> , 2022, 149, 1157-1165.	0.7	10
2536	Comprehensive analysis of recessive carrier status using exome and genome sequencing data in 1543 Southern Chinese. <i>Npj Genomic Medicine</i> , 2022, 7, 23.	1.7	6
2537	Case Report: Next-Generation Sequencing Identified a Novel Pair of Compound-Heterozygous Mutations of LPL Gene Causing Lipoprotein Lipase Deficiency. <i>Frontiers in Genetics</i> , 2022, 13, 831133.	1.1	2
2538	Systematic estimation of cystic fibrosis prevalence in Chinese and genetic spectrum comparison to Caucasians. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 129.	1.2	17
2540	The burden of rare protein-truncating genetic variants on human lifespan. <i>Nature Aging</i> , 2022, 2, 289-294.	5.3	6
2541	The Role of <i>GJD2</i> (<i>Cx36</i>) in Refractive Error Development. , 2022, 63, 5.		3
2542	Longitudinal analysis of electroencephalography pattern changes in an infant with <i>Schaaf-ÿang</i> syndrome and a novel mutation in <i>melanoma antigen L2</i> (<i>MAGEL2</i>) <i>Tj ETQp1 1 0.784314 rgBT</i>	1.1	1
2543	Current Status of Next-Generation Sequencing Approaches for Candidate Gene Discovery in Familial Parkinson's Disease. <i>Frontiers in Genetics</i> , 2022, 13, 781816.	1.1	3
2544	Predicting embryonic aneuploidy rate in IVF patients using whole-exome sequencing. <i>Human Genetics</i> , 2022, 141, 1615-1627.	1.8	9
2546	Dominant negative effects of <i>SCN5A</i> missense variants. <i>Genetics in Medicine</i> , 2022, 24, 1238-1248.	1.1	9
2547	Venus: Elucidating the Impact of Amino Acid Variants on Protein Function Beyond Structure Destabilisation. <i>Journal of Molecular Biology</i> , 2022, 434, 167567.	2.0	13
2549	A saturation-mutagenesis analysis of the interplay between stability and activation in Ras. <i>ELife</i> , 2022, 11, .	2.8	13
2550	Sequencing of <i>BRCA1/2</i> -alterations using NGS-based technology: annotation as a challenge. <i>Oncotarget</i> , 2022, 13, 464-475.	0.8	0

#	ARTICLE	IF	CITATIONS
2553	Whole-genome sequencing reveals host factors underlying critical COVID-19. <i>Nature</i> , 2022, 607, 97-103.	13.7	174
2554	Evaluating a Causal Relationship between Complement Factor I Protein Level and Advanced Age-Related Macular Degeneration Using Mendelian Randomization. <i>Ophthalmology Science</i> , 2022, 2, 100146.	1.0	6
2555	De novo variants in ATP2B1 lead to neurodevelopmental delay. <i>American Journal of Human Genetics</i> , 2022, 109, 944-952.	2.6	11
2556	Need for Inclusive Genomic Research. <i>Circulation Genomic and Precision Medicine</i> , 2022, , CIRCGEN122003736.	1.6	0
2558	Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing. <i>Nature Biotechnology</i> , 2022, 40, 1035-1041.	9.4	45
2560	Region-based analysis of rare genomic variants in whole-genome sequencing datasets reveal two novel Alzheimer's disease-associated genes: DTNB and DLG2. <i>Molecular Psychiatry</i> , 2022, 27, 1963-1969.	4.1	9
2562	Rare and common variants in ROM1 and PRPH2 genes trans-modify Stargardt/ABCA4 disease. <i>PLoS Genetics</i> , 2022, 18, e1010129.	1.5	8
2563	Classification of KRAS-Activating Mutations and the Implications for Therapeutic Intervention. <i>Cancer Discovery</i> , 2022, 12, 913-923.	7.7	32
2565	Whole-genome sequencing of 1,171 elderly admixed individuals from Brazil. <i>Nature Communications</i> , 2022, 13, 1004.	5.8	35
2566	Pathogenic variants in MDL1 cause recessive central conducting lymphatic anomaly with lymphedema. <i>Science Translational Medicine</i> , 2022, 14, eabm4869.	5.8	14
2567	Consolidation of the clinical and genetic definition of a SOX4-related neurodevelopmental syndrome. <i>Journal of Medical Genetics</i> , 2022, 59, 1058-1068.	1.5	10
2569	Whole exome analysis of patients in Japan with hearing loss reveals high heterogeneity among responsible and novel candidate genes. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 114.	1.2	3
2571	Genetic alteration of human MYH6 is mimicked by SARS-CoV-2 polyprotein: mapping viral variants of cardiac interest. <i>Cell Death Discovery</i> , 2022, 8, 124.	2.0	4
2572	Functional analysis of a novel de novo variant in PPP5C associated with microcephaly, seizures, and developmental delay. <i>Molecular Genetics and Metabolism</i> , 2022, 136, 65-73.	0.5	4
2573	The proteogenomic subtypes of acute myeloid leukemia. <i>Cancer Cell</i> , 2022, 40, 301-317.e12.	7.7	43
2574	Hornerin deposits in neuronal intranuclear inclusion disease: direct identification of proteins with compositionally biased regions in inclusions. <i>Acta Neuropathologica Communications</i> , 2022, 10, 28.	2.4	4
2575	Whole genome sequences discriminate hereditary hemorrhagic telangiectasia phenotypes by non-HHT deleterious DNA variation. <i>Blood Advances</i> , 2022, 6, 3956-3969.	2.5	9
2576	Novel mutations in the HADHB gene causing a mild phenotype of mitochondrial trifunctional protein (MTP) deficiency. <i>JIMD Reports</i> , 2022, 63, 193-198.	0.7	1

#	ARTICLE	IF	CITATIONS
2578	Extrahepatic manifestations of progressive familial intrahepatic cholestasis syndromes: Presentation of a case series and literature review. <i>Liver International</i> , 2022, 42, 1084-1096.	1.9	7
2579	A clinical laboratory's experience using GeneMatcher®"Building stronger gene-disease relationships. <i>Human Mutation</i> , 2022, , .	1.1	3
2580	Novel Pathogenic Sequence Variation m.5789T>C Causes NARP Syndrome and Promotes Formation of Deletions of the Mitochondrial Genome. <i>Neurology: Genetics</i> , 2022, 8, e660.	0.9	3
2582	Whole-Genome Sequencing Identifies Novel Heterozygous Mutation in ALMS1 in Three Men With Both Peyronie's and Dupuytren's Disease. <i>Urology</i> , 2022, 166, 76-78.	0.5	3
2583	Whole-genome sequencing reveals novel ethnicity-specific rare variants associated with Alzheimer's disease. <i>Molecular Psychiatry</i> , 2022, 27, 2554-2562.	4.1	14
2586	Neonatal Arterial Tortuosity and Adult Aortic Aneurysm"Is There a Missing Link?"A Case Report. <i>Frontiers in Pediatrics</i> , 2021, 9, 814773.	0.9	2
2588	Oral Adrenergic Agents Produced Ventricular Fibrillation and QT Prolongation in an Elderly Patient Carrying an <i>RYR2</i> Variant. <i>International Heart Journal</i> , 2022, 63, 398-403.	0.5	0
2589	Ultrasensitive profiling of UV-induced mutations identifies thousands of subclinical facial tumors in tuberous sclerosis complex. <i>Journal of Clinical Investigation</i> , 2022, 132, .	3.9	6
2590	Causes and Pathogenesis of Malignant Mesothelioma. , 0, , .		2
2591	Molecular Modeling is an Enabling Approach to Complement and Enhance Channelopathy Research. , 2022, 12, 3141-3166.		0
2592	Complex Autism Spectrum Disorder with Epilepsy, Strabismus and Self-Injurious Behaviors in a Patient with a De Novo Heterozygous POLR2A Variant. <i>Genes</i> , 2022, 13, 470.	1.0	3
2593	Extending the prenatal Noonan's phenotype by review of ultrasound and autopsy data. <i>Prenatal Diagnosis</i> , 2022, 42, 574-582.	1.1	4
2594	A Genome-Wide Association Study and Machine-Learning Algorithm Analysis on the Prediction of Facial Phenotypes by Genotypes in Korean Women. <i>Clinical, Cosmetic and Investigational Dermatology</i> , 2022, Volume 15, 433-445.	0.8	1
2595	Fibrillar Collagen Variants in Spontaneous Coronary Artery Dissection. <i>JAMA Cardiology</i> , 2022, 7, 396.	3.0	19
2597	MEN4, the MEN1 Mimicker: A Case Series of three Phenotypically Heterogenous Patients With Unique <i>CDKN1B</i> Mutations. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 2339-2349.	1.8	14
2598	Single-cell landscapes of primary glioblastomas and matched explants and cell lines show variable retention of inter- and intratumor heterogeneity. <i>Cancer Cell</i> , 2022, 40, 379-392.e9.	7.7	54
2599	Defining novel causal SNPs and linked phenotypes at melanoma-associated loci. <i>Human Molecular Genetics</i> , 2022, 31, 2845-2856.	1.4	3
2600	Structural Plasticity Is a Feature of Rheostat Positions in the Human Na ⁺ /Taurocholate Cotransporting Polypeptide (NTCP). <i>International Journal of Molecular Sciences</i> , 2022, 23, 3211.	1.8	4

#	ARTICLE	IF	CITATIONS
2601	Characteristic Immune Dynamics in COVID-19 Patients with Cardiac Dysfunction. <i>Journal of Clinical Medicine</i> , 2022, 11, 1880.	1.0	3
2602	Genome-wide analysis of haploinsufficiency in human embryonic stem cells. <i>Cell Reports</i> , 2022, 38, 110573.	2.9	4
2603	A novel <i>GNAS</i> variant presents with disorders of <i>GNAS</i> inactivation and cardiomyopathy. <i>American Journal of Medical Genetics, Part A</i> , 2022, , .	0.7	0
2604	Whole-exome DNA sequencing in childhood anxiety disorders identifies rare de novo damaging coding variants. <i>Depression and Anxiety</i> , 2022, 39, 474-484.	2.0	5
2605	Variant-level matching for diagnosis and discovery: Challenges and opportunities. <i>Human Mutation</i> , 2022, , .	1.1	11
2606	Performance Comparison of Computational Methods for the Prediction of the Function and Pathogenicity of Non-Coding Variants. <i>Genomics, Proteomics and Bioinformatics</i> , 2023, 21, 649-661.	3.0	7
2607	Whole-genome analysis of human embryonic stem cells enables rational line selection based on genetic variation. <i>Cell Stem Cell</i> , 2022, 29, 472-486.e7.	5.2	27
2608	Possible Catch-Up Developmental Trajectories for Children with Mild Developmental Delay Caused by NAA15 Pathogenic Variants. <i>Genes</i> , 2022, 13, 536.	1.0	3
2609	Enrichment of Motilin Receptor Loss-of-Function Variants in Gastroparesis. <i>Clinical and Translational Gastroenterology</i> , 2022, 13, e00474.	1.3	3
2610	Whole exome sequencing of pediatric leukemia reveals a novel InDel within FLT-3 gene in AML patient from Mizo tribal population, Northeast India. <i>BMC Genomic Data</i> , 2022, 23, 23.	0.7	4
2611	Biallelic variants in <i>TTC21B</i> as a rare cause of early-onset arterial hypertension and tubuloglomerular kidney disease. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2022, 190, 109-120.	0.7	6
2612	Contiguously hydrophobic sequences are functionally significant throughout the human exome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, e2116267119.	3.3	1
2614	Identification of New Vulnerabilities in Conjunctival Melanoma Using Image-Based High Content Drug Screening. <i>Cancers</i> , 2022, 14, 1575.	1.7	0
2615	Multicenter Consensus Approach to Evaluation of Neonatal Hypotonia in the Genomic Era: A Review. <i>JAMA Neurology</i> , 2022, 79, 405.	4.5	7
2617	Novel dominant and recessive variants in human <i>ROBO1</i> cause distinct neurodevelopmental defects through different mechanisms. <i>Human Molecular Genetics</i> , 2022, 31, 2751-2765.	1.4	3
2620	Molecular Properties of Human Guanylate Cyclase-Activating Protein 3 (GCAP3) and Its Possible Association with Retinitis Pigmentosa. <i>International Journal of Molecular Sciences</i> , 2022, 23, 3240.	1.8	3
2623	A comprehensive WGS-based pipeline for the identification of new candidate genes in inherited retinal dystrophies. <i>Npj Genomic Medicine</i> , 2022, 7, 17.	1.7	7
2624	Pan-human consensus genome significantly improves the accuracy of RNA-seq analyses. <i>Genome Research</i> , 2022, 32, 738-749.	2.4	6

#	ARTICLE	IF	CITATIONS
2625	Federated analysis of BRCA1 and BRCA2 variation in a Japanese cohort. <i>Cell Genomics</i> , 2022, 2, 100109.	3.0	1
2626	Whole genome sequencing delineates regulatory, copy number, and cryptic splice variants in early onset cardiomyopathy. <i>Npj Genomic Medicine</i> , 2022, 7, 18.	1.7	14
2627	Genome Alert!: A standardized procedure for genomic variant reinterpretation and automated geneâ€œphenotype reassessment in clinical routine. <i>Genetics in Medicine</i> , 2022, 24, 1316-1327.	1.1	5
2628	Inherited and De Novo Variation in Lithuanian Genomes: Introduction to the Analysis of the Generational Shift. <i>Genes</i> , 2022, 13, 569.	1.0	3
2629	Combining familial hypercholesterolemia and statin genetic studies as a strategy for the implementation of pharmacogenomics. A multidisciplinary approach. <i>Pharmacogenomics Journal</i> , 2022, , .	0.9	0
2630	<i>BAG3</i> Genetic Cardiomyopathy May Overlap Fulminant Myocarditis Clinical Findings. <i>Circulation: Heart Failure</i> , 2022, 15, e008443.	1.6	1
2631	Transcription factor protein interactomes reveal genetic determinants in heart disease. <i>Cell</i> , 2022, 185, 794-814.e30.	13.5	39
2632	Middle Eastern Genetic Variation Improves Clinical Annotation of the Human Genome. <i>Journal of Personalized Medicine</i> , 2022, 12, 423.	1.1	7
2633	Red Herring Pathogenic Variants: A Case Report of Premature Ventricular Contraction-Triggered Ventricular Fibrillation with an Incidental Pathogenic <i>LMNA</i> Variant. <i>European Heart Journal - Case Reports</i> , 2022, 6, ytac115.	0.3	0
2634	Comprehensive characterization of PTEN mutational profile in a series of 34,129 colorectal cancers. <i>Nature Communications</i> , 2022, 13, 1618.	5.8	23
2635	Germline Variants in Cancer Genes from Young Breast Cancer Mexican Patients. <i>Cancers</i> , 2022, 14, 1647.	1.7	5
2636	An orally available, brain penetrant, small molecule lowers huntingtin levels by enhancing pseudoexon inclusion. <i>Nature Communications</i> , 2022, 13, 1150.	5.8	58
2638	De novo variants in <i>EMC1</i> lead to neurodevelopmental delay and cerebellar degeneration and affect glial function in <i>Drosophila</i> . <i>Human Molecular Genetics</i> , 2022, 31, 3231-3244.	1.4	5
2641	PTCHD1: Identification and Neurodevelopmental Contributions of an Autism Spectrum Disorder and Intellectual Disability Susceptibility Gene. <i>Genes</i> , 2022, 13, 527.	1.0	7
2642	Validation of New Gene Variant Classification Methods: a Field-Test in Diagnostic Cardiogenetics. <i>Frontiers in Genetics</i> , 2022, 13, 824510.	1.1	1
2643	Selective requirement for polycomb repressor complex 2 in the generation of specific hypothalamic neuronal subtypes. <i>Development (Cambridge)</i> , 2022, 149, .	1.2	4
2644	Expanding the clinical spectrum of COL2A1 related disorders by a mass like phenotype. <i>Scientific Reports</i> , 2022, 12, 4489.	1.6	2
2645	CFTR bearing variant p.Phe312del exhibits function inconsistent with phenotype and negligible response to ivacaftor. <i>JCI Insight</i> , 2022, 7, .	2.3	3

#	ARTICLE	IF	CITATIONS
2646	Functional Characterization of the MYO6 Variant p.E60Q in Non-Syndromic Hearing Loss Patients. <i>International Journal of Molecular Sciences</i> , 2022, 23, 3369.	1.8	1
2647	Gain-of-function and loss-of-function GABRB3 variants lead to distinct clinical phenotypes in patients with developmental and epileptic encephalopathies. <i>Nature Communications</i> , 2022, 13, 1822.	5.8	32
2648	Novel truncating variants in <i>FGD1</i> detected in two Danish families with Aarskog-Scott syndrome and myopathic features. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2251-2257.	0.7	3
2649	Analyzing human knockouts to validate GPR151 as a therapeutic target for reduction of body mass index. <i>PLoS Genetics</i> , 2022, 18, e1010093.	1.5	1
2650	Use of machine learning to classify high-risk variants of uncertain significance in lamin A/C cardiac disease. <i>Heart Rhythm</i> , 2022, 19, 676-685.	0.3	3
2651	Phenotype Expansion for Atypical Gaucher Disease Due to Homozygous Missense PSAP Variant in a Large Consanguineous Pakistani Family. <i>Genes</i> , 2022, 13, 662.	1.0	3
2652	Population-based estimates of age-specific cumulative risk of breast cancer for pathogenic variants in ATM. <i>Breast Cancer Research</i> , 2022, 24, 24.	2.2	3
2653	A year of COVID-19 GWAS results from the GRASP portal reveals potential genetic risk factors. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100095.	1.0	21
2655	Seven Years of Selective Genetic Screening Program and Follow-Up of Asymptomatic Carriers With Hereditary Transthyretin Amyloidosis in Bulgaria. <i>Frontiers in Neurology</i> , 2022, 13, 844595.	1.1	3
2656	Mutation profiling of the c.1521_1523delCTT (p.Phe508del, F508del) cystic fibrosis transmembrane conductance regulator allele using haplotype-resolved long-read next generation sequencing. <i>Human Mutation</i> , 2022, 43, 595-603.	1.1	1
2657	Germline mosaicism of a missense variant in <i>KCNC2</i> in a multiplex family with autism and epilepsy characterized by long-read sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2071-2081.	0.7	7
2658	Exome sequencing of individuals with Huntington's disease implicates FAN1 nuclease activity in slowing CAG expansion and disease onset. <i>Nature Neuroscience</i> , 2022, 25, 446-457.	7.1	31
2659	Molecular pathogenesis and heterogeneity in type 3 VWD families in U.S. Zimmerman program. <i>Journal of Thrombosis and Haemostasis</i> , 2022, 20, 1576-1588.	1.9	5
2660	Lack of association of TP73 with amyotrophic lateral sclerosis in a large cohort of cases. <i>Neurobiology of Aging</i> , 2022, 115, 109-111.	1.5	2
2661	Artificial intelligence informed toxicity screening of amine chemistries used in the synthesis of hybrid organic-inorganic perovskites. <i>AIChE Journal</i> , 2022, 68, .	1.8	1
2662	Single-cell Atlas of common variable immunodeficiency shows germinal center-associated epigenetic dysregulation in B-cell responses. <i>Nature Communications</i> , 2022, 13, 1779.	5.8	25
2663	<i>BRCA2</i> promoter hypermethylation as a biomarker for the leukemic transformation of myeloproliferative neoplasms. <i>Epigenomics</i> , 2022, 14, 391-403.	1.0	1
2664	De novo heterozygous variants in <i>KIF5B</i> cause kyphomelic dysplasia. <i>Clinical Genetics</i> , 2022, 102, 3-11.	1.0	5

#	ARTICLE	IF	CITATIONS
2665	Identification and Management of Pathogenic Variants in <i>BRCA1</i> , <i>BRCA2</i> , and <i>PALB2</i> in a Tumor-Only Genomic Testing Program. <i>Clinical Cancer Research</i> , 2022, 28, 2349-2360.	3.2	8
2666	Pseudohypoaldosteronism Type 1: The Presentation and Management of a Neonate With a Novel Mutation of the <i>SCNN1B</i> Gene Found in Two Hispanic Siblings. <i>Cureus</i> , 2022, 14, e23918.	0.2	1
2667	A joint NCBI and EMBL-EBI transcript set for clinical genomics and research. <i>Nature</i> , 2022, 604, 310-315.	13.7	162
2670	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. <i>Genome Medicine</i> , 2022, 14, 38.	3.6	85
2671	Clinical Phenotypes and Outcomes in Monogenic Versus Non-monogenic Very Early Onset Inflammatory Bowel Disease. <i>Journal of Crohn's and Colitis</i> , 2022, 16, 1380-1396.	0.6	19
2672	Detecting drug resistance in pancreatic cancer organoids guides optimized chemotherapy treatment. <i>Journal of Pathology</i> , 2022, 257, 607-619.	2.1	13
2673	A complete reference genome improves analysis of human genetic variation. <i>Science</i> , 2022, 376, eabl3533.	6.0	144
2674	Integrative genomic analysis reveals low T-cell infiltration as the primary feature of tobacco use in HPV-positive oropharyngeal cancer. <i>IScience</i> , 2022, 25, 104216.	1.9	6
2675	Genome-wide analysis of somatic noncoding mutation patterns in cancer. <i>Science</i> , 2022, 376, eabg5601.	6.0	33
2677	STRipy: A graphical application for enhanced genotyping of pathogenic short tandem repeats in sequencing data. <i>Human Mutation</i> , 2022, 43, 859-868.	1.1	11
2678	Rare coding variants in ten genes confer substantial risk for schizophrenia. <i>Nature</i> , 2022, 604, 509-516.	13.7	326
2679	Preserved cardiac performance and adrenergic response in a rabbit model with decreased ryanodine receptor 2 expression. <i>Journal of Molecular and Cellular Cardiology</i> , 2022, 167, 118-128.	0.9	5
2680	Functional investigation of two simultaneous or separately segregating <i>DSP</i> variants within a single family supports the theory of a dose-dependent disease severity. <i>Experimental Dermatology</i> , 2022, , .	1.4	3
2681	Rare and Common Variants Uncover the Role of the Atria in Coarctation of the Aorta. <i>Genes</i> , 2022, 13, 636.	1.0	4
2682	A next-generation human genome sequence. <i>Science</i> , 2022, 376, 34-35.	6.0	11
2683	Genetic obstacles to developing and tolerizing human B cells. <i>WIREs Mechanisms of Disease</i> , 2022, 14, e1554.	1.5	1
2684	SWAN pathway-network identification of common aneuploidy-based oncogenic drivers. <i>Nucleic Acids Research</i> , 2022, 50, 3673-3692.	6.5	10
2685	16p13.11p11.2 triplication syndrome: a new recognizable genomic disorder characterized by optical genome mapping and whole genome sequencing. <i>European Journal of Human Genetics</i> , 2022, , .	1.4	5

#	ARTICLE	IF	CITATIONS
2686	Monogenic and Polygenic Contributions to QTc Prolongation in the Population. <i>Circulation</i> , 2022, 145, 1524-1533.	1.6	14
2687	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2022, 109, 750-758.	2.6	13
2688	Uncompensated mitochondrial oxidative stress underlies heart failure in an iPSC-derived model of congenital heart disease. <i>Cell Stem Cell</i> , 2022, 29, 840-855.e7.	5.2	18
2689	AOPEP variants as a novel cause of recessive dystonia: Generalized dystonia and dystonia-parkinsonism. <i>Parkinsonism and Related Disorders</i> , 2022, 97, 52-56.	1.1	7
2691	The individual and global impact of copy-number variants on complex human traits. <i>American Journal of Human Genetics</i> , 2022, 109, 647-668.	2.6	31
2692	Bioinformatic Challenges Detecting Genetic Variation in Precision Medicine Programs. <i>Frontiers in Medicine</i> , 2022, 9, 806696.	1.2	3
2693	Searching thousands of genomes to classify somatic and novel structural variants using STIX. <i>Nature Methods</i> , 2022, 19, 445-448.	9.0	8
2694	Diagnostic yield using whole-genome sequencing and <i>in silico</i> panel of 281 genes associated with non-immune hydrops fetalis in clinical setting. <i>Ultrasound in Obstetrics and Gynecology</i> , 2022, 60, 487-493.	0.9	11
2695	RAREsim: A simulation method for very rare genetic variants. <i>American Journal of Human Genetics</i> , 2022, 109, 680-691.	2.6	1
2696	Partitioning gene-level contributions to complex-trait heritability by allele frequency identifies disease-relevant genes. <i>American Journal of Human Genetics</i> , 2022, 109, 692-709.	2.6	2
2697	Loss-of-function variants in TIAM1 are associated with developmental delay, intellectual disability, and seizures. <i>American Journal of Human Genetics</i> , 2022, 109, 571-586.	2.6	19
2698	Phenotype-driven approaches to enhance variant prioritization and diagnosis of rare disease. <i>Human Mutation</i> , 2022, 43, 1071-1081.	1.1	17
2699	Novel missense SETD1A variants in Japanese patients with schizophrenia: Resequencing and association analysis. <i>Psychiatry Research</i> , 2022, 310, 114481.	1.7	2
2700	A novel, de novo intronic variant in <i>POGZ</i> causes White-Sutton syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2198-2203.	0.7	4
2701	Status Epilepticus due to Asfotase Alfa Interruption in Perinatal Severe Hypophosphatasia. <i>Pediatric Neurology</i> , 2022, 130, 4-6.	1.0	1
2702	Missense Variants Reveal Functional Insights Into the Human ARID Family of Gene Regulators. <i>Journal of Molecular Biology</i> , 2022, 434, 167529.	2.0	6
2703	Whole-genome sequencing identifies rare missense variants of WNT16 and ERVW-1 causing the systemic lupus erythematosus. <i>Genomics</i> , 2022, 114, 110332.	1.3	9
2704	Compound heterozygote variants: c.848A>G; p.Glu283Gly and c.890C>A; p.Ala297Val, of Isovaleric acid-CoA dehydrogenase (IVD) gene causing severe Isovaleric acidemia with hyperammonemia. <i>Molecular Genetics and Metabolism Reports</i> , 2022, 31, 100859.	0.4	0

#	ARTICLE	IF	CITATIONS
2705	Integrated bioinformatic pipeline using whole-exome and RNAseq data to identify germline variants correlated with cancer. STAR Protocols, 2022, 3, 101273.	0.5	0
2706	Neovascularization of the optic disc and peripheral retinal ischemia in a child with a novel variant in ALMS1 (Alström syndrome). American Journal of Ophthalmology Case Reports, 2022, 26, 101506.	0.4	0
2707	Assessment of 13 in silico pathogenicity methods on cancer-related variants. Computers in Biology and Medicine, 2022, 145, 105434.	3.9	2
2708	Dominant osteogenesis imperfecta with low bone turnover caused by a heterozygous SP7 variant. Bone, 2022, 160, 116400.	1.4	10
2709	Modeling Human Genetic Disorders with CRISPR Technologies in <i>Xenopus</i> . Cold Spring Harbor Protocols, 2022, 2022, pdb.prot106997.	0.2	5
2710	Assessing Prevalence and Carrier Frequency of Succinic Semialdehyde Dehydrogenase Deficiency. Journal of Child Neurology, 2021, 36, 1218-1222.	0.7	7
2711	Variant Library Annotation Tool (VaLiAnT): an oligonucleotide library design and annotation tool for saturation genome editing and other deep mutational scanning experiments. Bioinformatics, 2022, 38, 892-899.	1.8	3
2712	Interpreting ciliopathy-associated missense variants of uncertain significance (VUS) in <i>Caenorhabditis elegans</i> . Human Molecular Genetics, 2022, 31, 1574-1587.	1.4	9
2714	Predicting disease variants using biodiversity and machine learning. Nature Biotechnology, 2022, 40, 27-28.	9.4	3
2716	Epithelioid cell granuloma formation in <i>CARD14</i> -associated papulosquamous eruptions. Journal of the European Academy of Dermatology and Venereology, 2022, 36, .	1.3	0
2717	A Nationwide Study of GATA2 Deficiency in Norway—the Majority of Patients Have Undergone Allo-HSCT. Journal of Clinical Immunology, 2022, 42, 404-420.	2.0	10
2718	Sleep Polygenic Risk Score Is Associated with Cognitive Changes over Time. Genes, 2022, 13, 63.	1.0	5
2719	<i>EFEMP1</i> rare variants cause familial juvenile-onset open-angle glaucoma. Human Mutation, 2022, 43, 240-252.	1.1	19
2720	Silent variant in <i>F8</i> :c.222G>T (p.Thr74Thr) causes a partial exon skipping in a patient with mild hemophilia A. Molecular Genetics & Genomic Medicine, 2022, 10, e1856.	0.6	1
2721	High-impact rare genetic variants in severe schizophrenia. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	29
2722	The good, the bad, and the ugly: Evolutionary and pathological aspects of gene dosage alterations. PLoS Genetics, 2021, 17, e1009906.	1.5	5
2725	Domestication reshaped the genetic basis of inbreeding depression in a maize landrace compared to its wild relative, teosinte. PLoS Genetics, 2021, 17, e1009797.	1.5	5
2726	Systems biology analysis of human genomes points to key pathways conferring spina bifida risk. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	11

#	ARTICLE	IF	CITATIONS
2727	dbTMM: an integrated database of large-scale cohort, genome and clinical data for the Tohoku Medical Megabank Project. <i>Human Genome Variation</i> , 2021, 8, 44.	0.4	7
2729	Limitations of lymphoblastoid cell lines for establishing genetic reference datasets in the immunoglobulin loci. <i>PLoS ONE</i> , 2021, 16, e0261374.	1.1	4
2730	The flagellar germâ€line hypothesis: How flagellate and ciliate gametes significantly shaped the evolution of organismal complexity. <i>BioEssays</i> , 2022, 44, 2100143.	1.2	1
2731	Variant-specific effects define the phenotypic spectrum of HNRNP2-associated neurodevelopmental disorders in males. <i>Human Genetics</i> , 2022, 141, 257-272.	1.8	8
2732	Computational analysis of cancer genome sequencing data. <i>Nature Reviews Genetics</i> , 2022, 23, 298-314.	7.7	38
2733	Genetic architecture of microRNA expression and its link to complex diseases in the Japanese population. <i>Human Molecular Genetics</i> , 2022, 31, 1806-1820.	1.4	14
2734	Driving mosaicism: somatic variants in reference population databases and effect on variant interpretation in rare genetic disease. <i>Human Genomics</i> , 2021, 15, 71.	1.4	5
2735	Association of the Polygenic Risk Score With the Probability of Prodromal Parkinsonâ€™s Disease in Older Adults. <i>Frontiers in Molecular Neuroscience</i> , 2021, 14, 739571.	1.4	6
2736	Deviation from baseline mutation burden provides powerful and robust rare-variants association test for complex diseases. <i>Nucleic Acids Research</i> , 2022, 50, e34-e34.	6.5	3
2738	Intellectual disability genomics: current state, pitfalls and future challenges. <i>BMC Genomics</i> , 2021, 22, 909.	1.2	31
2739	Accurate Prediction of Protein Sequences for Proteogenomics Data Integration. <i>Methods in Molecular Biology</i> , 2022, 2420, 233-260.	0.4	0
2740	Genomic study of a large family with complex neurological phenotype including hearing loss, imbalance and action tremor. <i>Neurobiology of Aging</i> , 2022, 113, 137-142.	1.5	1
2741	Compound heterozygous c.598_612del and c.1746-20Câ€™>â€™G CAPN3 genotype cause autosomal recessive limb-girdle muscular dystrophy-1: a case report. <i>BMC Musculoskeletal Disorders</i> , 2021, 22, 1020.	0.8	2
2742	Common ALDH3A1 Gene Variant Associated with Keratoconus Risk in the Polish Population. <i>Journal of Clinical Medicine</i> , 2022, 11, 8.	1.0	2
2743	Origins of biallelic inactivation of NF2 in neurofibromatosis type 2. <i>Neuro-Oncology</i> , 2022, 24, 903-913.	0.6	4
2744	Frequency of <i>DPYD</i> gene variants and phenotype inference in a Southern Brazilian population. <i>Annals of Human Genetics</i> , 2022, 86, 102-107.	0.3	2
2745	Genetic epidemiology of autoinflammatory disease variants in Indian population from 1029 whole genomes. <i>Journal of Genetic Engineering and Biotechnology</i> , 2021, 19, 183.	1.5	0
2746	Efficient ancestry and mutation simulation with msprime 1.0. <i>Genetics</i> , 2022, 220, .	1.2	133

#	ARTICLE	IF	CITATIONS
2748	PPIL4 is essential for brain angiogenesis and implicated in intracranial aneurysms in humans. <i>Nature Medicine</i> , 2021, 27, 2165-2175.	15.2	23
2751	Identification and functional characterization of a novel susceptibility locus for small vessel vasculitis with MPO-ANCA. <i>Rheumatology</i> , 2022, 61, 3461-3470.	0.9	8
2752	High Frequency of Juxtamembrane Domain ERBB2 Mutation in Gastric Cancer. <i>Cancer Genomics and Proteomics</i> , 2022, 19, 105-112.	1.0	7
2755	Whole-Exome Sequencing of HPV Positive Tonsillar and Base of Tongue Squamous Cell Carcinomas Reveals a Global Mutational Pattern along with Relapse-Specific Somatic Variants. <i>Cancers</i> , 2022, 14, 77.	1.7	4
2756	Clinical, Histological, and Genetic Features of 25 Patients with Autosomal Dominant Progressive External Ophthalmoplegia (ad-PEO)/PEO-Plus Due to TWNK Mutations. <i>Journal of Clinical Medicine</i> , 2022, 11, 22.	1.0	5
2757	ROHMM: A flexible hidden Markov model framework to detect runs of homozygosity from genotyping data. <i>Human Mutation</i> , 2022, 43, 158-168.	1.1	1
2758	Genomics of Pulmonary Hypertension. <i>Advances in Pulmonary Hypertension</i> , 2021, 20, 142-149.	0.1	0
2759	De novo mutations in childhood cases of sudden unexplained death that disrupt intracellular Ca ²⁺ regulation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	3.3	21
2760	CDH1 germline variants are enriched in patients with colorectal cancer, gastric cancer, and breast cancer. <i>British Journal of Cancer</i> , 2022, 126, 797-803.	2.9	17
2761	Ethnic Diversity of DPD Activity and the DPYD Gene: Review of the Literature. <i>Pharmacogenomics and Personalized Medicine</i> , 2021, Volume 14, 1603-1617.	0.4	6
2763	A homozygous in-frame duplication within the LRRCT consensus sequence of CFAP410 causes cone-rod dystrophy, macular staphyloma and short stature. <i>Ophthalmic Genetics</i> , 2022, 43, 378-384.	0.5	3
2765	Generation and mutational analysis of a transgenic mouse model of human SRY. <i>Human Mutation</i> , 2022, 43, 362-379.	1.1	3
2767	Systematic analysis of PINK1 variants of unknown significance shows intact mitophagy function for most variants. <i>Npj Parkinson's Disease</i> , 2021, 7, 113.	2.5	6
2769	Aberrations in SMAD family of genes among HNSCC patients. <i>Bioinformatics</i> , 2021, 17, 1113-1119.	0.2	2
2770	Biological Pathways Associated With the Development of Pulmonary Toxicities in Mesothelioma Patients Treated With Radical Hemithoracic Radiation Therapy: A Preliminary Study. <i>Frontiers in Oncology</i> , 2021, 11, 784081.	1.3	3
2771	An Iranian Congenital Adrenal Hypoplasia Patient with Elevated Testosterone in Infancy due to a Novel Pathogenic Frameshift Variant in NROB1. <i>International Journal of Endocrinology</i> , 2021, 2021, 1-5.	0.6	0
2772	Epigenome-wide association study of serum urate reveals insights into urate co-regulation and the SLC2A9 locus. <i>Nature Communications</i> , 2021, 12, 7173.	5.8	8
2773	Analysing an allelic series of rare missense variants of CACNA1I in a Swedish schizophrenia cohort. <i>Brain</i> , 2022, 145, 1839-1853.	3.7	18

#	ARTICLE	IF	CITATIONS
2775	Combination of Genomic and Transcriptomic Approaches Highlights Vascular and Circadian Clock Components in Multiple Sclerosis. <i>International Journal of Molecular Sciences</i> , 2022, 23, 310.	1.8	9
2776	Analysis of <i>CYP2C19</i> genetic variants with ischaemic events in UK patients prescribed clopidogrel in primary care: a retrospective cohort study. <i>BMJ Open</i> , 2021, 11, e053905.	0.8	6
2777	De novo pathogenic variant in <i>SETX</i> causes a rapidly progressive neurodegenerative disorder of early childhood-onset with severe axonal polyneuropathy. <i>Acta Neuropathologica Communications</i> , 2021, 9, 194.	2.4	5
2778	Truncating and zinc-finger variants in <i>GLI2</i> are associated with hypopituitarism. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1065-1074.	0.7	3
2781	Predicting deleterious missense genetic variants via integrative supervised nonnegative matrix tri-factorization. <i>Scientific Reports</i> , 2021, 11, 23747.	1.6	0
2782	Machine learning enables new insights into genetic contributions to liver fat accumulation. <i>Cell Genomics</i> , 2021, 1, 100066.	3.0	34
2784	A disorder clinically resembling cystic fibrosis caused by biallelic variants in the <i>AGR2</i> gene. <i>Journal of Medical Genetics</i> , 2022, 59, 993-1001.	1.5	5
2785	Family-Based Whole-Exome Analysis of Specific Language Impairment (SLI) Identifies Rare Variants in <i>BUD13</i> , a Component of the Retention and Splicing (RES) Complex. <i>Brain Sciences</i> , 2022, 12, 47.	1.1	13
2786	Annotating and prioritizing genomic variants using the Ensembl Variant Effect Predictor – A tutorial. <i>Human Mutation</i> , 2022, 43, 986-997.	1.1	30
2788	A Case of Microsatellite Instability-High Colon Cancer in a Young Woman With Familial Adenomatous Polyposis. <i>Journal of the National Comprehensive Cancer Network: JNCCN</i> , 2021, 19, 1377-1381.	2.3	3
2789	Founder genetic variants of <i>ABCC4</i> and <i>ABCC11</i> in the Japanese population are not associated with the development of subacute myelo-optic neuropathy (SMON). <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1845.	0.6	2
2790	Mutations in Hsp90 Cochaperones Result in a Wide Variety of Human Disorders. <i>Frontiers in Molecular Biosciences</i> , 2021, 8, 787260.	1.6	11
2791	Does the adenosine deaminase (<i>ADA</i>) gene confer risk of sleepwalking?. <i>Journal of Sleep Research</i> , 2022, 31, e13537.	1.7	4
2792	<i>CDH5</i> , a Possible New Candidate Gene for Genetic Testing of Lymphedema. <i>Lymphatic Research and Biology</i> , 2021, , .	0.5	3
2794	De novo variants in H3-3A and H3-3B are associated with neurodevelopmental delay, dysmorphic features, and structural brain abnormalities. <i>Npj Genomic Medicine</i> , 2021, 6, 104.	1.7	7
2795	Whole genome sequencing-based copy number variations reveal novel pathways and targets in Alzheimer's disease. <i>Alzheimer's and Dementia</i> , 2022, 18, 1846-1867.	0.4	13
2797	A Case of Type 2 Sialidosis With Deletion of a Single Nucleotide at Position c.947 of the Neuraminidase 1 (<i>NEU1</i>) Gene. <i>Cureus</i> , 2021, 13, e20389.	0.2	0
2798	A functionally impaired missense variant identified in French Canadian families implicates <i>FANCI</i> as a candidate ovarian cancer-predisposing gene. <i>Genome Medicine</i> , 2021, 13, 186.	3.6	12

#	ARTICLE	IF	CITATIONS
2799	A rare CTSC mutation in Papillon-Lefèvre Syndrome results in abolished serine protease activity and reduced NET formation but otherwise normal neutrophil function. <i>PLoS ONE</i> , 2021, 16, e0261724.	1.1	4
2801	The microRNA processor <i>DROSHA</i> is a candidate gene for a severe progressive neurological disorder. <i>Human Molecular Genetics</i> , 2022, 31, 2934-2950.	1.4	6
2802	The use of base editing technology to characterize single nucleotide variants. <i>Computational and Structural Biotechnology Journal</i> , 2022, 20, 1670-1680.	1.9	4
2803	AutozygosityMapper: Identification of disease-mutations in consanguineous families. <i>Nucleic Acids Research</i> , 2022, 50, W83-W89.	6.5	2
2804	OUP accepted manuscript. Database: the Journal of Biological Databases and Curation, 2022, 2022, .	1.4	1
2805	New onset mitral regurgitation caused by mitral valve prolapse in a male patient with dominant pretibial dystrophic epidermolysis bullosa. <i>Indian Journal of Dermatology</i> , 2022, 67, 72.	0.1	2
2806	OUP accepted manuscript. <i>Nucleic Acids Research</i> , 2022, , .	6.5	3
2807	Maternal Uniparental Isodisomy of Chromosome 4 and 8 in Patients with Retinal Dystrophy: SRD5A3-Congenital Disorders of Glycosylation and RP1-Related Retinitis Pigmentosa. <i>Genes</i> , 2022, 13, 359.	1.0	4
2808	Loss of Nexilin function leads to a recessive lethal fetal cardiomyopathy characterized by cardiomegaly and endocardial fibroelastosis. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1676-1687.	0.7	9
2809	Mice with a deficiency in Peroxisomal Membrane Protein 4 (PXMP4) display mild changes in hepatic lipid metabolism. <i>Scientific Reports</i> , 2022, 12, 2512.	1.6	7
2810	Anchored Multiplex PCR Custom Melanoma Next Generation Sequencing Panel for Analysis of Circulating Tumor DNA. <i>Frontiers in Oncology</i> , 2022, 12, 820510.	1.3	2
2811	The Many Faces of G Protein-Coupled Receptor 143, an Atypical Intracellular Receptor. <i>Frontiers in Molecular Biosciences</i> , 2022, 9, 873777.	1.6	3
2812	A Severe Dementia Syndrome Caused by Intron Retention and Cryptic Splice Site Activation in <i>STUB1</i> and Exacerbated by TBP Repeat Expansions. <i>Frontiers in Molecular Neuroscience</i> , 2022, 15, 878236.	1.4	8
2813	The genomic landscape of blood groups in Indigenous Australians in remote communities. <i>Transfusion</i> , 2022, , .	0.8	4
2815	Associations of APOE Gene Variants rs429358 and rs7412 with Parameters of the Blood Lipid Profile and the Risk of Myocardial Infarction and Death in a White Population of Western Siberia. <i>Current Issues in Molecular Biology</i> , 2022, 44, 1713-1724.	1.0	4
2816	Characterization of the mechanism by which a nonsense variant in <i>RYR2</i> leads to disordered calcium handling. <i>Physiological Reports</i> , 2022, 10, e15265.	0.7	7
2817	A Novel FGFR1 Missense Mutation in a Portuguese Family with Congenital Hypogonadotropic Hypogonadism. <i>International Journal of Molecular Sciences</i> , 2022, 23, 4423.	1.8	0
2818	Somatic mosaicism reveals clonal distributions of neocortical development. <i>Nature</i> , 2022, 604, 689-696.	13.7	26

#	ARTICLE	IF	CITATIONS
2819	Genomics, convergent neuroscience and progress in understanding autism spectrum disorder. <i>Nature Reviews Neuroscience</i> , 2022, 23, 323-341.	4.9	81
2820	Predominant Founder Effect among Recurrent Pathogenic Variants for an X-Linked Disorder. <i>Genes</i> , 2022, 13, 675.	1.0	2
2822	Unraveling the Genetic Architecture of Hepatoblastoma Risk: Birth Defects and Increased Burden of Germline Damaging Variants in Gastrointestinal/Renal Cancer Predisposition and DNA Repair Genes. <i>Frontiers in Genetics</i> , 2022, 13, 858396.	1.1	6
2823	Mitochondrial 1555 G>A variant as a potential risk factor for childhood glioblastoma. <i>Neuro-Oncology Advances</i> , 2022, 4, vda045.	0.4	1
2824	Identification and validation of candidate risk genes in endocytic vesicular trafficking associated with esophageal atresia and tracheoesophageal fistulas. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100107.	1.0	2
2825	KBC syndrome mimicking genetic generalized epilepsy. <i>Epilepsy and Behavior Reports</i> , 2022, 19, 100545.	0.5	1
2826	Life-threatening viral disease in a novel form of autosomal recessive <i>IFNAR2</i> deficiency in the Arctic. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	33
2827	Computational Methods for the Study of Peroxisomes in Health and Disease. <i>Physiology</i> , 0, , .	4.0	0
2828	Using whole-genome sequencing to characterize clinically significant blood groups among healthy older Australians. <i>Blood Advances</i> , 2022, 6, 4593-4604.	2.5	1
2829	Genetic variability in exon 1 of the glucocorticoid receptor gene NR3C1 is associated with postoperative complications. <i>Molecular Medicine Reports</i> , 2022, 25, .	1.1	0
2832	Present status of germline findings in precision medicine for Japanese cancer patients: issues in the current system. <i>Japanese Journal of Clinical Oncology</i> , 2022, 52, 599-608.	0.6	2
2833	Long-read sequencing to resolve the parent of origin of a de novo pathogenic <i>UBE3A</i> variant. <i>Journal of Medical Genetics</i> , 2022, 59, 1082-1086.	1.5	4
2834	<i>SCN2A</i>-related epilepsy of infancy with migrating focal seizures: report of a variant with apparent gain- and loss-of-function effects. <i>Journal of Neurophysiology</i> , 2022, 127, 1388-1397.	0.9	6
2835	Predicting causal genes from psychiatric genome-wide association studies using high-level etiological knowledge. <i>Molecular Psychiatry</i> , 2022, 27, 3095-3106.	4.1	4
2836	Panâ€cancer analysis of mutations in open chromatin regions and their possible association with cancer pathogenesis. <i>Cancer Medicine</i> , 2022, , .	1.3	3
2837	Genetic diagnosis of immune dysregulation can lead to targeted therapy for interstitial lung disease: A case series and single center approach. <i>Pediatric Pulmonology</i> , 2022, 57, 1577-1587.	1.0	4
2838	Genetic and environmental determinants of diastolic heart function. , 2022, 1, 361-371.		12
2839	Expansion of Cancer Risk Profile for <i>BRCA1</i> and <i>BRCA2</i> Pathogenic Variants. <i>JAMA Oncology</i> , 2022, 8, 871.	3.4	70

#	ARTICLE	IF	CITATIONS
2840	A leukemia-protective germline variant mediates chromatin module formation via transcription factor nucleation. <i>Nature Communications</i> , 2022, 13, 2042.	5.8	6
2841	A novel desmoplakin mutation causes dilated cardiomyopathy with palmoplantar keratoderma as an early clinical sign. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2022, 36, 1349-1358.	1.3	3
2842	Diagnostic yield of whole exome data in fetuses aborted for conotruncal malformations. <i>Prenatal Diagnosis</i> , 2022, 42, 852-861.	1.1	1
2843	TTR variants in patients with dilated cardiomyopathy: An investigation of the DCM Precision Medicine Study. <i>Genetics in Medicine</i> , 2022, 24, 1495-1502.	1.1	5
2844	Machine Learning Prediction of Non-Coding Variant Impact in Human Retinal <i>cis</i> -Regulatory Elements. <i>Translational Vision Science and Technology</i> , 2022, 11, 16.	1.1	5
2845	Frequency of pathogenic germline variants in cancer susceptibility genes in 1336 renal cell carcinoma cases. <i>Human Molecular Genetics</i> , 2022, 31, 3001-3011.	1.4	9
2849	The DUB Club: Deubiquitinating Enzymes and Neurodevelopmental Disorders. <i>Biological Psychiatry</i> , 2022, 92, 614-625.	0.7	8
2850	Guiding the global evolution of cytogenetic testing for hematologic malignancies. <i>Blood</i> , 2022, 139, 2273-2284.	0.6	29
2851	Comment on Balsamo et al.: Birt-Hogg-Dubé syndrome with simultaneous hyperplastic polyposis of the gastrointestinal tract: case report and review of the literature. <i>BMC Medical Genomics</i> , 2022, 15, 84.	0.7	0
2852	Genome-wide compound heterozygote analysis highlights DPY19L2 alleles in a non-consanguineous Spanish family with total globozoospermia. <i>Reproductive BioMedicine Online</i> , 2022, 45, 332-340.	1.1	1
2853	Exome sequencing of families from Ghana reveals known and candidate hearing impairment genes. <i>Communications Biology</i> , 2022, 5, 369.	2.0	8
2855	Emerging technologies for prenatal diagnosis: The application of whole genome and RNA sequencing. <i>Prenatal Diagnosis</i> , 2022, 42, 686-696.	1.1	6
2856	A loss-of-function <i>IFNAR1</i> allele in Polynesia underlies severe viral diseases in homozygotes. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	28
2857	Common Variant in <i>ALDH2</i> Modifies the Risk of Breast Cancer Among Carriers of the p.K3326* Variant in <i>BRCA2</i> . <i>JCO Precision Oncology</i> , 2022, 6, e2100450.	1.5	1
2858	Exome sequencing in bipolar disorder identifies <i>AKAP11</i> as a risk gene shared with schizophrenia. <i>Nature Genetics</i> , 2022, 54, 541-547.	9.4	65
2866	uORF-introducing variants in the 5'UTR of the <i>NIPBL</i> gene as a cause of Cornelia de Lange syndrome. <i>Human Mutation</i> , 2022, 43, 1239-1248.	1.1	6
2867	Polygenic risk score as a possible tool for identifying familial monogenic causes of complex diseases. <i>Genetics in Medicine</i> , 2022, 24, 1545-1555.	1.1	12
2868	Whole-genome sequencing analysis of clozapine-induced myocarditis. <i>Pharmacogenomics Journal</i> , 2022, 22, 173-179.	0.9	1

#	ARTICLE	IF	CITATIONS
2869	A de novo startâ€lost variant in <i>ANKRD28</i> in a Holstein calf with dwarfism. <i>Animal Genetics</i> , 2022, 53, 470-471.	0.6	3
2870	Clinically actionable cancer somatic variants (CACSV): a tumor interpreted dataset for analytical workflows. <i>BMC Medical Genomics</i> , 2022, 15, 95.	0.7	1
2871	Large-scale discovery of novel neurodevelopmental disorder-related genes through a unified analysis of single-nucleotide and copy number variants. <i>Genome Medicine</i> , 2022, 14, 40.	3.6	13
2873	VHL-P138R and VHL-L163R Novel Variants: Mechanisms of VHL Pathogenicity Involving HIF-Dependent and HIF-Independent Actions. <i>Frontiers in Endocrinology</i> , 2022, 13, 854365.	1.5	0
2874	Whole-Exome Sequencing Reveals Recurrent but Heterogeneous Mutational Profiles in Sporadic WHO Grade 1 Meningiomas. <i>Frontiers in Oncology</i> , 2021, 11, 740782.	1.3	5
2876	Cross-species analysis of LZTR1 loss-of-function mutants demonstrates dependency to RIT1 orthologs. <i>ELife</i> , 2022, 11, .	2.8	8
2877	FABIAN-variant: predicting the effects of DNA variants on transcription factor binding. <i>Nucleic Acids Research</i> , 2022, 50, W322-W329.	6.5	12
2878	Structural bioinformatics enhances the interpretation of somatic mutations in KDM6A found in human cancers. <i>Computational and Structural Biotechnology Journal</i> , 2022, 20, 2200-2211.	1.9	5
2879	Vascular burden and genetic risk in association with cognitive performance and dementia in a population-based study. <i>Cerebral Circulation - Cognition and Behavior</i> , 2022, 3, 100145.	0.4	0
2881	Possible role for rare <i>TRPM7</i> variants in patients with hypomagnesaemia with secondary hypocalcaemia. <i>Nephrology Dialysis Transplantation</i> , 2023, 38, 679-690.	0.4	6
2883	Implementation of the User-Friendly Odds Ratio Calculator for Unvaccinated Individuals in a Country with a High COVID-19 Death Toll. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0
2884	A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, 24, 1567-1582.	1.1	5
2887	'Fly-ing' from rare to common neurodegenerative disease mechanisms. <i>Trends in Genetics</i> , 2022, 38, 972-984.	2.9	16
2888	Combination of late gadolinium enhancement and genotype improves prediction of prognosis in nonâ€ischaemic dilated cardiomyopathy. <i>European Journal of Heart Failure</i> , 2022, 24, 1183-1196.	2.9	13
2889	The Thousand Polish Genomesâ€A Database of Polish Variant Allele Frequencies. <i>International Journal of Molecular Sciences</i> , 2022, 23, 4532.	1.8	15
2891	Role of testisâ€specific serine kinase 1B in undiagnosed male infertility. <i>Molecular Medicine Reports</i> , 2022, 25, .	1.1	1
2893	Preparation of Duplex Sequencing Libraries for Archival Paraffin-Embedded Tissue Samples Using Single-Strand-Specific Nuclease P1. <i>International Journal of Molecular Sciences</i> , 2022, 23, 4586.	1.8	1
2894	Loss, Gain and Altered Function of GlyR Î±2 Subunit Mutations in Neurodevelopmental Disorders. <i>Frontiers in Molecular Neuroscience</i> , 2022, 15, 886729.	1.4	7

#	ARTICLE	IF	CITATIONS
2895	Genome interpretation using in silico predictors of variant impact. <i>Human Genetics</i> , 2022, 141, 1549-1577.	1.8	26
2896	The Genetic and Molecular Analyses of RAD51C and RAD51D Identifies Rare Variants Implicated in Hereditary Ovarian Cancer from a Genetically Unique Population. <i>Cancers</i> , 2022, 14, 2251.	1.7	4
2897	Inferring Potential Cancer Driving Synonymous Variants. <i>Genes</i> , 2022, 13, 778.	1.0	1
2899	Population-based screening in children for early diagnosis and treatment of familial hypercholesterolemia: design of the VRONI study. <i>Medizinische Genetik</i> , 2022, 34, 41-51.	0.1	0
2900	Single-nucleotide polymorphisms and the effectiveness of taxane-based chemotherapy in premenopausal breast cancer: a population-based cohort study in Denmark. <i>Breast Cancer Research and Treatment</i> , 2022, , 1.	1.1	0
2901	SARS-CoV-2 Susceptibility and ACE2 Gene Variations Within Diverse Ethnic Backgrounds. <i>Frontiers in Genetics</i> , 2022, 13, 888025.	1.1	14
2902	ACLY and CKD: A Mendelian Randomization Analysis. <i>Kidney International Reports</i> , 2022, 7, 1673-1681.	0.4	1
2903	Single-Nucleotide Variations, Insertions/Deletions and Copy Number Variations in Myelodysplastic Syndrome during Disease Progression Revealed by a Single-Cell DNA Sequencing Platform. <i>International Journal of Molecular Sciences</i> , 2022, 23, 4647.	1.8	3
2904	Whole-Exome Sequencing of Germline Variants in Non-BRCA Families with Hereditary Breast Cancer. <i>Biomedicines</i> , 2022, 10, 1004.	1.4	1
2905	Technical Performance of a 430-Gene Preventative Genomics Assay to Identify Multiple Variant Types Associated with Adult-Onset Monogenic Conditions, Susceptibility Loci, and Pharmacogenetic Insights. <i>Journal of Personalized Medicine</i> , 2022, 12, 667.	1.1	1
2906	Exome sequencing in individuals with cardiovascular laterality defects identifies potential candidate genes. <i>European Journal of Human Genetics</i> , 2022, , .	1.4	1
2907	SCN1A Variants as the Underlying Cause of Genetic Epilepsy with Febrile Seizures Plus in Two Multi-Generational Colombian Families. <i>Genes</i> , 2022, 13, 754.	1.0	2
2908	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. <i>Nature Communications</i> , 2022, 13, 2306.	5.8	20
2909	Genetic insight into Birtâ€“Hoggâ€“DubÃ© syndrome in Indian patients reveals novel mutations at FLCN. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 176.	1.2	1
2910	Extremely low arylsulfatase A enzyme activity does not necessarily cause symptoms: A longâ€“term followâ€“up and review of the literature. <i>JIMD Reports</i> , 2022, 63, 292-302.	0.7	5
2912	Genetic architecture of 11 major psychiatric disorders at biobehavioral, functional genomic and molecular genetic levels of analysis. <i>Nature Genetics</i> , 2022, 54, 548-559.	9.4	101
2913	The human genetic epidemiology of COVID-19. <i>Nature Reviews Genetics</i> , 2022, 23, 533-546.	7.7	64
2914	The Genetics of Primary Ciliary Dyskinesia in Puerto Rico. <i>Diagnostics</i> , 2022, 12, 1127.	1.3	6

#	ARTICLE	IF	CITATIONS
2915	Association of Pathogenic DNA Variants Predisposing to Cardiomyopathy With Cardiovascular Disease Outcomes and All-Cause Mortality. <i>JAMA Cardiology</i> , 2022, 7, 723.	3.0	15
2916	Genetic and chemotherapeutic influences on germline hypermutation. <i>Nature</i> , 2022, 605, 503-508.	13.7	43
2917	The Mutation Analysis of the AMT Gene in a Chinese Family With Nonketotic Hyperglycinemia. <i>Frontiers in Genetics</i> , 2022, 13, .	1.1	0
2918	Evidence for reduced BRCA2 functional activity in Homo sapiens after divergence from the chimpanzee-human last common ancestor. <i>Cell Reports</i> , 2022, 39, 110771.	2.9	5
2919	A rare variant analysis framework using public genotype summary counts to prioritize disease-predisposition genes. <i>Nature Communications</i> , 2022, 13, 2592.	5.8	6
2920	Integration of Protein Structure and Population-Scale DNA Sequence Data for Disease Gene Discovery and Variant Interpretation. <i>Annual Review of Biomedical Data Science</i> , 2022, 5, .	2.8	0
2922	Genome-wide tandem repeat expansions contribute to schizophrenia risk. <i>Molecular Psychiatry</i> , 2022, 27, 3692-3698.	4.1	20
2923	Promoting the genomic revolution in Africa through the Nigerian 100K Genome Project. <i>Nature Genetics</i> , 2022, 54, 531-536.	9.4	25
2924	Complement Gene Variant Effect on Relapse of Complement-Mediated Thrombotic Microangiopathy after Eculizumab Cessation. <i>Blood Advances</i> , 2022, , .	2.5	2
2925	Classic infantile-onset Pompe disease with histopathological neurologic findings linked to a novel <i>GAA</i> gene 4Åbp deletion: A case study. <i>Molecular Genetics & Genomic Medicine</i> , 2022, , e1957.	0.6	1
2926	Accounting for population structure in genetic studies of cystic fibrosis. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100117.	1.0	1
2927	Ablation of <i>Tmcc2</i> Gene Impairs Erythropoiesis in Mice. <i>International Journal of Molecular Sciences</i> , 2022, 23, 5263.	1.8	3
2928	Tumor Purity in Preclinical Mouse Tumor Models. <i>Cancer Research Communications</i> , 2022, 2, 353-365.	0.7	4
2929	How to proceed after "negative" exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. <i>Journal of Inherited Metabolic Disease</i> , 2022, 45, 663-681.	1.7	20
2930	Meta-analysis of sub-Saharan African studies provides insights into genetic architecture of lipid traits. <i>Nature Communications</i> , 2022, 13, 2578.	5.8	18
2931	A minority of somatically mutated genes in pre-existing fatty liver disease have prognostic importance in the development of <i>NAFLD</i> . <i>Liver International</i> , 2022, 42, 1823-1835.	1.9	3
2932	Anticoagulant SERPINS: Endogenous Regulators of Hemostasis and Thrombosis. <i>Frontiers in Cardiovascular Medicine</i> , 2022, 9, 878199.	1.1	24
2933	Targeted long-read sequencing identifies missing pathogenic variants in unsolved Werner syndrome cases. <i>Journal of Medical Genetics</i> , 2022, 59, 1087-1094.	1.5	14

#	ARTICLE	IF	CITATIONS
2934	ANXA11 rs1049550 Associates with LÄ¶fgrerÄ¶s Syndrome and Chronic Sarcoidosis. <i>Cells</i> , 2022, 11, 1557.	1.8	5
2935	Heterozygous variants in <i>PRPF8</i> are associated with neurodevelopmental disorders. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2750-2759.	0.7	4
2936	Autosomal dominantly inherited myopathy likely caused by the <i>TNNT1</i> variant p.(Asp65Ala). <i>Human Mutation</i> , 2022, 43, 1224-1233.	1.1	0
2937	Longitudinal neurodevelopmental profile of a pediatric patient with de novo SPTAN1, epilepsy, and left hippocampal sclerosis. <i>Epilepsy and Behavior Reports</i> , 2022, 19, 100550.	0.5	2
2938	Large scale genotype- and phenotype-driven machine learning in Von Hippel-Lindau disease. <i>Human Mutation</i> , 2022, 43, 1268-1285.	1.1	6
2939	Copy Number Variant Risk Scores Associated With Cognition, Psychopathology, and Brain Structure in Youths in the Philadelphia Neurodevelopmental Cohort. <i>JAMA Psychiatry</i> , 2022, 79, 699.	6.0	8
2940	Rare variants implicate NMDA receptor signaling and cerebellar gene networks in risk for bipolar disorder. <i>Molecular Psychiatry</i> , 2022, 27, 3842-3856.	4.1	5
2941	Delusions, Hallucinations, and Cognitive Decline in Middle Age: A Case of Dementia, GIGYF2 Gene Mutation, and 22q11 Duplication. <i>Indian Journal of Psychological Medicine</i> , 0, , 025371762210848.	0.6	2
2942	Whole-Genome Sequencing Identifies PPARGC1A as a Putative Modifier of Cancer Risk in BRCA1/2 Mutation Carriers. <i>Cancers</i> , 2022, 14, 2350.	1.7	1
2943	Complement Factor I Variants in Complement-Mediated Renal Diseases. <i>Frontiers in Immunology</i> , 2022, 13, .	2.2	1
2944	Plasma proteome analyses in individuals of European and African ancestry identify cis-pQTLs and models for proteome-wide association studies. <i>Nature Genetics</i> , 2022, 54, 593-602.	9.4	98
2945	Rapid Molecular Diagnosis of Genetically Inherited Neuromuscular Disorders Using Next-Generation Sequencing Technologies. <i>Journal of Clinical Medicine</i> , 2022, 11, 2750.	1.0	3
2946	Analysis of MRI-derived spleen iron in the UK Biobank identifies genetic variation linked to iron homeostasis and hemolysis. <i>American Journal of Human Genetics</i> , 2022, 109, 1092-1104.	2.6	7
2947	CCL22 mutations drive natural killer cell lymphoproliferative disease by deregulating microenvironmental crosstalk. <i>Nature Genetics</i> , 2022, 54, 637-648.	9.4	13
2950	<i>TADeus2</i> : a web server facilitating the clinical diagnosis by pathogenicity assessment of structural variations disarranging 3D chromatin structure. <i>Nucleic Acids Research</i> , 2022, 50, W744-W752.	6.5	7
2951	Transcriptome analysis provides critical answers to the "variants of uncertain significance" conundrum. <i>Human Mutation</i> , 2022, 43, 1590-1608.	1.1	7
2952	Clonal Hematopoiesis Is Associated with Increased Risk of Severe Neurotoxicity in Axicabtagene Ciloleucele Therapy of Large B-Cell Lymphoma. <i>Blood Cancer Discovery</i> , 2022, 3, 385-393.	2.6	29
2953	Germline predisposition to pediatric Ewing sarcoma is characterized by inherited pathogenic variants in DNA damage repair genes. <i>American Journal of Human Genetics</i> , 2022, 109, 1026-1037.	2.6	19

#	ARTICLE	IF	CITATIONS
2954	LT1, an ONT long-read-based assembly scaffolded with Hi-C data and polished with short reads. <i>GigaByte</i> , 0, 2022, 1-16.	0.0	0
2955	Contribution of rare whole-genome sequencing variants to plasma protein levels and the missing heritability. <i>Nature Communications</i> , 2022, 13, 2532.	5.8	9
2956	Further description of two patients with biallelic variants in <i>NADSYN1</i> in association with cardiac and vertebral anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2479-2484.	0.7	8
2958	A gene-to-patient approach uplifts novel disease gene discovery and identifies 18 putative novel disease genes. <i>Genetics in Medicine</i> , 2022, 24, 1697-1707.	1.1	14
2959	GWAS of thyroid dysgenesis identifies a risk locus at 2q33.3 linked to regulation of Wnt signaling. <i>Human Molecular Genetics</i> , 2022, 31, 3967-3974.	1.4	2
2960	Refinements and considerations for trio whole-genome sequence analysis when investigating Mendelian diseases presenting in early childhood. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100113.	1.0	4
2961	Stx4 is required to regulate cardiomyocyte Ca ²⁺ handling during vertebrate cardiac development. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100115.	1.0	1
2962	Techniques for estimating genetically variable peptides and semi-continuous likelihoods from massively parallel sequencing data. <i>Forensic Science International: Genetics</i> , 2022, 59, 102719.	1.6	2
2963	Oxytocin and vasotocin receptor variation and the evolution of human prosociality. <i>Comprehensive Psychoneuroendocrinology</i> , 2022, 11, 100139.	0.7	6
2964	Haplotype phasing of a bipolar disorder pedigree revealed rare multiple mutations of SPOCD1 gene in the 1p36 susceptibility locus. <i>Journal of Affective Disorders</i> , 2022, 310, 96-105.	2.0	2
2965	Are polygenic risk scores ready for the cancer clinic? a perspective. <i>Translational Lung Cancer Research</i> , 2022, 11, 910-919.	1.3	6
2966	Genomic architecture of fetal central nervous system anomalies using whole-genome sequencing. <i>Npj Genomic Medicine</i> , 2022, 7, 31.	1.7	6
2968	Nonsense Mutations in Eukaryotes. <i>Biochemistry (Moscow)</i> , 2022, 87, 400-412.	0.7	1
2969	Defining pathogenicity of <i>NOTCH2</i> variants for diagnosis of Alagille syndrome type 2 using a large cohort of patients. <i>Liver International</i> , 2022, 42, 1836-1848.	1.9	3
2971	Opportunities and challenges for the use of common controls in sequencing studies. <i>Nature Reviews Genetics</i> , 2022, 23, 665-679.	7.7	13
2972	Novel Missense and Splice Site Mutations in <i>USH2A</i> , <i>CDH23</i> , <i>PCDH15</i> , and <i>ADGRV1</i> Are Associated With Usher Syndrome in Lebanon. <i>Frontiers in Genetics</i> , 2022, 13, .	1.1	2
2973	Prevalence and Phenotypic Effects of Copy Number Variants in Isolated Hypogonadotropic Hypogonadism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 2228-2242.	1.8	10
2974	SHROOM4 Variants Are Associated With X-Linked Epilepsy With Features of Generalized Seizures or Generalized Discharges. <i>Frontiers in Molecular Neuroscience</i> , 2022, 15, .	1.4	5

#	ARTICLE	IF	CITATIONS
2975	Spectrum of Mutations in NDP Resulting in Ocular Disease; a Systematic Review. <i>Frontiers in Genetics</i> , 2022, 13, .	1.1	7
2976	Annotating functional effects of non-coding variants in neuropsychiatric cell types by deep transfer learning. <i>PLoS Computational Biology</i> , 2022, 18, e1010011.	1.5	7
2977	An R307H substitution in GATA1 that prevents Ser310 phosphorylation causes severe fetal anemia. <i>Blood Advances</i> , 2022, 6, 4330-4334.	2.5	4
2978	Identifying common transcriptome signatures of cancer by interpreting deep learning models. <i>Genome Biology</i> , 2022, 23, 117.	3.8	11
2979	MiRLog and dbmiR: Prioritization and functional annotation tools to study human microRNA sequence variants. <i>Human Mutation</i> , 2022, . .	1.1	1
2980	Bioinformatics for the Origin and Evolution of Viruses. <i>Advances in Experimental Medicine and Biology</i> , 2022, 1368, 53-71.	0.8	2
2981	Brief Report: Evidence of Autism Spectrum Disorder Caused by a Mutation in ATRX Gene: A Case Report. <i>Journal of Autism and Developmental Disorders</i> , 2024, 54, 379-388.	1.7	1
2982	Integration of rare expression outlier-associated variants improves polygenic risk prediction. <i>American Journal of Human Genetics</i> , 2022, 109, 1055-1064.	2.6	8
2983	Pediatric T-ALL type-1 and type-2 relapses develop along distinct pathways of clonal evolution. <i>Leukemia</i> , 2022, 36, 1759-1768.	3.3	4
2984	Feasibility of whole genome and transcriptome profiling in pediatric and young adult cancers. <i>Nature Communications</i> , 2022, 13, 2485.	5.8	31
2985	Evaluation of cfDNA as an early detection assay for dense tissue breast cancer. <i>Scientific Reports</i> , 2022, 12, 8458.	1.6	3
2986	A complete pedigree-based graph workflow for rare candidate variant analysis. <i>Genome Research</i> , 2022, . .	2.4	1
2987	A de novo pathogenic variant identified in a boy with Poland syndrome.. <i>Cold Spring Harbor Molecular Case Studies</i> , 2022, 8, .	0.7	0
2988	Computational and experimental methods for classifying variants of unknown clinical significance.. <i>Cold Spring Harbor Molecular Case Studies</i> , 2022, 8, .	0.7	7
2989	A basement membrane discovery pipeline uncovers network complexity, regulators, and human disease associations. <i>Science Advances</i> , 2022, 8, eabn2265.	4.7	76
2990	Human OTULIN haploinsufficiency impairs cell-intrinsic immunity to staphylococcal Î±-toxin. <i>Science</i> , 2022, 376, eabm6380.	6.0	25
2991	Phase II Clinical Trial of Eribulinâ€™Gemcitabine Combination Therapy in Previously Treated Patients With Advanced Liposarcoma or Leiomyosarcoma. <i>Clinical Cancer Research</i> , 2022, 28, 3225-3234.	3.2	5
2992	Exploring the Genetic Architecture of Spontaneous Coronary Artery Dissection Using Whole-Genome Sequencing. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, 101161CIRCGEN121003527.	1.6	14

#	ARTICLE	IF	CITATIONS
2993	Psychiatric manifestations of rare variation in medically actionable genes: a PheWAS approach. <i>BMC Genomics</i> , 2022, 23, 385.	1.2	1
2994	Tumor and Constitutional Sequencing for Neurofibromatosis Type 1. <i>JCO Precision Oncology</i> , 2022, 6, e2100540.	1.5	4
2995	Human phospho- signaling networks of SARS-CoV-2 infection are rewired by population genetic variants. <i>Molecular Systems Biology</i> , 2022, 18, e10823.	3.2	8
2999	A familial case of inherited autosomal-dominant non-syndromic hearing loss caused by a <i>TECTA</i> mutation in the zona pellucida domain of alpha-tectorin: indication of elevation of hearing thresholds in high frequencies with age. <i>Audiology Japan</i> , 2022, 65, 145-151.	0.1	0
3002	Association of <i>APOE</i>-Independent Alzheimer Disease Polygenic Risk Score With Brain Amyloid Deposition in Asymptomatic Older Adults. <i>Neurology</i> , 2022, 99, .	1.5	6
3003	Overview of Neuromuscular Disorder Molecular Diagnostic Experience for the Population of Latvia. <i>Neurology: Genetics</i> , 2022, 8, .	0.9	3
3004	Familial Brain Calcifications With Leukoencephalopathy. <i>Neurology: Genetics</i> , 2022, 8, .	0.9	2
3005	Prevalence of alcohol dehydrogenase 1B and aldehyde dehydrogenase 2 genotypes in Kashmir, an Asian high-risk region of esophageal squamous cell carcinoma. , 2022, 33, 201042.		0
3006	Automated next-generation profiling of genomic alterations in human cancers. <i>Nature Communications</i> , 2022, 13, .	5.8	8
3007	Case Report: Brachydactyly Type A1 Induced by a Novel Variant of in-Frame Insertion in the IHH Gene. <i>Frontiers in Genetics</i> , 2022, 13, .	1.1	1
3008	Genetics of bipolar disorder: insights into its complex architecture and biology from common and rare variants. <i>Journal of Human Genetics</i> , 2023, 68, 183-191.	1.1	5
3009	A promoter variant in the <i>OTC</i> gene associated with late and variable age of onset hyperammonemia. <i>Journal of Inherited Metabolic Disease</i> , 2022, 45, 710-718.	1.7	5
3010	Identification of a Complex Allele in <i>IMPG2</i> as a Cause of Adult-Onset Vitelliform Macular Dystrophy. , 2022, 63, 27.		13
3014	Computational analysis of missense filamin-A variants, including the novel p.Arg484Gln variant of two brothers with periventricular nodular heterotopia. <i>PLoS ONE</i> , 2022, 17, e0265400.	1.1	0
3015	Lipoprotein(a) beyond the kringle IV repeat polymorphism: The complexity of genetic variation in the LPA gene. <i>Atherosclerosis</i> , 2022, 349, 17-35.	0.4	61
3017	Distinct landscapes of deleterious variants in DNA damage repair system in ethnic human populations. <i>Life Science Alliance</i> , 2022, 5, e202101319.	1.3	3
3018	Regionally defined proteomic profiles of human cerebral tissue and organoids reveal conserved molecular modules of neurodevelopment. <i>Cell Reports</i> , 2022, 39, 110846.	2.9	7
3019	Maternal exome analysis for the diagnosis of oocyte maturation defects and early embryonic developmental arrest. <i>Reproductive BioMedicine Online</i> , 2022, 45, 508-518.	1.1	4

#	ARTICLE	IF	CITATIONS
3023	Genetic changes associated with relapse in favorable histology Wilms tumor: A Children's Oncology Group AREN03B2 study. <i>Cell Reports Medicine</i> , 2022, 3, 100644.	3.3	13
3024	The Genetics of Autoimmune Myositis. <i>Frontiers in Immunology</i> , 2022, 13, .	2.2	6
3025	ezQTL: A Web Platform for Interactive Visualization and Colocalization of QTLs and GWAS Loci. <i>Genomics, Proteomics and Bioinformatics</i> , 2022, 20, 541-548.	3.0	17
3026	Hereditary hemorrhagic telangiectasia: First demonstration of a founder effect in Italy; the <i>ACVRL1</i> c.289_294del variant originated in the country of Bergamo 200 years ago. <i>Molecular Genetics & Genomic Medicine</i> , 0, , .	0.6	3
3028	Clinical validity assessment of genes frequently tested on intellectual disability/autism sequencing panels. <i>Genetics in Medicine</i> , 2022, 24, 1899-1908.	1.1	9
3029	Detecting Tandem Repeat Expansions Using Short-Read Sequencing for Clinical Use. <i>NeuroMethods</i> , 2022, , 15-42.	0.2	2
3030	Amplification is the Primary Mode of Gene-by-Sex Interaction in Complex Human Traits. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0
3031	An <i>IGHG1</i> Variant Introgressed From Vindija Neanderthal Archaic Hominin Confers Enhanced Antibody Immunity Against SARS-CoV-2. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0
3032	Targeted resequencing of the 13q13 spondyloarthritis-linked locus identifies a rare variant in <i>FREM2</i> possibly associated with familial spondyloarthritis. <i>Joint Bone Spine</i> , 2022, 89, 105419.	0.8	2
3033	Rare Germline Variants Are Associated with Rapid Biochemical Recurrence After Radical Prostate Cancer Treatment: A Pan Prostate Cancer Group Study. <i>European Urology</i> , 2022, 82, 201-211.	0.9	2
3037	A de novo missense mutation in <i>KIT</i> is responsible for dominant white spotting phenotype in a Standardbred horse. <i>Animal Genetics</i> , 2022, 53, 534-537.	0.6	5
3038	Identification of potential biomarkers and novel therapeutic targets through genomic analysis of small cell bladder carcinoma and associated clinical outcomes. <i>Urologic Oncology: Seminars and Original Investigations</i> , 2022, , .	0.8	1
3039	Crovalimab for treatment of patients with paroxysmal nocturnal haemoglobinuria and complement <i>C5</i> polymorphism: Subanalysis of the phase 1/2 <i>COMPOSER</i> study. <i>British Journal of Haematology</i> , 2022, 198, .	1.2	4
3040	Genetic and Clinical Characteristics of Patients in the Middle East With Multisystem Inflammatory Syndrome in Children. <i>JAMA Network Open</i> , 2022, 5, e2214985.	2.8	13
3041	Exome sequencing identifies rare mutations of <i>LDLR</i> and <i>QTRT1</i> conferring risk for early-onset coronary artery disease in Chinese. <i>National Science Review</i> , 2022, 9, .	4.6	1
3044	Screening of the <i>TMEM151A</i> Gene in Patients With Paroxysmal Kinesigenic Dyskinesia and Other Movement Disorders. <i>Frontiers in Neurology</i> , 0, 13, .	1.1	3
3045	Mapping the O-GlcNAc Modified Proteome: Applications for Health and Disease. <i>Frontiers in Molecular Biosciences</i> , 2022, 9, .	1.6	6
3046	Eph and Ephrin Variants in Malaysian Neural Tube Defect Families. <i>Genes</i> , 2022, 13, 952.	1.0	1

#	ARTICLE	IF	CITATIONS
3048	Defining mitochondrial protein functions through deep multiomic profiling. <i>Nature</i> , 2022, 606, 382-388.	13.7	49
3049	Molecular Genetics and Pathogenesis of the Floating Harbor Syndrome: Case Report of Long-Term Growth Hormone Treatment and a Literature Review. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	6
3050	Analysis of Mutational Profile of Hypopharyngeal and Laryngeal Head and Neck Squamous Cell Carcinomas Identifies KMT2C as a Potential Tumor Suppressor. <i>Frontiers in Oncology</i> , 0, 12, .	1.3	3
3051	Expanding the Molecular Spectrum of ANKRD11 Gene Defects in 33 Patients with a Clinical Presentation of KBG Syndrome. <i>International Journal of Molecular Sciences</i> , 2022, 23, 5912.	1.8	6
3052	Cancer-related Mutations with Local or Long-range Effects on an Allosteric Loop of p53. <i>Journal of Molecular Biology</i> , 2022, 434, 167663.	2.0	17
3054	PHACT: Phylogeny-Aware Computing of Tolerance for Missense Mutations. <i>Molecular Biology and Evolution</i> , 2022, 39, .	3.5	3
3055	Recessive LAMA5 Variants Associated With Partial Epilepsy and Spasms in Infancy. <i>Frontiers in Molecular Neuroscience</i> , 2022, 15, .	1.4	12
3056	A Role for Gene-Environment Interactions in Autism Spectrum Disorder Is Supported by Variants in Genes Regulating the Effects of Exposure to Xenobiotics. <i>Frontiers in Neuroscience</i> , 2022, 16, .	1.4	16
3058	Rare Variants in Inborn Errors of Immunity Genes Associated With Covid-19 Severity. <i>Frontiers in Cellular and Infection Microbiology</i> , 2022, 12, .	1.8	5
3059	Prediction of Neurodevelopmental Disorders Based on De Novo Coding Variation. <i>Journal of Autism and Developmental Disorders</i> , 2023, 53, 963-976.	1.7	2
3060	An N-glycosylation hotspot in immunoglobulin λ light chains is associated with AL amyloidosis. <i>Leukemia</i> , 2022, 36, 2076-2085.	3.3	10
3061	Viral and Genomic Drivers of Squamous Cell Neoplasms Arising in the Lacrimal Drainage System. <i>Cancers</i> , 2022, 14, 2558.	1.7	3
3062	Hypomorphic GINS3 variants alter DNA replication and cause Meier-Gorlin syndrome. <i>JCI Insight</i> , 2022, 7, .	2.3	6
3063	Mutational signatures are markers of drug sensitivity of cancer cells. <i>Nature Communications</i> , 2022, 13, .	5.8	29
3064	Phenotype expansion of variants affecting p38 MAPK signaling in hypospadias patients. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, .	1.2	0
3065	Association of Rare <i>APOE</i> Missense Variants V236E and R251G With Risk of Alzheimer Disease. <i>JAMA Neurology</i> , 2022, 79, 652.	4.5	31
3066	Integrating Genetic Structural Variations and Whole-Genome Sequencing Into Clinical Neurology. <i>Neurology: Genetics</i> , 2022, 8, e200005.	0.9	4
3067	Clinical and genetic findings in <i>TRPM1</i> -related congenital stationary night blindness. <i>Acta Ophthalmologica</i> , 2022, 100, .	0.6	4

#	ARTICLE	IF	CITATIONS
3068	Determinants of trafficking, conduction, and disease within a K ⁺ channel revealed through multiparametric deep mutational scanning. <i>ELife</i> , 0, 11, .	2.8	23
3069	Omeprazole Treatment Failure in Gastroesophageal Reflux Disease and Genetic Variation at the CYP2C Locus. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	5
3071	Genetic architecture of band neutrophil fraction in Iceland. <i>Communications Biology</i> , 2022, 5, .	2.0	1
3072	A phenome-wide association study identifies effects of copy-number variation of VNTRs and multicopy genes on multiple human traits. <i>American Journal of Human Genetics</i> , 2022, 109, 1065-1076.	2.6	12
3074	Combined exome and transcriptome sequencing of non-muscle-invasive bladder cancer: associations between genomic changes, expression subtypes, and clinical outcomes. <i>Genome Medicine</i> , 2022, 14, .	3.6	5
3075	Ultra-rare and common genetic variant analysis converge to implicate negative selection and neuronal processes in the aetiology of schizophrenia. <i>Molecular Psychiatry</i> , 2022, 27, 3699-3707.	4.1	4
3076	Rare and population-specific functional variation across pig lines. <i>Genetics Selection Evolution</i> , 2022, 54, .	1.2	9
3077	Targeted copy number variant identification across the neurodegenerative disease spectrum. <i>Molecular Genetics & Genomic Medicine</i> , 0, , .	0.6	3
3078	Mapping the genetic landscape of early-onset Alzheimer's disease in a cohort of 36 families. <i>Alzheimer's Research and Therapy</i> , 2022, 14, .	3.0	5
3079	Multiomic analysis reveals cell-type-specific molecular determinants of COVID-19 severity. <i>Cell Systems</i> , 2022, 13, 598-614.e6.	2.9	10
3080	A Novel Homozygous Variant in the COMP Gene Causing a Multiple Epiphyseal Dysplasia 1 with Autosomal Recessive Inheritance. <i>International Journal of Translational Medicine</i> , 2022, 2, 210-219.	0.1	0
3081	Protocol for unbiased, consolidated variant calling from whole exome sequencing data. <i>STAR Protocols</i> , 2022, 3, 101418.	0.5	1
3082	Impaired activity of the fusogenic micropeptide Myomixer causes myopathy resembling Carey-Fineman-Ziter syndrome. <i>Journal of Clinical Investigation</i> , 2022, 132, .	3.9	7
3083	Estimating tumor mutational burden from RNA-sequencing without a matched-normal sample. <i>Nature Communications</i> , 2022, 13, .	5.8	4
3084	Comprehensive evaluation and efficient classification of BRCA1 RING domain missense substitutions. <i>American Journal of Human Genetics</i> , 2022, 109, 1153-1174.	2.6	6
3085	Whole exome sequencing reveals genetic variants in HLA class II genes associated with transplant-free survival of indeterminate acute liver failure. <i>Clinical and Translational Gastroenterology</i> , 2022, Publish Ahead of Print, .	1.3	0
3086	Spontaneous reshaping of vertebral fractures in an adolescent with osteogenesis imperfecta. <i>Bone Reports</i> , 2022, 16, 101595.	0.2	3
3091	Major sex differences in allele frequencies for X chromosomal variants in both the 1000 Genomes Project and gnomAD. <i>PLoS Genetics</i> , 2022, 18, e1010231.	1.5	12

#	ARTICLE	IF	CITATIONS
3092	Presence of rare potential pathogenic variants in subjects under 65 years old with very severe or fatal COVID-19. <i>Scientific Reports</i> , 2022, 12, .	1.6	6
3093	Association Between the LZTFL1 rs11385942 Polymorphism and COVID-19 Severity in Colombian Population. <i>Frontiers in Medicine</i> , 0, 9, .	1.2	11
3094	Function and dynamics of the Mediator complex: novel insights and new frontiers. <i>Transcription</i> , 2022, 13, 39-52.	1.7	2
3096	Genetic pain loss disorders. <i>Nature Reviews Disease Primers</i> , 2022, 8, .	18.1	18
3097	Investigating DNA methylation as a mediator of genetic risk in childhood acute lymphoblastic leukemia. <i>Human Molecular Genetics</i> , 2022, 31, 3741-3756.	1.4	0
3098	Scrutinizing pathogenicity of the USH2A c.2276G>T; p.(Cys759Phe) variant. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	5
3099	Expanding the MYOD1 phenotype: A case report of a patient diagnosed whilst pregnant. <i>Journal of Neuromuscular Diseases</i> , 2022, , 1-4.	1.1	0
3101	Combining SNP-to-gene linking strategies to identify disease genes and assess disease omnigenicity. <i>Nature Genetics</i> , 2022, 54, 827-836.	9.4	61
3102	The Involvement of ALPK3 in Hypertrophic Cardiomyopathy in East Asia. <i>Frontiers in Medicine</i> , 0, 9, .	1.2	2
3103	MYO1H is a novel candidate gene for autosomal dominant pure hereditary spastic paraplegia. <i>Molecular Genetics and Genomics</i> , 0, , .	1.0	0
3104	Gain and loss of TASK3 channel function and its regulation by novel variation cause KCNK9 imprinting syndrome. <i>Genome Medicine</i> , 2022, 14, .	3.6	6
3105	Integrated genomic analyses of acral and mucosal melanomas nominate novel driver genes. <i>Genome Medicine</i> , 2022, 14, .	3.6	13
3106	Mosaic Deletions of Known Genes Explain Skeletal Dysplasias With High and Low Bone Mass. <i>JBMR Plus</i> , 2022, 6, .	1.3	2
3107	HDAC9 structural variants disrupting TWIST1 transcriptional regulation lead to craniofacial and limb malformations. <i>Genome Research</i> , 2022, 32, 1242-1253.	2.4	5
3108	Rare pathogenic variants in WNK3 cause X-linked intellectual disability. <i>Genetics in Medicine</i> , 2022, 24, 1941-1951.	1.1	5
3109	TET2 mutant clonal hematopoiesis and risk of gout. <i>Blood</i> , 2022, 140, 1094-1103.	0.6	57
3110	A comprehensive genomic reporting structure for communicating all clinically significant primary and secondary findings. <i>Human Genetics</i> , 2022, 141, 1875-1885.	1.8	1
3111	Sonic Hedgehog Intron Variant Associated With an Unusual Pediatric Cortical Cataract. , 2022, 63, 25.		0

#	ARTICLE	IF	CITATIONS
3112	Interpreting protein variant effects with computational predictors and deep mutational scanning. <i>DMM Disease Models and Mechanisms</i> , 2022, 15, .	1.2	25
3113	XSIâ€”a genotype compression tool for compressive genomics in large biobanks. <i>Bioinformatics</i> , 2022, 38, 3778-3784.	1.8	6
3115	Founder & BRCA1& mutations in Nepalese population. <i>Journal of Pathology and Translational Medicine</i> , 2022, 56, 212-216.	0.4	2
3116	Mutations in <i>MINAR2</i> encoding membrane integral NOTCH2-associated receptor 2 cause deafness in humans and mice. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	3
3117	Epigenetic Dysregulation in Meningiomas. <i>Neuro-Oncology Advances</i> , 0, , .	0.4	0
3120	Triangulating variation in the population to define mechanisms for precision management of genetic disease. <i>Structure</i> , 2022, , .	1.6	4
3122	Compound Heterozygous FKTN Variants in a Patient with Dilated Cardiomyopathy Led to an Aberrant Î±-Dystroglycan Pattern. <i>International Journal of Molecular Sciences</i> , 2022, 23, 6685.	1.8	3
3123	Reanalysis of exome negative patients with rare disease: a pragmatic workflow for diagnostic applications. <i>Genome Medicine</i> , 2022, 14, .	3.6	17
3124	Quantifying concordant genetic effects of de novo mutations on multiple disorders. <i>ELife</i> , 0, 11, .	2.8	3
3125	Structural and dynamic investigation of nonâ€”synonymous variations in Reninâ€”AGT complex revealed altered binding via hydrogenâ€”bonding network reprogramming to accelerate the hypertension pathway. <i>Chemical Biology and Drug Design</i> , 2022, 100, 730-746.	1.5	1
3126	The Arginine Methyltransferase <i>Carm1</i> is Necessary for Heart Development. <i>G3: Genes, Genomes, Genetics</i> , 0, , .	0.8	2
3127	ADGRV1 Variants in Febrile Seizures/Epilepsy With Antecedent Febrile Seizures and Their Associations With Audio-Visual Abnormalities. <i>Frontiers in Molecular Neuroscience</i> , 0, 15, .	1.4	7
3128	Detection of brain somatic variation in epilepsyâ€”associated developmental lesions. <i>Epilepsia</i> , 2022, 63, 1981-1997.	2.6	29
3129	Screening of potential novel candidate genes in schwannomatosis patients. <i>Human Mutation</i> , 2022, 43, 1368-1376.	1.1	3
3130	Spatio-temporal dynamics of pathogenic variants associated with monogenic disorders reconstructed with ancient DNA. <i>PLoS ONE</i> , 2022, 17, e0269628.	1.1	1
3131	<i>CAPN3</i> c.1746â€”G variant is hypomorphic for LGMD R1 calpain 3â€”related. <i>Human Mutation</i> , 2022, 43, 1347-1353.	1.1	4
3132	State of the Science for Kidney Disorders in Phelan-McDermid Syndrome: UPK3A, FBLN1, WNT7B, and CELSR1 as Candidate Genes. <i>Genes</i> , 2022, 13, 1042.	1.0	4
3133	Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome. <i>Brain</i> , 2022, 145, 3383-3390.	3.7	3

#	ARTICLE	IF	CITATIONS
3134	Exome sequencing in multiplex families with left-sided cardiac defects has high yield for disease gene discovery. <i>PLoS Genetics</i> , 2022, 18, e1010236.	1.5	8
3135	Case Report: Novel CSF1R Variant in a Patient With Behavioral Variant Frontotemporal Dementia Syndrome With Prodromal Repetitive Scratching Behavior. <i>Frontiers in Neurology</i> , 0, 13, .	1.1	1
3136	Whole genome duplication in oral squamous cell carcinoma in patients younger than 50%years: implications for prognosis and adverse clinicopathological factors. <i>Genes Chromosomes and Cancer</i> , 2022, 61, 561-571.	1.5	2
3139	Pericarditis and Autoinflammation: A Clinical and Genetic Analysis of Patients With Idiopathic Recurrent Pericarditis and Monogenic Autoinflammatory Diseases at a National Referral Center. <i>Journal of the American Heart Association</i> , 2022, 11, .	1.6	15
3140	Clinicopathological features in two families with <scp>MARS</scp>â€related <scp>Charcotâ€Marieâ€Tooth</scp> disease. <i>Neuropathology</i> , 2022, 42, 505-511.	0.7	2
3141	Genome-wide mapping of somatic mutation rates uncovers drivers of cancer. <i>Nature Biotechnology</i> , 2022, 40, 1634-1643.	9.4	23
3142	Analysis of TMIE gene mutations including the first large deletion of exon 1 with autosomal recessive non-syndromic deafness. <i>BMC Medical Genomics</i> , 2022, 15, .	0.7	1
3143	Discovery of eQTL Alleles Associated with Autism Spectrum Disorder: A Caseâ€Control Study. <i>Journal of Autism and Developmental Disorders</i> , 0, , .	1.7	2
3144	Clinical exome sequencing of 1000 families with complex immune phenotypes: Toward comprehensive genomic evaluations. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 150, 947-954.	1.5	13
3145	Genome-wide meta-analysis of iron status biomarkers and the effect of iron on all-cause mortality in HUNT. <i>Communications Biology</i> , 2022, 5, .	2.0	11
3147	Pilot study of bempedalesleukin in combination with nivolumab in patients with metastatic sarcoma. <i>Nature Communications</i> , 2022, 13, .	5.8	21
3148	Loss-of-Function <i>FLNC</i> Variants Are Associated With Arrhythmogenic Cardiomyopathy Phenotypes When Identified Through Exome Sequencing of a General Clinical Population. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, .	1.6	8
3151	Molecular and clinical descriptions of patients with <scp>GABA_A</scp> receptor gene variants (<i>GABRA1</i>, <scp>GABRB2</scp>, <scp>GABRB3</scp>, <scp>GABRG2</scp> </i>): A cohort study, review of literature, and genotypeâ€phenotype correlation. <i>Epilepsia</i> , 2022, 63, 2519-2533.	2.6	23
3152	Impact of Genetic Testing on Therapeutic Decision-Making in Childhood-Onset Epilepsiesâ€a Study in a Tertiary Epilepsy Center. <i>Neurotherapeutics</i> , 2022, 19, 1353-1367.	2.1	14
3155	Respiratory viral infections in otherwise healthy humans with inherited IRF7 deficiency. <i>Journal of Experimental Medicine</i> , 2022, 219, .	4.2	21
3156	Case Report: A Novel EIF2B3 Pathogenic Variant in Central Nervous System Hypomyelination/Vanishing White Matter. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	0
3157	The 3D mutational constraint on amino acid sites in the human proteome. <i>Nature Communications</i> , 2022, 13, .	5.8	15
3158	Filaggrin loss-of-function mutations are associated with persistence of egg and milk allergy. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 150, 1125-1134.	1.5	17

#	ARTICLE	IF	CITATIONS
3159	Variants in ASPH cause exertional heat illness and are associated with malignant hyperthermia susceptibility. <i>Nature Communications</i> , 2022, 13, .	5.8	7
3160	Impact of germline mutations in cancer-predisposing genes on long-term survival in patients with epithelial ovarian cancer. <i>British Journal of Cancer</i> , 2022, 127, 879-885.	2.9	2
3161	Combined Preimplantation Genetic Testing for Genetic Kidney Disease: Genetic Risk Identification, Assisted Reproductive Cycle, and Pregnancy Outcome Analysis. <i>Frontiers in Medicine</i> , 0, 9, .	1.2	2
3162	Functional Analyses of Two Novel <sc><i>LRRK2</i></sc> Pathogenic Variants in Familial Parkinson's Disease. <i>Movement Disorders</i> , 2022, 37, 1761-1767.	2.2	5
3163	Gastroesophageal Glomus Tumors. <i>American Journal of Surgical Pathology</i> , 2022, 46, 1436-1446.	2.1	9
3164	A de novo start-loss in <i>EFTUD2</i> associated with mandibulofacial dysostosis with microcephaly: case report. <i>Journal of Physical Education and Sports Management</i> , 2022, 8, a006206.	0.5	2
3165	Characterization of functionally deficient SIM2 variants found in patients with neurological phenotypes. <i>Biochemical Journal</i> , 0, , .	1.7	0
3166	Missense variants causing Wiedemann-Steiner syndrome preferentially occur in the KMT2A-CXXC domain and are accurately classified using AlphaFold2. <i>PLoS Genetics</i> , 2022, 18, e1010278.	1.5	3
3167	Polishing copy number variant calls on exome sequencing data via deep learning. <i>Genome Research</i> , 2022, 32, 1170-1182.	2.4	5
3168	Expanding the HPSE2 Genotypic Spectrum in Urofacial Syndrome, A Disease Featuring a Peripheral Neuropathy of the Urinary Bladder. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	4
3169	Extended genetic diagnostics for children with profound sensorineural hearing loss by implementing massive parallel sequencing. Diagnostic outcome, family experience and clinical implementation. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2022, 159, 111218.	0.4	5
3171	Confirmation of a Phenotypic Entity for TSPEAR Variants in Egyptian Ectodermal Dysplasia Patients and Role of Ethnicity. <i>Genes</i> , 2022, 13, 1056.	1.0	0
3172	Elucidation of the genetic causes of bicuspid aortic valve disease. <i>Cardiovascular Research</i> , 2023, 119, 857-866.	1.8	11
3173	Non-HLA Gene Polymorphisms in the Pathogenesis of Type 1 Diabetes: Phase and Endotype Specific Effects. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	5
3174	The Compleat Human Genome. , 2022, 1, 234-236.		1
3177	The genetics of drug-induced QT prolongation: evaluating the evidence for pharmacodynamic variants. <i>Pharmacogenomics</i> , 2022, 23, 543-557.	0.6	4
3179	Promotion Effects of Smoking in Polyp Development in Monozygotic Twins with Atypical Colorectal Polyposis. <i>Case Reports in Gastroenterology</i> , 0, , 375-381.	0.3	1
3181	The role of common genetic variation in presumed monogenic epilepsies. <i>EBioMedicine</i> , 2022, 81, 104098.	2.7	12

#	ARTICLE	IF	CITATIONS
3182	Mixed model-based eQTL analysis reveals lncRNAs associated with regulation of genes involved in sex determination and spermatogenesis: The key to understanding human gender imbalance. <i>Computational Biology and Chemistry</i> , 2022, 99, 107713.	1.1	0
3183	Retinal dystrophies: A look beyond the eyes. <i>American Journal of Ophthalmology Case Reports</i> , 2022, 27, 101613.	0.4	1
3184	Functional Studies of Genetic Variants Associated with Human Diseases in Notch Signaling-Related Genes Using <i>Drosophila</i> . <i>Methods in Molecular Biology</i> , 2022, , 235-276.	0.4	1
3185	Current status and future perspectives of the evaluation of missense variants by using three-dimensional structures of proteins. <i>Biophysics and Physicobiology</i> , 2022, , .	0.5	0
3186	Impact of Copy Number Variants and Polygenic Risk Scores on Psychopathology in the UK Biobank. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0
3188	Expanding the phenotype of <i>PIK3C2A</i> related syndrome: Report of two siblings with novel features and genotype. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	0
3192	The impact of rare germline variants on human somatic mutation processes. <i>Nature Communications</i> , 2022, 13, .	5.8	13
3193	Molecular Modelling Hurdle in the Next-Generation Sequencing Era. <i>International Journal of Molecular Sciences</i> , 2022, 23, 7176.	1.8	0
3195	Common genetic variation associated with Mendelian disease severity revealed through cryptic phenotype analysis. <i>Nature Communications</i> , 2022, 13, .	5.8	5
3196	The Location of Missense Variants in the Human GIP Gene Is Indicative for Natural Selection. <i>Frontiers in Endocrinology</i> , 0, 13, .	1.5	1
3197	Penetrance estimation of Alzheimer disease in SORL1 loss-of-function variant carriers using a family-based strategy and stratification by APOE genotypes. <i>Genome Medicine</i> , 2022, 14, .	3.6	7
3198	ANKRD26-Related Thrombocytopenia and Predisposition to Myeloid Neoplasms. <i>Current Hematologic Malignancy Reports</i> , 2022, 17, 105-112.	1.2	7
3199	Concurrent Germline <i>BRCA1</i> and <i>BRCA2</i> and Mismatch Repair Mutations in Young-Onset Pancreatic and Colorectal Cancer: The Importance of Comprehensive Germline and Somatic Characterization to Inform Therapeutic Options. <i>JCO Precision Oncology</i> , 2022, , .	1.5	2
3201	Heterogeneous Distribution of Genetic Mutations in Myosin Binding Protein-C Paralogs. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	5
3202	Neurofibromatosis-1 Gene Mutational Profiles Differ Between Syndromic Disease and Sporadic Cancers. <i>Neurology: Genetics</i> , 2022, 8, .	0.9	3
3203	Uncovering the burden of hidden ciliopathies in the 100 000 Genomes Project: a reverse phenotyping approach. <i>Journal of Medical Genetics</i> , 0, , jmedgenet-2022-108476.	1.5	3
3204	Characterization of ADME Gene Variation in Colombian Population by Exome Sequencing. <i>Frontiers in Pharmacology</i> , 0, 13, .	1.6	4
3205	Six genetically linked mutations in the CD36 gene significantly delay the onset of Alzheimer's disease. <i>Scientific Reports</i> , 2022, 12, .	1.6	5

#	ARTICLE	IF	CITATIONS
3206	Medium-chain acyl-CoA dehydrogenase deficiency: prevalence of ACADM pathogenic variants c.985A>G and c.199T>C in a healthy population in Rio Grande do Sul, Brazil. <i>Reproductive and Developmental Medicine</i> , 2022, 6, 92-97.	0.2	0
3209	3D spatial genome organization in the nervous system: From development and plasticity to disease. <i>Neuron</i> , 2022, 110, 2902-2915.	3.8	10
3210	Protein Quality Control at the Sarcomere: Titin Protection and Turnover and Implications for Disease Development. <i>Frontiers in Physiology</i> , 0, 13, .	1.3	2
3211	Inherited basis of visceral, abdominal subcutaneous and gluteofemoral fat depots. <i>Nature Communications</i> , 2022, 13, .	5.8	43
3212	Investigating the characteristics of genes and variants associated with self-reported hearing difficulty in older adults in the UK Biobank. <i>BMC Biology</i> , 2022, 20, .	1.7	7
3214	Network-based meta-analysis and the candidate gene association studies reveal novel ethnicity-specific variants in <i>MFSD3</i> and <i>MRPL43</i> associated with dementia with Lewy bodies. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 0, , .	1.1	1
3217	Estimation of the number of inherited prion disease mutation carriers in the UK. <i>European Journal of Human Genetics</i> , 2022, 30, 1167-1170.	1.4	3
3218	Improved diagnosis of citrin deficiency by newborn screening using a molecular second-tier test. <i>Molecular Genetics and Metabolism</i> , 2022, 136, 330-336.	0.5	10
3219	Thousands of human mutation clusters are explained by short-range template switching. <i>Genome Research</i> , 0, , gr.276478.121.	2.4	3
3220	The rs35217482 (T755I) Single-Nucleotide Polymorphism in Aldehyde Oxidase-1 Attenuates Protein Dimer Formation and Reduces the Rates of Phthalazine Metabolism. <i>Drug Metabolism and Disposition</i> , 2022, 50, 1126-1131.	1.7	3
3222	Translation of cytoplasmic UBA1 contributes to VEXAS syndrome pathogenesis. <i>Blood</i> , 2022, 140, 1496-1506.	0.6	54
3223	Questioning the Association of the <i>STMN2</i> Dinucleotide Repeat With Amyotrophic Lateral Sclerosis. <i>Neurology: Genetics</i> , 2022, 8, e678.	0.9	1
3225	Genetic variations in methotrexate metabolic pathway genes influence methotrexate responses in rheumatoid arthritis patients in Malaysia. <i>Scientific Reports</i> , 2022, 12, .	1.6	4
3226	Burden of rare variants in arrhythmogenic cardiomyopathy with right dominant form-associated genes provides new insights for molecular diagnosis and clinical management. <i>Human Mutation</i> , 2022, 43, 1333-1342.	1.1	2
3227	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. <i>Genome Medicine</i> , 2022, 14, .	3.6	65
3228	STAT3 gain-of-function is not responsible for low total IgE levels in patients with autoimmune chronic spontaneous urticaria. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	2
3229	3'RNA and whole-genome sequencing of archival uterine leiomyomas reveal a tumor subtype with chromosomal rearrangements affecting either <i>HMGA2</i> , <i>HMGA1</i> , or <i>PLAG1</i> . <i>Genes Chromosomes and Cancer</i> , 0, , .	1.5	2
3230	Case Report: DPM1-CDG: Novel Variant with Severe Phenotype and Literature Review. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1

#	ARTICLE	IF	CITATIONS
3231	Molecular Subclasses of Clear Cell Ovarian Carcinoma and Their Impact on Disease Behavior and Outcomes. <i>Clinical Cancer Research</i> , 2022, 28, 4947-4956.	3.2	22
3233	PRENATAL ULTRASOUND FINDINGS ASSOCIATED WITH <i>PICW</i> VARIANTS:: ONE MORE PIECE IN THE FRYNS SYNDROME PUZZLE? <i>PICW</i> -related prenatal findings. <i>Prenatal Diagnosis</i> , 0, , .	1.1	1
3234	A novel mutation in <i>ATP13A2</i> gene in a patient with complicated hereditary spastic paraplegia accompanied by tubulopathy. <i>Acta Neurologica Belgica</i> , 0, , .	0.5	0
3235	Pathogenicity of missense variants affecting the collagen IV ± 5 carboxy non-collagenous domain in X-linked Alport syndrome. <i>Scientific Reports</i> , 2022, 12, .	1.6	0
3236	Longitudinal dynamics of clonal hematopoiesis identifies gene-specific fitness effects. <i>Nature Medicine</i> , 2022, 28, 1439-1446.	15.2	36
3239	Predominance of <i>BRCA2</i> Mutation and Estrogen Receptor Positivity in Unselected Breast Cancer with <i>BRCA1</i> or <i>BRCA2</i> Mutation. <i>Cancers</i> , 2022, 14, 3266.	1.7	3
3240	<i>MODY</i> probability calculator utility in individuals' selection for genetic testing: Its accuracy and performance. <i>Endocrinology, Diabetes and Metabolism</i> , 2022, 5, .	1.0	7
3241	Mitochondrial DNA variation in Parkinson's disease: Analysis of "out-of-place" population variants as a risk factor. <i>Frontiers in Aging Neuroscience</i> , 0, 14, .	1.7	7
3245	Isolated aneurysmal disease as an underestimated finding in individuals with <i>JAG1</i> pathogenic variants. <i>Human Mutation</i> , 2022, 43, 1824-1828.	1.1	3
3246	Investigating the Genetic Etiology of Pediatric Patients with Peripheral Hypotonia Using the Next-Generation Sequencing Method. <i>Global Medical Genetics</i> , 2022, 09, 200-207.	0.4	0
3248	Establishing the relationship between familial dysbetalipoproteinemia and genetic variants in the <i>APOE</i> gene. <i>Clinical Genetics</i> , 2022, 102, 253-261.	1.0	5
3249	MAFDash: An easy-to-use dashboard builder for mutation data. <i>F1000Research</i> , 0, 11, 748.	0.8	0
3250	LMNA Variants and Risk of Adult-Onset Cardiac Disease. <i>Journal of the American College of Cardiology</i> , 2022, 80, 50-59.	1.2	14
3251	A multiplex pedigree with pathologically confirmed multiple system atrophy and Parkinson's disease with dementia. <i>Brain Communications</i> , 2022, 4, .	1.5	3
3252	StabilitySort: assessment of protein stability changes on a genome-wide scale to prioritize potentially pathogenic genetic variation. <i>Bioinformatics</i> , 2022, 38, 4220-4222.	1.8	2
3253	Missense variants in <i>ANKRD11</i> cause KBG syndrome by impairment of stability or transcriptional activity of the encoded protein. <i>Genetics in Medicine</i> , 2022, 24, 2051-2064.	1.1	12
3255	Uncovering the Contribution of Moderate-Penetrance Susceptibility Genes to Breast Cancer by Whole-Exome Sequencing and Targeted Enrichment Sequencing of Candidate Genes in Women of European Ancestry. <i>Cancers</i> , 2022, 14, 3363.	1.7	2
3259	The pZRS non-coding regulatory mutation resulting in triphalangeal thumb "polysyndactyly syndrome changes the pattern of local interactions. <i>Molecular Genetics and Genomics</i> , 2022, 297, 1343-1352.	1.0	1

#	ARTICLE	IF	CITATIONS
3260	Complex genomic rearrangements: an underestimated cause of rare diseases. <i>Trends in Genetics</i> , 2022, 38, 1134-1146.	2.9	19
3261	Autophagy-associated immune dysregulation and hyperplasia in a patient with compound heterozygous mutations in <i>ATG9A</i> . <i>Autophagy</i> , 2023, 19, 678-691.	4.3	4
3262	Benign SNPs in the Coding Region of <i>TP53</i> : Finding the Needles in a Haystack of Pathogenic Variants. <i>Cancer Research</i> , 2022, 82, 3420-3431.	0.4	6
3263	Deep intronic <i>NIPBL</i> de novo mutations and differential diagnoses revealed by whole genome and RNA sequencing in Cornelia de Lange syndrome patients. <i>Human Mutation</i> , 2022, 43, 1882-1897.	1.1	1
3264	Results of genetic analysis of 11,341 participants enrolled in the My Life, Our Future hemophilia genotyping initiative in the United States. <i>Journal of Thrombosis and Haemostasis</i> , 2022, 20, 2022-2034.	1.9	10
3265	Genetics and therapeutic responses to TIL therapy of pancreatic cancer PDX models. , 2022, , .		2
3266	Genome-wide analyses of 200,453 individuals yield new insights into the causes and consequences of clonal hematopoiesis. <i>Nature Genetics</i> , 2022, 54, 1155-1166.	9.4	109
3267	The contribution of common regulatory and protein-coding TYR variants to the genetic architecture of albinism. <i>Nature Communications</i> , 2022, 13, .	5.8	17
3268	Frequency, Penetrance, and Variable Expressivity of Dilated Cardiomyopathy-Associated Putative Pathogenic Gene Variants in UK Biobank Participants. <i>Circulation</i> , 2022, 146, 110-124.	1.6	25
3269	SNP-to-gene linking strategies reveal contributions of enhancer-related and candidate master-regulator genes to autoimmune disease. <i>Cell Genomics</i> , 2022, 2, 100145.	3.0	19
3270	Dysregulated minor intron splicing in cancer. <i>Cancer Science</i> , 2022, 113, 2934-2942.	1.7	7
3271	Gastrointestinal stromal tumors caused by novel germline variants in <i>SDHB</i> and <i>KIT</i> : a report of two cases and literature review. <i>Clinical Journal of Gastroenterology</i> , 0, , .	0.4	1
3272	Genotypic and phenotypic spectrum of cytosolic phosphoenolpyruvate carboxykinase deficiency. <i>Molecular Genetics and Metabolism</i> , 2022, 137, 18-25.	0.5	5
3273	How Functional Genomics Can Keep Pace With VUS Identification. <i>Frontiers in Cardiovascular Medicine</i> , 0, 9, .	1.1	8
3274	A novel <i>SCN9A</i> gene variant identified in a Chinese girl with paroxysmal extreme pain disorder (PEPD): a rare case report. <i>BMC Medical Genomics</i> , 2022, 15, .	0.7	2
3277	Comprehensive Assessment of Indian Variations in the Druggable Kinome Landscape Highlights Distinct Insights at the Sequence, Structure and Pharmacogenomic Stratum. <i>Frontiers in Pharmacology</i> , 0, 13, .	1.6	1
3278	Bi-allelic loss-of-function variants in <i>PPFIBP1</i> cause a neurodevelopmental disorder with microcephaly, epilepsy, and periventricular calcifications. <i>American Journal of Human Genetics</i> , 2022, 109, 1421-1435.	2.6	6
3279	Analysis of <i>ACE2</i> and <i>TMPRSS2</i> coding variants as a risk factor for SARS-CoV-2 from 946 whole-exome sequencing data in the Turkish population. <i>Journal of Medical Virology</i> , 2022, 94, 5225-5243.	2.5	8

#	ARTICLE	IF	CITATIONS
3280	The evolutionary history of human spindle genes includes back-and-forth gene flow with Neandertals. <i>ELife</i> , 0, 11, .	2.8	12
3281	Mutational landscape of pan-cancer patients with PIK3CA alterations in Chinese population. <i>BMC Medical Genomics</i> , 2022, 15, .	0.7	4
3282	Exploring the Mutational Landscape of Isolated Congenital Heart Defects: An Exome Sequencing Study Using Cardiac DNA. <i>Genes</i> , 2022, 13, 1214.	1.0	4
3283	Molecular Genetic Screening in Patients With ACE Inhibitor/Angiotensin Receptor Blocker-Induced Angioedema to Explore the Role of Hereditary Angioedema Genes. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	4
3285	Prenatal ultrasonographic features in Blomstrand osteochondrodysplasia: Antenatal case series confirmed by postmortem radiology and molecular diagnosis. <i>Prenatal Diagnosis</i> , 0, , .	1.1	2
3286	Association of mitochondrial DNA content, heteroplasmies and inter-generational transmission with autism. <i>Nature Communications</i> , 2022, 13, .	5.8	14
3287	Targeted Next-Generation Sequencing of Thymic Epithelial Tumours Revealed Pathogenic Variants in KIT, ERBB2, KRAS, and TP53 in 30% of Thymic Carcinomas. <i>Cancers</i> , 2022, 14, 3388.	1.7	5
3288	Including diverse and admixed populations in genetic epidemiology research. <i>Genetic Epidemiology</i> , 2022, 46, 347-371.	0.6	11
3289	The Phenotypic Continuum of <i>ATP1A3</i> -Related Disorders. <i>Neurology</i> , 2022, 99, .	1.5	16
3290	SLC25A12 Missense Variant in Nova Scotia Duck Tolling Retrievers Affected by Cerebellar Degenerationâ€”Myositis Complex (CDMC). <i>Genes</i> , 2022, 13, 1223.	1.0	2
3294	Identification of Pathogenic Variant Burden and Selection of Optimal Diagnostic Method Is a Way to Improve Carrier Screening for Autosomal Recessive Diseases. <i>Journal of Personalized Medicine</i> , 2022, 12, 1132.	1.1	5
3295	Functional analysis of missense DARS2 variants in siblings with leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation. <i>Molecular Genetics and Metabolism</i> , 2022, 136, 260-267.	0.5	1
3296	SIMPSON-GOLABI-BEHMEL syndrome type 1: How placental immunohistochemistry can rapidly Predict the diagnosis. <i>Placenta</i> , 2022, 126, 119-124.	0.7	0
3297	PLA2G6-associated neurodegeneration in four different populations-case series and literature review. <i>Parkinsonism and Related Disorders</i> , 2022, 101, 66-74.	1.1	4
3298	Benefit of 5 years of enzyme replacement therapy in advanced late onset Pompe. A case report of misdiagnosis for three decades with acute respiratory failure at presentation. <i>Molecular Genetics and Metabolism Reports</i> , 2022, 32, 100896.	0.4	0
3299	Atypical <i>TDP-43</i> protein expression in an <i>ALS</i> pedigree carrying a p. <i>Y374X</i> truncation mutation in <i>TARDBP</i> . <i>Brain Pathology</i> , 0, , .	2.1	4
3300	<i>PSMC1</i> variant causes a novel neurological syndrome. <i>Clinical Genetics</i> , 2022, 102, 324-332.	1.0	5
3301	Pathogenic variants identified using whole-exome sequencing in <i>Chinese</i> patients with primary ciliary dyskinesia. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 3024-3031.	0.7	0

#	ARTICLE	IF	CITATIONS
3302	Unravelling genetic variants of a swedish family with high risk of prostate cancer. Hereditary Cancer in Clinical Practice, 2022, 20, .	0.6	1
3303	Assessing the clinical utility of protein structural analysis in genomic variant classification: experiences from a diagnostic laboratory. Genome Medicine, 2022, 14, .	3.6	10
3304	Utility and Outcomes of the 2019 American College of Medical Genetics and Genomicsâ€“Clinical Genome Resource Guidelines for Interpretation of Copy Number Variants with Borderline Classifications at an Academic Clinical Diagnostic Laboratory. Journal of Molecular Diagnostics, 2022, 24, 1100-1111.	1.2	2
3306	Protein quality control of N-methyl-D-aspartate receptors. Frontiers in Cellular Neuroscience, 0, 16, .	1.8	5
3307	A dominant negative ADIPOQ mutation in a diabetic family with renal disease, hypoadiponectinemia, and hyperceramidemia. Npj Genomic Medicine, 2022, 7, .	1.7	5
3308	Whole genomic approach in mutation discovery of infantile spasms patients. Frontiers in Neurology, 0, 13, .	1.1	4
3309	GDF15 and ACE2 stratify COVID-19 patients according to severity while ACE2 mutations increase infection susceptibility. Frontiers in Cellular and Infection Microbiology, 0, 12, .	1.8	2
3310	The relationship between beta-ureidopropionase deficiency due to UPB1 variants and human phenotypes is uncertain. Molecular Genetics and Metabolism, 2022, 137, 62-67.	0.5	2
3311	Super-enhancer hypermutation alters oncogene expression in B cell lymphoma. Nature, 2022, 607, 808-815.	13.7	55
3312	The sequences of 150,119 genomes in the UK Biobank. Nature, 2022, 607, 732-740.	13.7	173
3313	Axenfeld-Rieger syndrome: more than meets the eye. Journal of Medical Genetics, 2023, 60, 368-379.	1.5	24
3315	Predicting and explaining the impact of genetic disruptions and interactions on organismal viability. Bioinformatics, 2022, 38, 4088-4099.	1.8	1
3316	The extended clinical and genetic spectrum of CTNNB1-related neurodevelopmental disorder. Frontiers in Pediatrics, 0, 10, .	0.9	5
3317	Novel RAB27A Variant Associated with Late-Onset Hemophagocytic Lymphohistiocytosis Alters Effector Protein Binding. Journal of Clinical Immunology, 2022, 42, 1685-1695.	2.0	1
3318	CMDB: the comprehensive population genome variation database of China. Nucleic Acids Research, 2023, 51, D890-D895.	6.5	4
3319	The ERICH3 rs11580409 polymorphism is associated with 6-month antidepressant response in depressed patients. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2022, 119, 110608.	2.5	0
3320	<sc>Î±â€“Synuclein V15A</sc> Variant in Familial Parkinson's Disease Exhibits a Weaker Lipidâ€“Binding Property. Movement Disorders, 2022, 37, 2075-2085.	2.2	11
3321	Deep Molecular Characterization of Milder Spinal Muscular Atrophy Patients Carrying the c.859G>C Variant in SMN2. International Journal of Molecular Sciences, 2022, 23, 8289.	1.8	6

#	ARTICLE	IF	CITATIONS
3323	A recurrent single-amino acid deletion (p.Glu500del) in the head domain of β -cardiac myosin in two unrelated boys presenting with polyhydramnios, congenital axial stiffness and skeletal myopathy. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, .	1.2	1
3324	Evidence of Two Novel LAMA2 Variants in a Patient With Muscular Dystrophy: Facing the Challenges of a Certain Diagnosis. <i>Frontiers in Neurology</i> , 0, 13, .	1.1	0
3325	Clinical Validation of Genome Reference Consortium Human Build 38 in a Laboratory Utilizing Next-Generation Sequencing Technologies. <i>Clinical Chemistry</i> , 2022, 68, 1177-1183.	1.5	5
3326	Genetic and immunohistochemical profiling of NK/T-cell lymphomas reveals prognostically relevant <i>BCOR</i> -MYC association. <i>Blood Advances</i> , 2023, 7, 178-189.	2.5	6
3327	CRISPR DNA Base Editing Strategies for Treating Retinitis Pigmentosa Caused by Mutations in Rhodopsin. <i>Genes</i> , 2022, 13, 1327.	1.0	5
3328	FUS-P525L Juvenile Amyotrophic Lateral Sclerosis and Intellectual Disability. <i>Neurology: Genetics</i> , 2022, 8, .	0.9	2
3329	Loss-of-function, gain-of-function and dominant-negative mutations have profoundly different effects on protein structure. <i>Nature Communications</i> , 2022, 13, .	5.8	82
3330	Spatially Distinct Genetic Determinants of Aortic Dimensions Influence Risks of Aneurysm and Stenosis. <i>Journal of the American College of Cardiology</i> , 2022, 80, 486-497.	1.2	15
3331	Comprehensive mapping of mutations in the C9ORF72 that affect folding and binding to SMCR8 protein. <i>Process Biochemistry</i> , 2022, 121, 312-321.	1.8	3
3332	Sitosterolemia. <i>Advances in Clinical Chemistry</i> , 2022, , 145-169.	1.8	6
3333	Whole exome sequencing identifies novel inherited genetic variants in tetralogy of Fallot. <i>Journal of Thoracic Disease</i> , 2022, .	0.6	0
3334	Gain-of-function mutations in <i>ALPK1</i> cause an NF- κ B-mediated autoinflammatory disease: functional assessment, clinical phenotyping and disease course of patients with ROSAH syndrome. <i>Annals of the Rheumatic Diseases</i> , 2022, 81, 1453-1464.	0.5	19
3335	Genetic predisposition and evolutionary traces of pediatric cancer risk: a prospective 5-year population-based genome sequencing study of children with CNS tumors. <i>Neuro-Oncology</i> , 2023, 25, 761-773.	0.6	7
3336	Rare loss of function variants in the hepatokine gene <i>INHBE</i> protect from abdominal obesity. <i>Nature Communications</i> , 2022, 13, .	5.8	15
3338	Gene-based burden analysis of damaging private variants in <i>PRKN</i> , <i>PARK7</i> and <i>PINK1</i> in Parkinson's disease cohorts of European descent. <i>Neurobiology of Aging</i> , 2022, 119, 136-138.	1.5	0
3339	Clinical impact of whole-genome sequencing in patients with early-onset dementia. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2022, 93, 1181-1189.	0.9	7
3340	A heterozygous <i>GRID2</i> mutation in autosomal dominant cerebellar ataxia. <i>Human Genome Variation</i> , 2022, 9, .	0.4	2
3341	Chromosomal Numerical Aberrations and Rare Copy Number Variation in Patients with Inflammatory Bowel Disease. <i>Journal of Crohn's and Colitis</i> , 0, , .	0.6	0

#	ARTICLE	IF	CITATIONS
3342	Does a rare mutation in PTPRA contribute to the development of Parkinson's disease in an Australian multi-incident family?. PLoS ONE, 2022, 17, e0271499.	1.1	0
3343	Identifying phenotypic expansions for congenital diaphragmatic hernia plus (<scp>CDH</scp>+) using <scp>DECIPHER</scp> data. American Journal of Medical Genetics, Part A, 2022, 188, 2958-2968.	0.7	2
3344	MicroRNA and MicroRNA-Target Variants Associated with Autism Spectrum Disorder and Related Disorders. Genes, 2022, 13, 1329.	1.0	6
3345	Inflammatory linear verrucous epidermal nevus (<scp>ILVEN</scp>) encompasses a spectrum of inflammatory mosaic disorders. Pediatric Dermatology, 2022, 39, 903-907.	0.5	10
3346	Gene-Based Variant Analysis of Whole-Exome Sequencing in Relation to Eosinophil Count. Frontiers in Immunology, 0, 13, .	2.2	3
3347	MYT1L in the making: emerging insights on functions of a neurodevelopmental disorder gene. Translational Psychiatry, 2022, 12, .	2.4	8
3348	Genetic variation as a long-distance modulator of RAD21 expression in humans. Scientific Reports, 2022, 12, .	1.6	1
3349	Discovering the drivers of clonal hematopoiesis. Nature Communications, 2022, 13, .	5.8	33
3350	HEM1 Actin Immunodysregulatory Disorder: Genotypes, Phenotypes, and Future Directions. Journal of Clinical Immunology, 2022, 42, 1583-1592.	2.0	5
3351	Aggregated Genomic Data as Cohort-Specific Allelic Frequencies can Boost Variants and Genes Prioritization in Non-Solved Cases of Inherited Retinal Dystrophies. International Journal of Molecular Sciences, 2022, 23, 8431.	1.8	4
3352	Extreme purifying selection against point mutations in the human genome. Nature Communications, 2022, 13, .	5.8	14
3353	ClinPharmSeq: A targeted sequencing panel for clinical pharmacogenetics implementation. PLoS ONE, 2022, 17, e0272129.	1.1	13
3354	<i>ACE2</i> and <i>TMPRSS2</i> SARS-CoV-2 infectivity genes: deep mutational scanning and characterization of missense variants. Human Molecular Genetics, 2022, 31, 4183-4192.	1.4	3
3355	Structural Insights into the Role of $\hat{2}3$ nAChR Subunit in the Activation of Nicotinic Receptors. Molecules, 2022, 27, 4642.	1.7	1
3356	X-CAP improves pathogenicity prediction of stopgain variants. Genome Medicine, 2022, 14, .	3.6	0
3357	Dairying, diseases and the evolution of lactase persistence in Europe. Nature, 2022, 608, 336-345.	13.7	54
3358	Cryo-EM structures of human fucosidase FucA1 reveal insight into substrate recognition and catalysis. Structure, 2022, 30, 1443-1451.e5.	1.6	6
3360	<i>CAPRIN1</i> haploinsufficiency causes a neurodevelopmental disorder with language impairment, ADHD and ASD. Brain, 2023, 146, 534-548.	3.7	8

#	ARTICLE	IF	CITATIONS
3361	Whole genome sequence association analysis of fasting glucose and fasting insulin levels in diverse cohorts from the NHLBI TOPMed program. <i>Communications Biology</i> , 2022, 5, .	2.0	5
3363	A Fast and Robust Strategy to Remove Variant-Level Artifacts in Alzheimer Disease Sequencing Project Data. <i>Neurology: Genetics</i> , 2022, 8, .	0.9	4
3364	Similar Rates of Deleterious Copy Number Variants in Early-Onset Psychosis and Autism Spectrum Disorder. <i>American Journal of Psychiatry</i> , 2022, 179, 853-861.	4.0	10
3365	Neurodevelopmental copy-number variants: A roadmap to improving outcomes by uniting patient advocates, researchers, and clinicians for collective impact. <i>American Journal of Human Genetics</i> , 2022, 109, 1353-1365.	2.6	9
3366	ADGRL1 haploinsufficiency causes a variable spectrum of neurodevelopmental disorders in humans and alters synaptic activity and behavior in a mouse model. <i>American Journal of Human Genetics</i> , 2022, 109, 1436-1457.	2.6	14
3367	Inframe insertion and splice site variants in <i>MFGE8</i> associate with protection against coronary atherosclerosis. <i>Communications Biology</i> , 2022, 5, .	2.0	9
3368	Loss-of-function variants in <i>SAT1</i> cause X-linked childhood-onset systemic lupus erythematosus. <i>Annals of the Rheumatic Diseases</i> , 2022, 81, 1712-1721.	0.5	6
3370	Cell-type-specific cis-eQTLs in eight human brain cell types identify novel risk genes for psychiatric and neurological disorders. <i>Nature Neuroscience</i> , 2022, 25, 1104-1112.	7.1	78
3371	Strategies to safely target widely expressed soluble adenylyl cyclase for contraception. <i>Frontiers in Pharmacology</i> , 0, 13, .	1.6	10
3372	Transcriptomic effects of propranolol and primidone converge on molecular pathways relevant to essential tremor. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	4
3374	Discovery and validation of dominantly inherited Alzheimer's disease mutations in populations from Latin America. <i>Alzheimer's Research and Therapy</i> , 2022, 14, .	3.0	2
3377	Three Newly Recognized Likely Pathogenic Gene Variants Associated with Hereditary Transthyretin Amyloidosis. <i>Neurology and Therapy</i> , 0, , .	1.4	1
3379	Curating the gnomAD database: Report of novel variants in the thyroid peroxidase gene using in silico bioinformatics algorithms and a literature review. <i>Molecular and Cellular Endocrinology</i> , 2022, 558, 111748.	1.6	0
3380	Disease Progression of WHIM Syndrome in an International Cohort of 66 Pediatric and Adult Patients. <i>Journal of Clinical Immunology</i> , 2022, 42, 1748-1765.	2.0	9
3381	De novo putative loss-of-function variants in <i>TAF4</i> are associated with a neurodevelopmental disorder. <i>Human Mutation</i> , 2022, 43, 1844-1851.	1.1	4
3382	Analysis of <i>COL7A1</i> pathogenic variants in a large cohort of dystrophic epidermolysis bullosa patients from Argentina reveals a new genotype-phenotype correlation. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 3153-3161.	0.7	2
3383	Compendium of proteins containing segments that exhibit zero-tolerance to amino acid variation in humans. <i>Protein Science</i> , 2022, 31, .	3.1	0
3384	CHDbase: A Comprehensive Knowledgebase for Congenital Heart Disease-related Genes and Clinical Manifestations. <i>Genomics, Proteomics and Bioinformatics</i> , 2023, 21, 216-227.	3.0	4

#	ARTICLE	IF	CITATIONS
3385	Congenital amegakaryocytic thrombocytopenia presenting with a new thrombopoietin receptor (MPL) pathogenic variant: An instructive neonatal case. <i>Journal of Paediatrics and Child Health</i> , 0, , .	0.4	0
3387	Whole-genome sequencing identifies new candidate genes for nonobstructive azoospermia. <i>Andrology</i> , 2022, 10, 1605-1624.	1.9	12
3390	Emerging phenotypes linked to variants in SAMD9 and MIRAGE syndrome. <i>Frontiers in Endocrinology</i> , 0, 13, .	1.5	4
3391	De novo variants in genes regulating stress granule assembly associate with neurodevelopmental disorders. <i>Science Advances</i> , 2022, 8, .	4.7	16
3392	The whole blood transcriptional regulation landscape in 465 COVID-19 infected samples from Japan COVID-19 Task Force. <i>Nature Communications</i> , 2022, 13, .	5.8	9
3393	Comparative transcriptome in large-scale human and cattle populations. <i>Genome Biology</i> , 2022, 23, .	3.8	11
3395	Approach to the patient with a variant of uncertain significance on genetic testing. <i>Clinical Endocrinology</i> , 0, , .	1.2	3
3397	Spatially restricted drivers and transitional cell populations cooperate with the microenvironment in untreated and chemo-resistant pancreatic cancer. <i>Nature Genetics</i> , 2022, 54, 1390-1405.	9.4	68
3398	Performance evaluation of differential splicing analysis methods and splicing analytics platform construction. <i>Nucleic Acids Research</i> , 2022, 50, 9115-9126.	6.5	11
3399	Ultra-deep sequencing validates safety of CRISPR/Cas9 genome editing in human hematopoietic stem and progenitor cells. <i>Nature Communications</i> , 2022, 13, .	5.8	22
3402	Differences in the genetic architecture of common and rare variants in childhood, persistent and late-diagnosed attention-deficit hyperactivity disorder. <i>Nature Genetics</i> , 2022, 54, 1117-1124.	9.4	27
3403	Pan-African genome demonstrates how population-specific genome graphs improve high-throughput sequencing data analysis. <i>Nature Communications</i> , 2022, 13, .	5.8	5
3404	Whole Genome Association Study of the Plasma Metabolome Identifies Metabolites Linked to Cardiometabolic Disease in Black Individuals. <i>Nature Communications</i> , 2022, 13, .	5.8	19
3405	Redefining germline predisposition in children with molecularly characterized ependymoma: a population-based 20-year cohort. <i>Acta Neuropathologica Communications</i> , 2022, 10, .	2.4	9
3406	1029 genomes of self-declared healthy individuals from India reveal prevalent and clinically relevant cardiac ion channelopathy variants. <i>Human Genomics</i> , 2022, 16, .	1.4	0
3407	Structural basis for receptor selectivity and inverse agonism in S1P5 receptors. <i>Nature Communications</i> , 2022, 13, .	5.8	6
3408	Compound-heterozygous <i>GRIN2A</i> null variants associated with severe developmental and epileptic encephalopathy. <i>Epilepsia</i> , 2022, 63, .	2.6	4
3410	Systematic single-variant and gene-based association testing of thousands of phenotypes in 394,841 UK Biobank exomes. <i>Cell Genomics</i> , 2022, 2, 100168.	3.0	89

#	ARTICLE	IF	CITATIONS
3411	Transposon-activated POU5F1B promotes colorectal cancer growth and metastasis. <i>Nature Communications</i> , 2022, 13, .	5.8	7
3412	Ultrarare Coding Variants and Cognitive Function in Schizophrenia. <i>JAMA Psychiatry</i> , 2022, 79, 963.	6.0	5
3413	Comprehensive cancer predisposition testing within the prospective MASTER trial identifies hereditary cancer patients and supports treatment decisions for rare cancers. <i>Annals of Oncology</i> , 2022, 33, 1186-1199.	0.6	21
3414	Heterozygous <i>BRCA1</i> and <i>BRCA2</i> and Mismatch Repair Gene Pathogenic Variants in Children and Adolescents With Cancer. <i>Journal of the National Cancer Institute</i> , 2022, 114, 1523-1532.	3.0	21
3415	Monoallelic pathogenic <i>ALG5</i> variants cause atypical polycystic kidney disease and interstitial fibrosis. <i>American Journal of Human Genetics</i> , 2022, 109, 1484-1499.	2.6	24
3417	Clonal hematopoiesis and risk of prostate cancer in large samples of European ancestry men. <i>Human Molecular Genetics</i> , 2023, 32, 489-495.	1.4	1
3418	<i>GIGYF1</i> disruption associates with autism and impaired IGF-1R signaling. <i>Journal of Clinical Investigation</i> , 2022, 132, .	3.9	10
3420	A novel missense variant in <i>endothelin2</i> (<i>EDN2</i>) causes a growth and respiratory lethal syndrome in bovine. <i>Animal Genetics</i> , 0, , .	0.6	2
3421	Novel Homozygous <i>TTI2</i> Variant Causing Autosomal Recessive Syndromic Intellectual Disability and Primary Microcephaly from Pakistan: A Case Report (Exome Report). <i>Case Reports in Genetics</i> , 2022, 2022, 1-5.	0.1	0
3422	Circulating interleukin-38 concentrations in healthy adults. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	2
3423	Estimating diagnostic noise in panel-based genomic analysis. <i>Genetics in Medicine</i> , 2022, 24, 2042-2050.	1.1	7
3424	An intermediate-effect size variant in <i>LIMOD</i> confers risk for chronic kidney disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	19
3425	Immunodeficiency, autoimmunity, and increased risk of B cell malignancy in humans with <i>TRAF3</i> mutations. <i>Science Immunology</i> , 2022, 7, .	5.6	9
3426	A homozygous variant in the <i>GPIHBP1</i> gene in a child with severe hypertriglyceridemia and a systematic literature review. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1
3427	GWAS meta-analysis of intrahepatic cholestasis of pregnancy implicates multiple hepatic genes and regulatory elements. <i>Nature Communications</i> , 2022, 13, .	5.8	11
3428	<i>FIBCD1</i> is an endocytic <i>GAG</i> receptor associated with a novel neurodevelopmental disorder. <i>EMBO Molecular Medicine</i> , 2022, 14, .	3.3	9
3429	Pathogenic variants damage cell composition and single cell transcription in cardiomyopathies. <i>Science</i> , 2022, 377, .	6.0	76
3430	A survey of genome-wide association studies, polygenic scores and UK Biobank highlights resources for autoimmune disease genetics. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	4

#	ARTICLE	IF	CITATIONS
3431	TP53-dependent toxicity of CRISPR/Cas9 cuts is differential across genomic loci and can confound genetic screening. <i>Nature Communications</i> , 2022, 13, .	5.8	28
3432	Multi-Omic Investigations of a 17q11.21 Translocation Links MINK1 Disruption to Autism, Epilepsy and Osteoporosis. <i>International Journal of Molecular Sciences</i> , 2022, 23, 9392.	1.8	2
3433	Heterozygous variants in MYH10 associated with neurodevelopmental disorders and congenital anomalies with evidence for primary cilia-dependent defects in Hedgehog signaling. <i>Genetics in Medicine</i> , 2022, 24, 2065-2078.	1.1	2
3434	Cross-species identification of cancer resistance-associated genes that may mediate human cancer risk. <i>Science Advances</i> , 2022, 8, .	4.7	5
3435	Candidate genes for infertility: an in-silico study based on cytogenetic analysis. <i>BMC Medical Genomics</i> , 2022, 15, .	0.7	3
3436	Diagnostic yield and clinical relevance of expanded genetic testing for cancer patients. <i>Genome Medicine</i> , 2022, 14, .	3.6	11
3437	The recurrent <i>TCF4</i> missense variant p.(Arg389Cys) causes a neurodevelopmental disorder overlapping with but not typical for Pitt-Hopkins syndrome. <i>Clinical Genetics</i> , 0, .	1.0	3
3438	Integrating de novo and inherited variants in 42,607 autism cases identifies mutations in new moderate-risk genes. <i>Nature Genetics</i> , 2022, 54, 1305-1319.	9.4	110
3439	Accelerating SLC Transporter Research: Streamlining Knowledge and Validated Tools. <i>Clinical Pharmacology and Therapeutics</i> , 2022, 112, 439-442.	2.3	5
3440	Germline Testing for the Evaluation of Hereditary Cancer Predisposition. <i>Clinics in Laboratory Medicine</i> , 2022, 42, 497-506.	0.7	0
3441	TREM2 Gene Compound Heterozygosity in Neurodegenerative Disorders. <i>Journal of Alzheimer's Disease</i> , 2022, , 1-9.	1.2	0
3443	Genetic predisposition to portal sinusoidal vascular disorder: A functional genomic-based, multigenerational family study. <i>Hepatology</i> , 2023, 77, 501-511.	3.6	7
3445	Molecular determinants of outcomes in relapsed or refractory mantle cell lymphoma treated with ibrutinib or temsirolimus in the MCL3001 (RAY) trial. <i>Leukemia</i> , 2022, 36, 2479-2487.	3.3	3
3446	mvPPT: A Highly Efficient and Sensitive Pathogenicity Prediction Tool for Missense Variants. <i>Genomics, Proteomics and Bioinformatics</i> , 2023, 21, 414-426.	3.0	2
3447	Further clinical delineation of microcephaly-capillary malformation syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 3350-3357.	0.7	2
3448	Germline genetic biomarkers to stratify patients for personalized radiation treatment. <i>Journal of Translational Medicine</i> , 2022, 20, .	1.8	5
3449	A Tale of Native American Whole-Genome Sequencing and Other Technologies. <i>Diversity</i> , 2022, 14, 647.	0.7	0
3450	A cross-disorder dosage sensitivity map of the human genome. <i>Cell</i> , 2022, 185, 3041-3055.e25.	13.5	117

#	ARTICLE	IF	CITATIONS
3451	Adult-Onset Focal Segmental Glomerulosclerosis With Steroid-Dependent Nephrotic Syndrome Caused by a Novel TBC1D8B Variant: A Case Report and Literature Review. American Journal of Kidney Diseases, 2022, , .	2.1	2
3454	Impact of 100 LRRK2 variants linked to Parkinson's disease on kinase activity and microtubule binding. Biochemical Journal, 2022, 479, 1759-1783.	1.7	39
3455	Wide range of phenotypic severity in individuals with late truncations unique to the predominant <i>CDKL5</i> transcript in the brain. American Journal of Medical Genetics, Part A, 0, , .	0.7	2
3457	Scalable approaches for functional analyses of whole-genome sequencing non-coding variants. Human Molecular Genetics, 2022, 31, R62-R72.	1.4	2
3458	Scaled Process Priors for Bayesian Nonparametric Estimation of the Unseen Genetic Variation. Journal of the American Statistical Association, 2024, 119, 320-331.	1.8	2
3459	Consequences of a Rare Complement Factor H Variant for Age-Related Macular Degeneration in the Amish. , 2022, 63, 8.		0
3460	Altered gene expression profiles impair the nervous system development in individuals with 15q13.3 microdeletion. Scientific Reports, 2022, 12, .	1.6	1
3461	Rapidly evolving viral motifs mostly target biophysically constrained binding pockets of host proteins. Cell Reports, 2022, 40, 111212.	2.9	10
3462	Expansion of the clinical and neuroimaging spectrum associated with <i>NDUFS8</i> -related disorder. JIMD Reports, 0, , .	0.7	0
3463	Hemizyosity can reveal variant pathogenicity on the X-chromosome. Human Genetics, 2023, 142, 11-19.	1.8	2
3464	Real-World Evaluation of a Population Germline Genetic Screening Initiative for Family Medicine Patients. Journal of Personalized Medicine, 2022, 12, 1297.	1.1	1
3465	CCAS: One-stop and comprehensive annotation system for individual cancer genome at multi-omics level. Frontiers in Genetics, 0, 13, .	1.1	2
3466	Can tandem alternative splicing and evasion of premature termination codon surveillance contribute to attenuated <i>Peutz-Jeghers</i> syndrome?. American Journal of Medical Genetics, Part A, 2022, 188, 3089-3095.	0.7	2
3467	Meier's Gorlin Syndrome: Clinical Misdiagnosis, Genetic Testing and Functional Analysis of <i>ORC6</i> Mutations and the Development of a Prenatal Test. International Journal of Molecular Sciences, 2022, 23, 9234.	1.8	2
3468	Heterozygous variants in <i>SIX3</i> and <i>POU1F1</i> cause pituitary hormone deficiency in mouse and man. Human Molecular Genetics, 2023, 32, 367-385.	1.4	1
3469	Performance evaluation of computational methods for splice-disrupting variants and improving the performance using the machine learning-based framework. Briefings in Bioinformatics, 0, , .	3.2	5
3471	AutoCaSc: Prioritizing candidate genes for neurodevelopmental disorders. Human Mutation, 2022, 43, 1795-1807.	1.1	5
3472	Autosomal recessive inheritance of a novel missense mutation of <i>ITGB4</i> for Epidermolysis-Bullosa pyloric-atresia: a case report. Molecular Genetics and Genomics, 0, , .	1.0	0

#	ARTICLE	IF	CITATIONS
3473	<i>NOD2</i> in Crohn's Disease—Unfinished Business. <i>Journal of Crohn's and Colitis</i> , 2023, 17, 450-458.	0.6	8
3474	Prediction of infectivity of SARS-CoV-2 virus based on Spike-hACE-2 interaction. <i>VirusDisease</i> , 0, , .	1.0	0
3475	Shared and Distinct Functional Effects of Patient-Specific <i>Tbr1</i> Mutations on Cortical Development. <i>Journal of Neuroscience</i> , 2022, 42, 7166-7181.	1.7	5
3476	AnFISA: An open-source computational platform for the analysis of sequencing data for rare genetic disease. <i>Journal of Biomedical Informatics</i> , 2022, 133, 104174.	2.5	0
3477	Genetics of circadian rhythms and sleep in human health and disease. <i>Nature Reviews Genetics</i> , 2023, 24, 4-20.	7.7	55
3479	Pathogenic genetic variants identified in Australian families with paediatric cataract. <i>BMJ Open Ophthalmology</i> , 2022, 7, e001064.	0.8	7
3480	Molecular and cellular evolution of the primate dorsolateral prefrontal cortex. <i>Science</i> , 2022, 377, .	6.0	61
3481	Loss of function of OTUD7A in the schizophrenia-associated 15q13.3 deletion impairs synapse development and function in human neurons. <i>American Journal of Human Genetics</i> , 2022, 109, 1500-1519.	2.6	5
3482	Whole Exome Sequencing in Patients With Ectopic Posterior Pituitary. <i>Journal of the Endocrine Society</i> , 2022, 6, .	0.1	2
3483	FOXI3 haploinsufficiency contributes to low T-cell receptor excision circles and T-cell lymphopenia. <i>Journal of Allergy and Clinical Immunology</i> , 2022, 150, 1556-1562.	1.5	6
3485	Clinical significance of genetic variation in hypertrophic cardiomyopathy: comparison of computational tools to prioritize missense variants. <i>Frontiers in Cardiovascular Medicine</i> , 0, 9, .	1.1	5
3486	Expanding ACMG variant classification guidelines into a general framework. <i>Human Genomics</i> , 2022, 16, .	1.4	27
3487	Genetic factors help explain the variable responses of young children with cystic fibrosis to vitamin D supplements. <i>Clinical Nutrition ESPEN</i> , 2022, 51, 367-376.	0.5	3
3488	Genomic features of renal cell carcinoma developed during end-stage renal disease and dialysis. <i>Human Molecular Genetics</i> , 2023, 32, 290-303.	1.4	4
3489	Prevalence of Fabry disease-causing variants in the UK Biobank. <i>Journal of Medical Genetics</i> , 2023, 60, 391-396.	1.5	10
3490	Clinical and Genetic Characteristics of Multiple Epiphyseal Dysplasia Type 4. <i>Genes</i> , 2022, 13, 1512.	1.0	2
3491	COVID-19 in pediatrics: Genetic susceptibility. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	5
3492	Cancer-driving mutations are enriched in genic regions intolerant to germline variation. <i>Science Advances</i> , 2022, 8, .	4.7	2

#	ARTICLE	IF	CITATIONS
3494	Whole exome sequencing reveals novel risk genes of pituitary neuroendocrine tumors. PLoS ONE, 2022, 17, e0265306.	1.1	2
3497	Expanding the phenotypic spectrum of <sc><i>COLEC10</i></sc>-Related 3MC</sc> syndrome: A glimpse into <sc><i>COLEC10</i></sc>-Related 3MC</sc> syndrome in the Ashkenazi Jewish population. American Journal of Medical Genetics, Part A, 2022, 188, 3110-3117.	0.7	1
3498	<sc><i>EEF1A2</i></sc> pathogenic variant presenting in an infant with failure to thrive and frequent apneas requiring respiratory support. American Journal of Medical Genetics, Part A, 2022, 188, 3106-3109.	0.7	1
3499	Identification and genetic analysis of rare variants in myosin family genes in 412 Han Chinese congenital heart disease patients. Molecular Genetics & Genomic Medicine, 0, , .	0.6	1
3500	Challenges and opportunities associated with rare-variant pharmacogenomics. Trends in Pharmacological Sciences, 2022, 43, 852-865.	4.0	14
3501	Rare coding variation provides insight into the genetic architecture and phenotypic context of autism. Nature Genetics, 2022, 54, 1320-1331.	9.4	155
3503	New Insights into Renal Failure in a Cohort of 317 Patients with Autosomal Dominant Forms of Alport Syndrome: Report of Two Novel Heterozygous Mutations in COL4A3. Journal of Clinical Medicine, 2022, 11, 4883.	1.0	3
3505	Genome wide association study identifies a novel variant associated with tacrolimus trough concentration in Chinese renal transplant recipients. Clinical and Translational Science, 0, , .	1.5	2
3506	Post-translational control of beige fat biogenesis by PRDM16 stabilization. Nature, 2022, 609, 151-158.	13.7	31
3507	Most frequently harboured missense variants of hACE2 across different populations exhibit varying patterns of binding interaction with spike glycoproteins of emerging SARS-CoV-2 of different lineages. Computers in Biology and Medicine, 2022, 148, 105903.	3.9	5
3508	Genetics of multiple sclerosis: lessons from polygenicity. Lancet Neurology, The, 2022, 21, 830-842.	4.9	26
3509	De novo SCN3A missense variant associated with self-limiting generalized epilepsy with fever sensitivity. European Journal of Medical Genetics, 2022, 65, 104577.	0.7	0
3510	Hepatic solute carrier transporters and drug therapy: Regulation of expression and impact of genetic variation. , 2022, 238, 108268.		8
3511	Severe spinal cord hypoplasia due to a novel ATAD3A compound heterozygous deletion. Molecular Genetics and Metabolism Reports, 2022, 33, 100912.	0.4	3
3513	Prevalence estimates of putatively pathogenic leptin variants in the gnomAD database. PLoS ONE, 2022, 17, e0266642.	1.1	2
3515	Identifying the BRCA1 c.-107A→T variant in Dutch patients with a tumor BRCA1 promoter hypermethylation. Familial Cancer, 2023, 22, 151-154.	0.9	1
3516	The recurrent de novo c.2011C→T missense variant in MTSS2 causes syndromic intellectual disability. American Journal of Human Genetics, 2022, 109, 1923-1931.	2.6	7
3517	Systems analysis of de novo mutations in congenital heart diseases identified a protein network in the hypoplastic left heart syndrome. Cell Systems, 2022, 13, 895-910.e4.	2.9	3

#	ARTICLE	IF	CITATIONS
3518	Mutations in SCNM1 cause orofacioidigital syndrome due to minor intron splicing defects affecting primary cilia. <i>American Journal of Human Genetics</i> , 2022, 109, 1828-1849.	2.6	5
3519	Analyses of rare predisposing variants of lung cancer in 6,004 whole genomes in Chinese. <i>Cancer Cell</i> , 2022, 40, 1223-1239.e6.	7.7	23
3521	Genetic variability shapes the alternative pathway complement activity and predisposition to complement-related diseases. <i>Immunological Reviews</i> , 2023, 313, 71-90.	2.8	15
3522	Expanding the pre- and postnatal phenotype of WASHC5 and CCDC22 -related Ritscher-Schinzel syndromes. <i>European Journal of Medical Genetics</i> , 2022, 65, 104624.	0.7	3
3523	Convergent and Divergent Contribution of Vitamin A and Oxytocin to Autism Spectrum Disorder Aetiology. <i>SSRN Electronic Journal</i> , 0, , .	0.4	0
3524	Heterozygosity for Neuronal Ceroid Lipofuscinosis predisposes to Bipolar Disorder. <i>Revista Brasileira De Psiquiatria</i> , 2022, , .	0.9	1
3525	Pathogenic Variants Spectrum and Allele Frequency of the <i>CFTR</i> Gene in Asians. <i>Allergy, Asthma and Immunology Research</i> , 2022, 14, 444.	1.1	0
3526	ChemBioPort: an online portal to navigate the structure, function and chemical inhibition of the human proteome. <i>Database: the Journal of Biological Databases and Curation</i> , 2022, 2022, .	1.4	2
3527	SNPs Classification and Terminology: dbSNP Reference SNP (rs) Gene and Consequence Annotation. , 2022, , 3-12.		0
3528	Association of Alpha 1 Antitrypsin Deficiency with COVID-19 Mortality: Basis for Clinical Trials. , 2022, , 325-336.		0
3529	Single Nucleotide Polymorphisms (SNPs). , 2022, , .		1
3530	The Pioneer Advantage: Filling the blank spots on the map of genome diversity in Europe. <i>GigaScience</i> , 2022, 11, .	3.3	3
3532	Isolated MLH1 Loss by Immunohistochemistry Because of Benign Germline <i>MLH1</i> Polymorphisms. <i>JCO Precision Oncology</i> , 2022, , .	1.5	2
3533	PRKG1 mutation identified by whole-exome sequencing: a potential genetic etiology for He-Zhao deficiency. <i>Journal of Bio-X Research</i> , 2022, 5, 118-124.	0.3	0
3534	Conserved patterns across ion channels correlate with variant pathogenicity and clinical phenotypes. <i>Brain</i> , 2023, 146, 923-934.	3.7	11
3535	Genomes from a medieval mass burial show Ashkenazi-associated hereditary diseases pre-date the 12th century. <i>Current Biology</i> , 2022, 32, 4350-4359.e6.	1.8	3
3536	When LUCA met gnomAD: genetic constraints on universal genes in humans. <i>Intractable and Rare Diseases Research</i> , 2022, 11, 149-152.	0.3	0
3537	Unfolding of Novel Independent Missense Mutations in VAMP2 and AGRN and Their Collective Role in Global Developmental Delay: A Case Report. <i>Cureus</i> , 2022, , .	0.2	0

#	ARTICLE	IF	CITATIONS
3538	Principal Component Analyses (PCA)-based findings in population genetic studies are highly biased and must be reevaluated. <i>Scientific Reports</i> , 2022, 12, .	1.6	48
3539	A splice site variant in <i>TCTN3</i> underlies an atypical form of orofaciogigital syndrome IV. <i>Annals of Human Genetics</i> , 0, , .	0.3	2
3540	A Novel Mutation in the TSEN2 Gene Among Two Iranian Families with Pontocerebellar Hypoplasia Type 2B. <i>Jentashapir Journal of Cellular and Molecular Biology</i> , 2022, 13, .	0.1	0
3542	Defining the HIV Capsid Binding Site of Nucleoporin 153. <i>MSphere</i> , 2022, 7, .	1.3	2
3543	Snail Track Lesion with Flat Keratometry in Anterior Segment Dysgenesis Caused by a Novel FOXC1 Variant. <i>Journal of Clinical Medicine</i> , 2022, 11, 5166.	1.0	0
3544	Loss of Protein Function Causing Severe Phenotypes of Female-Restricted Wieacker Wolff Syndrome due to a Novel Nonsense Mutation in the ZC4H2 Gene. <i>Genes</i> , 2022, 13, 1558.	1.0	3
3545	Diverse mutations in autism-related genes and their expression in the developing brain. <i>Nature Genetics</i> , 2022, 54, 1263-1264.	9.4	1
3546	Loss of seryl-tRNA synthetase (<i>SARS1</i>) causes complex spastic paraplegia and cellular senescence. <i>Journal of Medical Genetics</i> , 2022, 59, 1227-1233.	1.5	4
3547	Newly identified disorder of copper metabolism caused by variants in <i>CTR1</i> , a high-affinity copper transporter. <i>Human Molecular Genetics</i> , 2022, 31, 4121-4130.	1.4	12
3548	Characterization of ACE2 naturally occurring missense variants: impact on subcellular localization and trafficking. <i>Human Genomics</i> , 2022, 16, .	1.4	4
3549	Ancestry-driven recalibration of tumor mutational burden and disparate clinical outcomes in response to immune checkpoint inhibitors. <i>Cancer Cell</i> , 2022, 40, 1161-1172.e5.	7.7	44
3551	Huntington's disease age at motor onset is modified by the tandem hexamer repeat in TCERG1. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	5
3552	Identification and in-silico characterization of splice-site variants from a large cardiogenetic national registry. <i>European Journal of Human Genetics</i> , 2023, 31, 512-520.	1.4	3
3553	Ultra-Rare BRD9 Loss-of-Function Variants Limit the Antiviral Action of Interferon. <i>Scientific Reports</i> , 2022, 12, .	1.6	1
3554	Dissection of mendelian predisposition and complex genetic architecture of craniovertebral junction malformation. <i>Human Genetics</i> , 0, , .	1.8	0
3555	<i>PTPA</i> variants and impaired PP2A activity in early-onset parkinsonism with intellectual disability. <i>Brain</i> , 2023, 146, 1496-1510.	3.7	7
3557	Association analyses of rare variants identify two genes associated with refractive error. <i>PLoS ONE</i> , 2022, 17, e0272379.	1.1	1
3558	Genetic Architecture of Acute Myocarditis and the Overlap With Inherited Cardiomyopathy. <i>Circulation</i> , 2022, 146, 1123-1134.	1.6	46

#	ARTICLE	IF	CITATIONS
3559	Diverse ancestry whole-genome sequencing association study identifies TBX5 and PTK7 as susceptibility genes for posterior urethral valves. <i>ELife</i> , 0, 11, .	2.8	5
3560	SDC4-rs1981429 and ATM-rs228590 may provide early biomarkers of breast cancer risk. <i>Journal of Cancer Research and Clinical Oncology</i> , 0, , .	1.2	1
3561	<i>ATP6VOC</i> variants impair V-ATPase function causing a neurodevelopmental disorder often associated with epilepsy. <i>Brain</i> , 2023, 146, 1357-1372.	3.7	6
3565	Haplotyping SNPs for allele-specific gene editing of the expanded huntingtin allele using long-read sequencing. <i>Human Genetics and Genomics Advances</i> , 2023, 4, 100146.	1.0	6
3566	Haplotype sequence collection of <i>ABO</i> blood group alleles by long-read sequencing reveals putative <i>A1</i>-diagnostic variants. <i>Blood Advances</i> , 2023, 7, 878-892.	2.5	10
3569	A catalog of the genetic causes of hereditary angioedema in the Canary Islands (Spain). <i>Frontiers in Immunology</i> , 0, 13, .	2.2	2
3571	Rare copy number variation in posttraumatic stress disorder. <i>Molecular Psychiatry</i> , 2022, 27, 5062-5069.	4.1	1
3572	Radiogenomic markers enable risk stratification and inference of mutational pathway states in head and neck cancer. <i>European Journal of Nuclear Medicine and Molecular Imaging</i> , 2023, 50, 546-558.	3.3	3
3573	A novel variant in <scp><i>GATM</i></scp> causes idiopathic renal Fanconi syndrome and predicts progression to end-stage kidney disease. <i>Clinical Genetics</i> , 2023, 103, 214-218.	1.0	5
3574	Functional Testing of Bone Morphogenetic Protein (BMP) Pathway Variants Identified on Whole-Exome Sequencing in a Patient with Delayed-Onset Fibrodysplasia Ossificans Progressiva (FOP) Using ACVR1R206H-Specific Human Cellular and Zebrafish Models. <i>Journal of Bone and Mineral Research</i> , 2020, 37, 2058-2076.	3.1	2
3575	ENPP1 deficiency: A clinical update on the relevance of individual variants using a locus-specific patient database. <i>Human Mutation</i> , 2022, 43, 1673-1705.	1.1	1
3576	BiP inactivation due to loss of the deAMPylation function of FICD causes a motor neuron disease. <i>Genetics in Medicine</i> , 2022, 24, 2487-2500.	1.1	4
3577	GenOtoScope: Towards automating ACMG classification of variants associated with congenital hearing loss. <i>PLoS Computational Biology</i> , 2022, 18, e1009785.	1.5	1
3580	Interpreting the spectrum of gamma-secretase complex missense variation in the context of hidradenitis suppurativa—An in-silico study. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	9
3581	Genetic Heterogeneity Shapes Brain Connectivity in Psychiatry. <i>Biological Psychiatry</i> , 2023, 93, 45-58.	0.7	9
3582	Determining the origin of different variants associated with familial mediterranean fever by machine-learning. <i>Scientific Reports</i> , 2022, 12, .	1.6	1
3584	Genomic profiling for clinical decision making in Amyloid neoplasms and acute leukemia. <i>Blood</i> , 2022, 140, 2228-2247.	0.6	72
3585	More than a marker: potential pathogenic functions of MAP2. <i>Frontiers in Molecular Neuroscience</i> , 0, 15, .	1.4	26

#	ARTICLE	IF	CITATIONS
3587	Whole-exome sequencing reveals a comprehensive germline mutation landscape and identifies twelve novel predisposition genes in Chinese prostate cancer patients. <i>PLoS Genetics</i> , 2022, 18, e1010373.	1.5	4
3588	The impact of Mendelian sleep and circadian genetic variants in a population setting. <i>PLoS Genetics</i> , 2022, 18, e1010356.	1.5	2
3589	Characterization of <i>POR</i> haplotype distribution in African populations and comparison with other global populations. <i>Pharmacogenomics</i> , 2022, 23, 771-782.	0.6	1
3591	Multiplexed functional genomic assays to decipher the noncoding genome. <i>Human Molecular Genetics</i> , 2022, 31, R84-R96.	1.4	4
3592	Scalable Functional Assays for the Interpretation of Human Genetic Variation. <i>Annual Review of Genetics</i> , 2022, 56, 441-465.	3.2	18
3593	Mitochondrial Genome Variants as a Cause of Mitochondrial Cardiomyopathy. <i>Cells</i> , 2022, 11, 2835.	1.8	4
3596	A Large Case-Control Study Performed in Spanish Population Suggests That RECQL5 Is the Only RECQ Helicase Involved in Breast Cancer Susceptibility. <i>Cancers</i> , 2022, 14, 4738.	1.7	3
3597	Sequencing individual genomes with recurrent genomic disorder deletions: an approach to characterize genes for autosomal recessive rare disease traits. <i>Genome Medicine</i> , 2022, 14, .	3.6	3
3598	Pathological variants in genes associated with disorders of sex development and central causes of hypogonadism in a whole-genome reference panel of 8380 Japanese individuals. <i>Human Genome Variation</i> , 2022, 9, .	0.4	0
3599	Developing CIRdb as a catalog of natural genetic variation in the Canary Islanders. <i>Scientific Reports</i> , 2022, 12, .	1.6	1
3600	Rare Heterozygous PCSK1 Variants in Human Obesity: The Contribution of the p.Y181H Variant and a Literature Review. <i>Genes</i> , 2022, 13, 1746.	1.0	1
3601	Duplex Sequencing Uncovers Recurrent Low-frequency Cancer-associated Mutations in Infant and Childhood KMT2A-rearranged Acute Leukemia. <i>HemaSphere</i> , 2022, 6, e785.	1.2	1
3602	Dementia-related genetic variants in an Italian population of early-onset Alzheimer's disease. <i>Frontiers in Aging Neuroscience</i> , 0, 14, .	1.7	5
3603	Response to Ramos et al. <i>Genetics in Medicine</i> , 2022, , .	1.1	0
3606	Genetic analysis of CFH and MCP in Egyptian patients with immune-complex proliferative glomerulonephritis. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	0
3607	Familial Male-limited Precocious Puberty (FMPP) and Testicular Germ Cell Tumors. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 3035-3044.	1.8	4
3608	Systematic identification of intron retention associated variants from massive publicly available transcriptome sequencing data. <i>Nature Communications</i> , 2022, 13, .	5.8	8
3609	Genomic data integration and user-defined sample-set extraction for population variant analysis. <i>BMC Bioinformatics</i> , 2022, 23, .	1.2	1

#	ARTICLE	IF	CITATIONS
3610	Individualized Mini-Panel Sequencing of ctDNA Allows Tumor Monitoring in Complex Karyotype Sarcomas. <i>International Journal of Molecular Sciences</i> , 2022, 23, 10215.	1.8	1
3611	High Bone Mass Disorders: New Insights From Connecting the Clinic and the Bench. <i>Journal of Bone and Mineral Research</i> , 2020, 38, 229-247.	3.1	4
3612	Pathologic Alexander Disease with Normal GFAP Sequencing: An Autopsy Case Report and Literature Review. <i>Journal of Neuropathology and Experimental Neurology</i> , 2022, 81, 1033-1036.	0.9	2
3613	Exome sequencing efficacy and phenotypic expansions involving esophageal atresia/tracheoesophageal fistula plus. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	2
3614	Recurrent 17q12 microduplications contribute to renal disease but not diabetes. <i>Journal of Medical Genetics</i> , 2023, 60, 491-497.	1.5	1
3615	Vasor: Accurate prediction of variant effects for amino acid substitutions in multidrug resistance protein 3. <i>Hepatology Communications</i> , 2022, 6, 3098-3111.	2.0	3
3617	Estimating the prevalence of congenital disaccharidase deficiencies using allele frequencies from gnomAD. <i>Archives De Pediatrie</i> , 2022, , .	0.4	2
3618	Unique structural features govern the activity of a human mitochondrial AAA+ disaggregase, Skd3. <i>Cell Reports</i> , 2022, 40, 111408.	2.9	9
3619	A rapid turnaround gene panel for severe autoinflammation: Genetic results within 48 hours. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	1
3620	Characterization of <i>CYP2D6</i> Pharmacogenetic Variation in Sub-Saharan African Populations. <i>Clinical Pharmacology and Therapeutics</i> , 2023, 113, 643-659.	2.3	12
3621	Identifying interpretable gene-biomarker associations with functionally informed kernel-based tests in 190,000 exomes. <i>Nature Communications</i> , 2022, 13, .	5.8	6
3622	De Novo Missense Variants in <i>SLC32A1</i> Cause a Developmental and Epileptic Encephalopathy Due to Impaired GABAergic Neurotransmission. <i>Annals of Neurology</i> , 2022, 92, 958-973.	2.8	6
3623	Brief Report: Risk Variants Could Inform Early Neurodevelopmental Outcome in Children with Developmental Disabilities. <i>Journal of Autism and Developmental Disorders</i> , 0, , .	1.7	0
3624	Biallelic pathogenic variants in <i>COX11</i> are associated with an infantile-onset mitochondrial encephalopathy. <i>Human Mutation</i> , 2022, 43, 1970-1978.	1.1	4
3625	Whole-genome sequencing combined RNA-sequencing analysis of patients with mutations in SET binding protein 1. <i>Frontiers in Neuroscience</i> , 0, 16, .	1.4	2
3626	Disorder of Sex Development Due to 17-Beta-Hydroxysteroid Dehydrogenase Type 3 Deficiency: A Case Report and Review of 70 Different HSD17B3 Mutations Reported in 239 Patients. <i>International Journal of Molecular Sciences</i> , 2022, 23, 10026.	1.8	6
3627	Genetic Variation among Pharmacogenes in the Sardinian Population. <i>International Journal of Molecular Sciences</i> , 2022, 23, 10058.	1.8	1
3628	Genetic risk factors have a substantial impact on healthy life years. <i>Nature Medicine</i> , 2022, 28, 1893-1901.	15.2	24

#	ARTICLE	IF	CITATIONS
3629	Clinical Risk Score to Predict Pathogenic Genotypes in Patients With Dilated Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2022, 80, 1115-1126.	1.2	18
3630	Combined Focused Next-Generation Sequencing Assays to Guide Precision Oncology in Solid Tumors: A Retrospective Analysis from an Institutional Molecular Tumor Board. <i>Cancers</i> , 2022, 14, 4430.	1.7	7
3631	Use of race, ethnicity, and ancestry data in health research. <i>PLOS Global Public Health</i> , 2022, 2, e0001060.	0.5	32
3632	Rapid genome sequencing for pediatrics. <i>Human Mutation</i> , 2022, 43, 1507-1518.	1.1	9
3633	Local data commons: the sleeping beauty in the community of data commons. <i>BMC Bioinformatics</i> , 2022, 23, .	1.2	1
3634	Commentary: SPTBN5, encoding the β -spectrin protein, leads to a syndrome of intellectual disability, developmental delay, and seizures. <i>Frontiers in Molecular Neuroscience</i> , 0, 15, .	1.4	0
3635	Genome-wide meta-analysis for Alzheimer's disease cerebrospinal fluid biomarkers. <i>Acta Neuropathologica</i> , 2022, 144, 821-842.	3.9	38
3636	The p190 RhoGAPs, ARHGAP35, and ARHGAP5 are implicated in GnRH neuronal development: Evidence from patients with idiopathic hypogonadotropic hypogonadism, zebrafish, and in vitro GAP activity assay. <i>Genetics in Medicine</i> , 2022, 24, 2501-2515.	1.1	2
3637	Investigating a Genetic Link Between Alzheimer's Disease and CADASIL-Related Cerebral Small Vessel Disease. <i>Molecular Neurobiology</i> , 2022, 59, 7293-7302.	1.9	1
3638	Genomic and phenotypic characterization of 404 individuals with neurodevelopmental disorders caused by CTNNB1 variants. <i>Genetics in Medicine</i> , 2022, 24, 2351-2366.	1.1	12
3639	Genetic structure correlates with ethnolinguistic diversity in eastern and southern Africa. <i>American Journal of Human Genetics</i> , 2022, 109, 1667-1679.	2.6	10
3640	Computational approaches for predicting variant impact: An overview from resources, principles to applications. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	11
3641	Social and scientific motivations to move beyond groups in allele frequencies: The TOPMed experience. <i>American Journal of Human Genetics</i> , 2022, 109, 1582-1590.	2.6	1
3642	Long read sequencing and expression studies of AHDC1 deletions in Xia-Gibbs syndrome reveal a novel genetic regulatory mechanism. <i>Human Mutation</i> , 2022, 43, 2033-2053.	1.1	5
3643	Teenage-Onset Colorectal Cancers in a Digenic Cancer Predisposition Syndrome Provide Clues for the Interaction between Mismatch Repair and Polymerase ϵ Proofreading Deficiency in Tumorigenesis. <i>Biomolecules</i> , 2022, 12, 1350.	1.8	9
3644	The genetic landscape of major drug metabolizing cytochrome P450 genes—an updated analysis of population-scale sequencing data. <i>Pharmacogenomics Journal</i> , 2022, 22, 284-293.	0.9	31
3645	An assessment of prevalence of Type 1 CFI rare variants in European AMD, and why lack of broader genetic data hinders development of new treatments and healthcare access. <i>PLoS ONE</i> , 2022, 17, e0272260.	1.1	1
3646	Characterization of a novel variant in the HR1 domain of MFN2 in a patient with ataxia, optic atrophy and sensorineural hearing loss. <i>F1000Research</i> , 0, 10, 606.	0.8	1

#	ARTICLE	IF	CITATIONS
3647	A large deletion encompassing exon 2 of the ectodysplasin A (EDA) gene in a British blue crossbred calf with hypohidrotic ectodermal dysplasia. <i>Acta Veterinaria Scandinavica</i> , 2022, 64, .	0.5	2
3648	Prevalence and Phenotypic Burden of Monogenic Arrhythmias Using Integration of Electronic Health Records With Genetics. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, .	1.6	3
3649	Strategies to inhibit FGFR4 V550L-driven rhabdomyosarcoma. <i>British Journal of Cancer</i> , 2022, 127, 1939-1953.	2.9	2
3650	Comprehensive targeted next-generation sequencing in patients with slow-flow vascular malformations. <i>Journal of Human Genetics</i> , 2022, 67, 721-728.	1.1	7
3652	Newborn screening for Fabry disease in Oregon: Approaching the iceberg of <sc>A143T</sc> and variants of uncertain significance. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2022, 190, 206-214.	0.7	3
3653	A Xp22.11-p21.3 microdeletion in a three-generation family supports male lethality of POLA1 nullisomy resulting in reduced fertility of female carriers. <i>European Journal of Medical Genetics</i> , 2022, , 104628.	0.7	1
3654	Prediction of Crohnâ€™s Disease Structuring Phenotype Using a <i>NOD2</i>-derived Genomic Biomarker. <i>Inflammatory Bowel Diseases</i> , 0, , .	0.9	0
3655	Localized variation in ancestral admixture identifies pilocytic astrocytoma risk loci among Latino children. <i>PLoS Genetics</i> , 2022, 18, e1010388.	1.5	2
3656	Identifying enhancer properties associated with genetic risk for complex traits using regulome-wide association studies. <i>PLoS Computational Biology</i> , 2022, 18, e1010430.	1.5	4
3657	TP53 mutations in functional corticotroph tumors are linked to invasion and worse clinical outcome. <i>Acta Neuropathologica Communications</i> , 2022, 10, .	2.4	9
3658	Analysis of rare disruptive germline mutations in 2135 enriched BRCA-negative breast cancers excludes additional high-impact susceptibility genes. <i>Annals of Oncology</i> , 2022, 33, 1318-1327.	0.6	6
3660	Uniparental disomy screen of Irish rare disorder cohort unmask homozygous variants of clinical significance in the TMCO1 and PRKRA genes. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	4
3661	Never-homozygous genetic variants in healthy populations are potential recessive disease candidates. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	3
3662	RB1-deficient squamous cell carcinoma: the proposed source of combined Merkel cell carcinoma. <i>Modern Pathology</i> , 2022, 35, 1829-1836.	2.9	9
3663	Testing for association with rare variants in the coding and non-coding genome: RAVA-FIRST, a new approach based on CADD deleteriousness score. <i>PLoS Genetics</i> , 2022, 18, e1009923.	1.5	4
3664	A Splice Variant of the MYH7 Gene Is Causative in a Family with Isolated Left Ventricular Noncompaction Cardiomyopathy. <i>Genes</i> , 2022, 13, 1750.	1.0	2
3665	Kyphoscoliotic Ehlersâ€Danlos syndrome caused by pathogenic variants in <i>FKBP14</i> : Further insights into the phenotypic spectrum and pathogenic mechanisms. <i>Human Mutation</i> , 2022, 43, 1994-2009.	1.1	2
3666	The precision medicine process for treating rare disease using the artificial intelligence tool mediKanren. <i>Frontiers in Artificial Intelligence</i> , 0, 5, .	2.0	12

#	ARTICLE	IF	CITATIONS
3668	Immunogenetics associated with severe coccidioidomycosis. <i>JCI Insight</i> , 2022, 7, .	2.3	20
3669	Genome Sequencing and Transcriptome Profiling in Twins Discordant for Mayer-Rokitansky-Küster-Hauser Syndrome. <i>Journal of Clinical Medicine</i> , 2022, 11, 5598.	1.0	4
3670	Novel variants identified in a three-generation family with concomitant exotropia. <i>Experimental and Therapeutic Medicine</i> , 2022, 24, .	0.8	1
3671	Ten challenges for clinical translation in psychiatric genetics. <i>Nature Genetics</i> , 2022, 54, 1457-1465.	9.4	9
3672	High-coverage whole-genome sequencing of the expanded 1000 Genomes Project cohort including 602 trios. <i>Cell</i> , 2022, 185, 3426-3440.e19.	13.5	285
3673	Low-level constitutional mosaicism of BRCA1 in two women with young onset ovarian cancer. <i>Hereditary Cancer in Clinical Practice</i> , 2022, 20, .	0.6	4
3674	Identification of genetic mechanisms for tissue-specific genetic effects based on CRISPR screens. <i>Genetics</i> , 0, , .	1.2	1
3675	The performance of genome sequencing as a first-tier test for neurodevelopmental disorders. <i>European Journal of Human Genetics</i> , 2023, 31, 81-88.	1.4	28
3676	DPP9 deficiency: An inflammasomopathy that can be rescued by lowering NLRP1/IL-1 signaling. <i>Science Immunology</i> , 2022, 7, .	5.6	13
3678	Deep neural networks with controlled variable selection for the identification of putative causal genetic variants. <i>Nature Machine Intelligence</i> , 2022, 4, 761-771.	8.3	3
3679	Identifying the molecular drivers of ALS-implicated missense mutations. <i>Journal of Medical Genetics</i> , 2023, 60, 484-490.	1.5	5
3680	Lessons learned during the process of reporting individual genomic results to participants of a population-based biobank. <i>European Journal of Human Genetics</i> , 0, , .	1.4	0
3681	Clinical and genomic delineation of the new proximal 19p13.3 microduplication syndrome. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	1
3682	Assessing the Pathogenicity of In-Frame CACNA1F Indel Variants Using Structural Modeling. <i>Journal of Molecular Diagnostics</i> , 2022, 24, 1232-1239.	1.2	2
3683	Functional Assays Reclassify Suspected Splice-Altering Variants of Uncertain Significance in Mendelian Channelopathies. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, .	1.6	5
3684	Genetic Ancestry Correlates with Somatic Differences in a Real-World Clinical Cancer Sequencing Cohort. <i>Cancer Discovery</i> , 2022, 12, 2552-2565.	7.7	21
3685	Genetic and molecular architecture of familial hypercholesterolemia. <i>Journal of Internal Medicine</i> , 2023, 293, 144-165.	2.7	22
3686	Tissue- and cell-type-specific molecular and functional signatures of 16p11.2 reciprocal genomic disorder across mouse brain and human neuronal models. <i>American Journal of Human Genetics</i> , 2022, 109, 1789-1813.	2.6	13

#	ARTICLE	IF	CITATIONS
3687	Nuclear-embedded mitochondrial DNA sequences in 66,083 human genomes. <i>Nature</i> , 2022, 611, 105-114.	13.7	69
3689	Whole-genome sequencing of multiple related individuals with type 2 diabetes reveals an atypical likely pathogenic mutation in the PAX6 gene. <i>European Journal of Human Genetics</i> , 0, , .	1.4	1
3690	Real-world clinical and molecular management of 50 prospective patients with microphthalmia, anophthalmia and/or ocular coloboma. <i>British Journal of Ophthalmology</i> , 2023, 107, 1925-1935.	2.1	8
3691	Exome sequencing in a Swedish family with PMS2 mutation with varying penetrance of colorectal cancer: investigating the presence of genetic risk modifiers in colorectal cancer risk. <i>European Journal of Cancer Prevention</i> , 2023, 32, 113-118.	0.6	2
3692	Investigating the contributions of circadian pathway and insomnia risk genes to autism and sleep disturbances. <i>Translational Psychiatry</i> , 2022, 12, .	2.4	3
3693	Intestinal mucin is a chaperone of multivalent copper. <i>Cell</i> , 2022, 185, 4206-4215.e11.	13.5	13
3695	Loss-of-function mutations in <i>SCCE</i> found in Japanese patients with myoclonus-dystonia. <i>Clinical Genetics</i> , 2023, 103, 209-213.	1.0	0
3696	HTAADVar: Aggregation and fully automated clinical interpretation of genetic variants in heritable thoracic aortic aneurysm and dissection. <i>Genetics in Medicine</i> , 2022, 24, 2544-2554.	1.1	1
3697	Epithelioid and Spindle Cell Hemangioma. <i>American Journal of Surgical Pathology</i> , 2023, 47, 147-156.	2.1	4
3698	A patient with mosaic USP9X gene variant. <i>European Journal of Medical Genetics</i> , 2022, 65, 104638.	0.7	0
3699	Phenotypic spectrum in recessive STING-associated vasculopathy with onset in infancy: Four novel cases and analysis of previously reported cases. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	5
3700	The piRNA-pathway factor FKBP6 is essential for spermatogenesis but dispensable for control of meiotic LINE-1 expression in humans. <i>American Journal of Human Genetics</i> , 2022, 109, 1850-1866.	2.6	6
3701	Pleiotropic modifiers of age-related diabetes and neonatal intestinal obstruction in cystic fibrosis. <i>American Journal of Human Genetics</i> , 2022, 109, 1894-1908.	2.6	6
3702	Prevalence and mechanisms of somatic deletions in single human neurons during normal aging and in DNA repair disorders. <i>Nature Communications</i> , 2022, 13, .	5.8	10
3705	SVAT: Secure outsourcing of variant annotation and genotype aggregation. <i>BMC Bioinformatics</i> , 2022, 23, .	1.2	1
3706	Loss-of-function variants in <i>MYCBP2</i> cause neurobehavioural phenotypes and corpus callosum defects. <i>Brain</i> , 2023, 146, 1373-1387.	3.7	9
3707	Recent advances and challenges of rare variant association analysis in the biobank sequencing era. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	5
3708	A low-frequency <i>APOB</i> p.(Pro955Ser) variant contributes to the severity of/variability in familial hypercholesterolemia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 0, , .	1.8	1

#	ARTICLE	IF	CITATIONS
3709	ECFS standards of care on CFTR-related disorders: Diagnostic criteria of CFTR dysfunction. <i>Journal of Cystic Fibrosis</i> , 2022, 21, 922-936.	0.3	18
3710	Inherited bone marrow failure with macrothrombocytopenia due to germline tubulin beta class I () Tj ETQq1 1 0.784314 rgBT ₁ /Overlo 1.2	1.2	1
3711	FOSL2 truncating variants in the last exon cause a neurodevelopmental disorder with scalp and enamel defects. <i>Genetics in Medicine</i> , 2022, 24, 2475-2486.	1.1	2
3712	Gain-of-function mutations in KCNK3 cause a developmental disorder with sleep apnea. <i>Nature Genetics</i> , 2022, 54, 1534-1543.	9.4	10
3713	GABBR1 monoallelic de novo variants linked to neurodevelopmental delay and epilepsy. <i>American Journal of Human Genetics</i> , 2022, 109, 1885-1893.	2.6	6
3714	A pathogenic variant of TULP3 causes renal and hepatic fibrocystic disease. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	4
3715	Human papillomavirus-related neoplasia of the ocular adnexa. <i>Acta Ophthalmologica</i> , 2022, 100, 3-33.	0.6	4
3716	Destabilization of mutated human PUS3 protein causes intellectual disability. <i>Human Mutation</i> , 2022, 43, 2063-2078.	1.1	8
3717	Evaluating probabilistic genotyping for low-pass DNA sequencing. <i>Forensic Science International: Genetics Supplement Series</i> , 2022, 8, 112-114.	0.1	1
3719	De novo variants in FRMD5 are associated with developmental delay, intellectual disability, ataxia, and abnormalities of eye movement. <i>American Journal of Human Genetics</i> , 2022, 109, 1932-1943.	2.6	12
3720	Clinically relevant germline variants in allogeneic hematopoietic stem cell transplant recipients. <i>Bone Marrow Transplantation</i> , 2023, 58, 39-45.	1.3	2
3721	Biallelic variants in the <sc> <i>SLC13A1</i> </sc> sulfate transporter gene cause hyposulfatemia with a mild spondylo-epi-metaphyseal dysplasia. <i>Clinical Genetics</i> , 0, , .	1.0	4
3722	Gene panel to guide antiseizure medication prescribing: Does the cost justify the benefits?. <i>Epilepsia</i> , 0, , .	2.6	0
3723	Two novel CHD7 variants in patients with typical and mild features of CHARGE syndrome Co-occurring with esophageal atresia. <i>Journal of Pediatric Surgery Case Reports</i> , 2022, , 102478.	0.1	0
3724	The human disease gene LYSET is essential for lysosomal enzyme transport and viral infection. <i>Science</i> , 2022, 378, .	6.0	28
3725	Myhre syndrome is caused by dominant-negative dysregulation of SMAD4 and other co-factors. <i>Differentiation</i> , 2022, 128, 1-12.	1.0	2
3727	A retrospective single-centered, comprehensive targeted genetic sequencing analysis of prognostic survival using tissues from Korean patients with metastatic renal cell carcinoma after targeted therapy. <i>Investigative and Clinical Urology</i> , 2022, 63, 602.	1.0	0
3728	Harnessing AI and Genomics to Accelerate Drug Discovery. <i>Future of Business and Finance</i> , 2022, , 89-106.	0.3	1

#	ARTICLE	IF	CITATIONS
3729	Urinary Comprehensive Genomic Profiling Correlates Urothelial Carcinoma Mutations with Clinical Risk and Efficacy of Intervention. <i>Journal of Clinical Medicine</i> , 2022, 11, 5827.	1.0	6
3731	Recurrent <i>FOXP4</i> nonsense variant in two unrelated patients: Association with neurodevelopmental disease and congenital diaphragmatic hernia. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	0
3732	Both Heterozygous and Homozygous Loss of <i>JPH3</i> Variants Are Associated with a Paroxysmal Movement Disorder. <i>Movement Disorders</i> , 2023, 38, 155-157.	2.2	3
3733	A combined polygenic score of 21,293 rare and 22 common variants improves diabetes diagnosis based on hemoglobin A1C levels. <i>Nature Genetics</i> , 2022, 54, 1609-1614.	9.4	20
3734	Mendelian gene identification through mouse embryo viability screening. <i>Genome Medicine</i> , 2022, 14, .	3.6	2
3735	Cross-Ancestry Investigation of Venous Thromboembolism Genomic Predictors. <i>Circulation</i> , 2022, 146, 1225-1242.	1.6	25
3736	Distinctive Brain Malformations in Zhu-Tokita-Takenouchi-Kim Syndrome. <i>American Journal of Neuroradiology</i> , 0, , .	1.2	1
3737	Statistical and functional convergence of common and rare genetic influences on autism at chromosome 16p. <i>Nature Genetics</i> , 2022, 54, 1630-1639.	9.4	14
3738	Global Biobank Meta-analysis Initiative: Powering genetic discovery across human disease. <i>Cell Genomics</i> , 2022, 2, 100192.	3.0	85
3741	Modeling of ACTN4-Based Podocytopathy Using <i>Drosophila</i> Nephrocytes. <i>Kidney International Reports</i> , 2023, 8, 317-329.	0.4	6
3742	Heterozygous pathogenic variants involving <i>CBFB</i> cause a new skeletal disorder resembling cleidocranial dysplasia. <i>Journal of Medical Genetics</i> , 2023, 60, 498-504.	1.5	0
3743	Whole-genome sequencing for mitochondrial disorders identifies unexpected mimics. <i>Practical Neurology</i> , 0, , pn-2022-003570.	0.5	1
3744	<i>ERI1</i> : A case report of an autosomal recessive syndrome associated with developmental delay and distal limb abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 64-69.	0.7	2
3745	The Notch1/CD22 signaling axis disrupts Treg function in SARS-CoV-2-associated multisystem inflammatory syndrome in children. <i>Journal of Clinical Investigation</i> , 2023, 133, .	3.9	9
3746	The CancerMuts software package for the prioritization of missense cancer variants: a case study of <i>AMBRA1</i> in melanoma. <i>Cell Death and Disease</i> , 2022, 13, .	2.7	7
3747	Vidutolimod in Combination With Atezolizumab With and Without Radiation Therapy in Patients With Programmed Cell Death Protein 1 or Programmed Death-Ligand 1 Blockade-Resistant Advanced NSCLC. <i>JTO Clinical and Research Reports</i> , 2023, 4, 100423.	0.6	1
3749	Sex differences in interindividual gene expression variability across human tissues. , 2022, 1, .		5
3750	Informing variant assessment using structured evidence from prior classifications (PS1, PM5, and PVS1) Tj ETQq1 1,0.784314rgBT /Ove		

#	ARTICLE	IF	CITATIONS
3751	A Heterozygous Mutation in MFF Associated with a Mild Mitochondrial Phenotype. <i>Journal of Neuromuscular Diseases</i> , 2022, , 1-12.	1.1	0
3753	Delineation of functionally essential protein regions for 242 neurodevelopmental genes. <i>Brain</i> , 2023, 146, 519-533.	3.7	6
3754	Congenital Hypermetabolism and Uncoupled Oxidative Phosphorylation. <i>New England Journal of Medicine</i> , 2022, 387, 1395-1403.	13.9	10
3755	Reclassification of a likely pathogenic Dutch founder variant in KCNH2; implications of reduced penetrance. <i>Human Molecular Genetics</i> , 0, , .	1.4	1
3756	The genomic landscape across 474 surgically accessible epileptogenic human brain lesions. <i>Brain</i> , 2023, 146, 1342-1356.	3.7	21
3758	Genetic variants associated with psychiatric disorders are enriched at epigenetically active sites in lymphoid cells. <i>Nature Communications</i> , 2022, 13, .	5.8	13
3759	The <i>SYNGAP1</i> 3'UTR Variant in ALS Patients Causes Aberrant <i>SYNGAP1</i> Splicing and Dendritic Spine Loss by Recruiting HNRNPK. <i>Journal of Neuroscience</i> , 2022, 42, 8881-8896.	1.7	2
3760	Direct detection of natural selection in Bronze Age Britain. <i>Genome Research</i> , 2022, 32, 2057-2067.	2.4	20
3761	Genetic variations affecting ACE2 protein stability in minority populations. <i>Frontiers in Medicine</i> , 0, 9, .	1.2	1
3762	The fly homolog of <i>SUPT16H</i> , a gene associated with neurodevelopmental disorders, is required in a cell-autonomous fashion for cell survival. <i>Human Molecular Genetics</i> , 2023, 32, 984-997.	1.4	6
3763	Ancestry-specific high-risk gene variant profiling unmask diabetes-associated genes. <i>Human Molecular Genetics</i> , 2024, 33, 655-666.	1.4	0
3764	The Finnish genetic heritage in 2022 – from diagnosis to translational research. <i>DMM Disease Models and Mechanisms</i> , 2022, 15, .	1.2	7
3765	A missense, loss-of-function YARS1 variant in a patient with proximal-predominant motor neuropathy. <i>Journal of Physical Education and Sports Management</i> , 0, , mcs.a006246.	0.5	1
3766	Neurodevelopmental and Epilepsy Phenotypes in Individuals With Missense Variants in the Voltage-Sensing and Pore Domains of <i>KCNH5</i> . <i>Neurology</i> , 2023, 100, .	1.5	4
3767	Brain monoamine vesicular transport disease caused by homozygous SLC18A2 variants: A study in 42 affected individuals. <i>Genetics in Medicine</i> , 2023, 25, 90-102.	1.1	15
3768	The new uORFdb: integrating literature, sequence, and variation data in a central hub for uORF research. <i>Nucleic Acids Research</i> , 2023, 51, D328-D336.	6.5	7
3769	Clinical Features, Neuropathology, and Surgical Outcome in Patients With Refractory Epilepsy and Brain Somatic Variants in the <i>SLC35A2</i> Gene. <i>Neurology</i> , 2023, 100, .	1.5	14
3770	Transient regulation of focal adhesion via Tensin3 is required for nascent oligodendrocyte differentiation. <i>ELife</i> , 0, 11, .	2.8	5

#	ARTICLE	IF	CITATIONS
3771	Defective binding of ETS1 and STAT4 due to a mutation in the promoter region of THPO as a novel mechanism of congenital amegakaryocytic thrombocytopenia. <i>Haematologica</i> , 0, , .	1.7	3
3772	A Common Variant in the CDK8 Gene Is Associated with Sporadic Pituitary Adenomas in the Portuguese Population: A Case-Control Study. <i>International Journal of Molecular Sciences</i> , 2022, 23, 11749.	1.8	0
3773	Genetic Characterization in Familial Rotator Cuff Tear: An Exome Sequencing Study. <i>Biology</i> , 2022, 11, 1565.	1.3	1
3774	MetaRNN: differentiating rare pathogenic and rare benign missense SNVs and InDels using deep learning. <i>Genome Medicine</i> , 2022, 14, .	3.6	28
3775	De novo missense variants in the E3 ubiquitin ligase adaptor KLHL20 cause a developmental disorder with intellectual disability, epilepsy, and autism spectrum disorder. <i>Genetics in Medicine</i> , 2022, 24, 2464-2474.	1.1	1
3776	Trio-based whole exome sequencing in patients with suspected sporadic inborn errors of immunity: A retrospective cohort study. <i>ELife</i> , 0, 11, .	2.8	3
3777	Molecular characterization of renal cell carcinoma tumors from a phase III anti-angiogenic adjuvant therapy trial. <i>Nature Communications</i> , 2022, 13, .	5.8	4
3778	A Case of Bilateral Microphthalmia and Extensive Colobomas of the Globes Associated with a Likely Pathogenic Homozygous <i>SIX6</i> Variant. <i>Case Reports in Ophthalmology</i> , 0, , 804-808.	0.3	0
3779	A COL4A4-G394S Variant and Impaired Collagen IV Trimerization in a Patient with Mild Alport Syndrome. <i>Kidney360</i> , 2022, 3, 1899-1908.	0.9	1
3780	Genetic characterization of 1,210 Japanese pedigrees with inherited retinal diseases by whole-exome sequencing. <i>Human Mutation</i> , 0, , .	1.1	9
3781	Multiple endocrine neoplasia type 2 (MEN2) and <i>RET</i> specific modifications of the ACMG/AMP variant classification guidelines and impact on the MEN2 <i>RET</i> database. <i>Human Mutation</i> , 2022, 43, 1780-1794.	1.1	6
3782	A congenital <i>CSF3R</i> mutation in chronic neutropenia reveals a vital role for a cytokine receptor extracellular hinge motif in the response to granulocyte colony-stimulating factor. <i>Pediatric Blood and Cancer</i> , 2023, 70, .	0.8	0
3783	Maturity-onset diabetes of the young in a large Portuguese cohort. <i>Acta Diabetologica</i> , 2023, 60, 83-91.	1.2	1
3784	Whole genome sequence analysis of blood lipid levels in >66,000 individuals. <i>Nature Communications</i> , 2022, 13, .	5.8	26
3785	Risk Variants in the Exomes of Children With Critical Illness. <i>JAMA Network Open</i> , 2022, 5, e2239122.	2.8	1
3786	CVID-Associated B Cell Activating Factor Receptor Variants Change Receptor Oligomerization, Ligand Binding, and Signaling Responses. <i>Journal of Clinical Immunology</i> , 2023, 43, 391-405.	2.0	3
3787	Genetic map of regional sulcal morphology in the human brain from UK biobank data. <i>Nature Communications</i> , 2022, 13, .	5.8	9
3788	Investigating the role of common and rare variants in multiplex multiple sclerosis families reveals an increased burden of common risk variation. <i>Scientific Reports</i> , 2022, 12, .	1.6	1

#	ARTICLE	IF	CITATIONS
3790	RaScALL: Rapid (Ra) screening (Sc) of RNA-seq data for prognostically significant genomic alterations in acute lymphoblastic leukaemia (ALL). <i>PLoS Genetics</i> , 2022, 18, e1010300.	1.5	13
3791	Candidate Modifier Genes for the Penetrance of Leber's Hereditary Optic Neuropathy. <i>International Journal of Molecular Sciences</i> , 2022, 23, 11891.	1.8	3
3793	Identification of Clinical Variants beyond the Exome in Inborn Errors of Metabolism. <i>International Journal of Molecular Sciences</i> , 2022, 23, 12850.	1.8	3
3794	Cytochrome b5 reductases: Redox regulators of cell homeostasis. <i>Journal of Biological Chemistry</i> , 2022, 298, 102654.	1.6	13
3795	Genetic and Clinical Spectrum of GNE Myopathy in Russia. <i>Genes</i> , 2022, 13, 1991.	1.0	3
3796	Gene Sequencing Identifies Perturbation in Nitric Oxide Signaling as a Nonlipid Molecular Subtype of Coronary Artery Disease. <i>Circulation Genomic and Precision Medicine</i> , 2022, 15, .	1.6	4
3797	Truncating Variants in <i>RFC1</i> in Cerebellar Ataxia, Neuropathy, and Vestibular Areflexia Syndrome. <i>Neurology</i> , 2023, 100, .	1.5	16
3798	Using human genetics to improve safety assessment of therapeutics. <i>Nature Reviews Drug Discovery</i> , 2023, 22, 145-162.	21.5	20
3799	Common genetic risk factors in ASD and ADHD co-occurring families. <i>Human Genetics</i> , 2023, 142, 217-230.	1.8	6
3800	Computed cancer interactome explains the effects of somatic mutations in cancers. <i>Protein Science</i> , 2022, 31, .	3.1	8
3802	Transcriptome-wide and stratified genomic structural equation modeling identify neurobiological pathways shared across diverse cognitive traits. <i>Nature Communications</i> , 2022, 13, .	5.8	11
3803	Genetic Analysis of RASD1 as a Candidate Gene for Schizophrenia. <i>Balkan Medical Journal</i> , 2022, 39, 422-428.	0.3	1
3805	Evolution of an Iron-Detoxifying Protein: Eukaryotic and Rickettsia Frataxins Contain a Conserved Site Which Is Not Present in Their Bacterial Homologues. <i>International Journal of Molecular Sciences</i> , 2022, 23, 13151.	1.8	2
3806	Intragenic compensation through the lens of deep mutational scanning. <i>Biophysical Reviews</i> , 2022, 14, 1161-1182.	1.5	4
3807	Damaging variants in FOXI3 cause microtia and craniofacial microsomia. <i>Genetics in Medicine</i> , 2023, 25, 143-150.	1.1	8
3808	Genome-wide rare variant score associates with morphological subtypes of autism spectrum disorder. <i>Nature Communications</i> , 2022, 13, .	5.8	7
3809	Visual inspection reveals a novel pathogenic mutation in <i>PKD1</i> missed by the variant caller in whole-exome sequencing. <i>Molecular Medicine Reports</i> , 2022, 26, .	1.1	0
3810	Reproductive Phenotypes and Genotypes in Men With IHH. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2023, 108, 897-908.	1.8	3

#	ARTICLE	IF	CITATIONS
3811	Influences of rare copy-number variation on human complex traits. <i>Cell</i> , 2022, 185, 4233-4248.e27.	13.5	15
3812	Genome-wide detection of human variants that disrupt intronic branchpoints. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	21
3813	MARGINAL: An Automatic Classification of Variants in BRCA1 and BRCA2 Genes Using a Machine Learning Model. <i>Biomolecules</i> , 2022, 12, 1552.	1.8	3
3814	Echtvar: compressed variant representation for rapid annotation and filtering of SNPs and indels. <i>Nucleic Acids Research</i> , 0, , .	6.5	0
3815	Exome sequencing identified five novel <i>USH2A</i> variants in Korean patients with retinitis pigmentosa. <i>Ophthalmic Genetics</i> , 2023, 44, 163-170.	0.5	0
3816	Taiwan Biobank: A rich biomedical research database of the Taiwanese population. <i>Cell Genomics</i> , 2022, 2, 100197.	3.0	21
3817	Monogenic early-onset lymphoproliferation and autoimmunity: Natural history of STAT3 gain-of-function syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2023, 151, 1081-1095.	1.5	31
3819	Roles of DNA damage repair and precise targeted therapy in renal cancer (Review). <i>Oncology Reports</i> , 2022, 48, .	1.2	3
3822	Phenotypic variability in <i>LAMA3</i> -associated amelogenesis imperfecta. <i>Oral Diseases</i> , 2023, 29, 3514-3524.	1.5	4
3823	A database of 5305 healthy Korean individuals reveals genetic and clinical implications for an East Asian population. <i>Experimental and Molecular Medicine</i> , 2022, 54, 1862-1871.	3.2	13
3824	Genetic determinants for the racial disparities in the risk of prostate and testicular cancers. <i>Communications Medicine</i> , 2022, 2, .	1.9	0
3825	Common and rare variants of EGF increase the genetic risk of Alzheimer's disease as revealed by targeted sequencing of growth factors in Han Chinese. <i>Neurobiology of Aging</i> , 2022, , .	1.5	1
3826	Ophthalmic genetic counselling: emerging trends in practice perspectives in Asia. <i>Journal of Community Genetics</i> , 0, , .	0.5	0
3827	A founder event causing a dominant childhood epilepsy survives 800 years through weak selective pressure. <i>American Journal of Human Genetics</i> , 2022, 109, 2080-2087.	2.6	3
3828	Phenome-wide analysis of Taiwan Biobank reveals novel glycemia-related loci and genetic risks for diabetes. <i>Communications Biology</i> , 2022, 5, .	2.0	9
3829	Exome-wide association study to identify rare variants influencing COVID-19 outcomes: Results from the Host Genetics Initiative. <i>PLoS Genetics</i> , 2022, 18, e1010367.	1.5	21
3830	Whole-Genome and Long-Read Sequencing Identify a Novel Mechanism in <i>RFC1</i> Resulting in CANVAS Syndrome. <i>Neurology: Genetics</i> , 2022, 8, .	0.9	5
3831	Ceramide Analysis in Combination With Genetic Testing May Provide a Precise Diagnosis for Self-Healing Collodion Babies. <i>Journal of Lipid Research</i> , 2022, 63, 100308.	2.0	2

#	ARTICLE	IF	CITATIONS
3832	Delineation of a KDM2B-related neurodevelopmental disorder and its associated DNA methylation signature. <i>Genetics in Medicine</i> , 2023, 25, 49-62.	1.1	9
3833	Computational modeling of the effect of five mutations on the structure of the ACE2 receptor and their correlation with infectivity and virulence of some emerged variants of SARS-CoV-2 suggests mechanisms of binding affinity dysregulation. <i>Chemico-Biological Interactions</i> , 2022, 368, 110244.	1.7	4
3834	Clinical Efficacy and Whole-Exome Sequencing of Liquid Biopsies in a Phase IB/II Study of Bazedoxifene and Palbociclib in Advanced Hormone Receptor-Positive Breast Cancer. <i>Clinical Cancer Research</i> , 2022, 28, 5066-5078.	3.2	5
3836	Reversible hypogonadotropic hypogonadism in men with the fertile eunuch/Pasqualini syndrome: A single-center natural history study. <i>Frontiers in Endocrinology</i> , 0, 13, .	1.5	2
3838	Reduced penetrance of MODY-associated HNF1A/HNF4A variants but not GCK variants in clinically unselected cohorts. <i>American Journal of Human Genetics</i> , 2022, 109, 2018-2028.	2.6	31
3839	Multi-omics approach dissects cis-regulatory mechanisms underlying North Carolina macular dystrophy, a retinal enhanceropathy. <i>American Journal of Human Genetics</i> , 2022, 109, 2029-2048.	2.6	14
3840	Transcriptional and functional consequences of alterations to MEF2C and its topological organization in neuronal models. <i>American Journal of Human Genetics</i> , 2022, 109, 2049-2067.	2.6	8
3841	The inner junction protein CFAP20 functions in motile and non-motile cilia and is critical for vision. <i>Nature Communications</i> , 2022, 13, .	5.8	7
3842	A clustering of heterozygous missense variants in the crucial chromatin modifier WDR5 defines a new neurodevelopmental disorder. <i>Human Genetics and Genomics Advances</i> , 2023, 4, 100157.	1.0	2
3843	The pathogenic c.1171A>G (p.Arg391Gly) and c.2359C>A (p.Val787Ile) ABCC6 variants display incomplete penetrance causing pseudoxanthoma elasticum in a subset of individuals. <i>Human Mutation</i> , 0, , .	1.1	1
3845	Systematic analysis and prediction of genes associated with monogenic disorders on human chromosome X. <i>Nature Communications</i> , 2022, 13, .	5.8	14
3846	A Human Hereditary Cardiomyopathy Shares a Genetic Substrate With Bicuspid Aortic Valve. <i>Circulation</i> , 2023, 147, 47-65.	1.6	9
3847	Ensembl 2023. <i>Nucleic Acids Research</i> , 2023, 51, D933-D941.	6.5	153
3848	Characterization of Arabian Peninsula whole exomes: Contributing to the catalogue of human diversity. <i>IScience</i> , 2022, 25, 105336.	1.9	0
3849	Identification of Genetic Risk Factors for Monogenic and Complex Canine Diseases. <i>Annual Review of Animal Biosciences</i> , 2023, 11, 183-205.	3.6	3
3850	Genome-wide association studies of COVID-19: Connecting the dots. <i>Infection, Genetics and Evolution</i> , 2022, 106, 105379.	1.0	13
3851	Mapping the genetic features of T-ALL cases through simplified NGS approach. <i>Clinical Immunology</i> , 2022, 245, 109151.	1.4	1
3852	From Calcium Channels to New Therapeutics. , 2022, , 687-706.		0

#	ARTICLE	IF	CITATIONS
3853	Diagnostic reasoning in neurogenetics: a general approach. <i>Arquivos De Neuro-Psiquiatria</i> , 2022, 80, 944-952.	0.3	0
3854	Diagnosing, discarding, or de-VUSsing: A practical guide to (un)targeted metabolomics as a variant-transcending functional tests. <i>Genetics in Medicine</i> , 2023, 25, 125-134.	1.1	4
3855	Neuromuscular symptoms in patients with <i>RYR1</i> -related malignant hyperthermia and rhabdomyolysis. <i>Brain Communications</i> , 2022, 4, .	1.5	7
3856	Predicting functional effect of missense variants using graph attention neural networks. <i>Nature Machine Intelligence</i> , 2022, 4, 1017-1028.	8.3	15
3858	Inverted genomic regions between reference genome builds in humans impact imputation accuracy and decrease the power of association testing. <i>Human Genetics and Genomics Advances</i> , 2023, 4, 100159.	1.0	2
3859	Somatic mosaicism in STAG2-associated cohesinopathies: Expansion of the genotypic and phenotypic spectrum. <i>Frontiers in Cell and Developmental Biology</i> , 0, 10, .	1.8	2
3860	Combining genetic constraint with predictions of alternative splicing to prioritize deleterious splicing in rare disease studies. <i>BMC Bioinformatics</i> , 2022, 23, .	1.2	7
3861	The contribution of common and rare genetic variants to variation in metabolic traits in 288,137 East Asians. <i>Nature Communications</i> , 2022, 13, .	5.8	13
3862	Non-coding variants disrupting a tissue-specific regulatory element in HK1 cause congenital hyperinsulinism. <i>Nature Genetics</i> , 2022, 54, 1615-1620.	9.4	12
3863	SCIP: software for efficient clinical interpretation of copy number variants detected by whole-genome sequencing. <i>Human Genetics</i> , 2023, 142, 201-216.	1.8	1
3864	Assessment of an automated approach for variant interpretation in screening for monogenic disorders: A single-center study. <i>Molecular Genetics & Genomic Medicine</i> , 0, , .	0.6	1
3866	14-fold increased prevalence of rare glucokinase gene variant carriers in unselected Danish patients with newly diagnosed type 2 diabetes. <i>Diabetes Research and Clinical Practice</i> , 2022, , 110159.	1.1	4
3868	Identification of a novel large multigene deletion and a frameshift indel in <i>PDE6B</i> as the underlying cause of early onset recessive rod-cone degeneration. <i>Journal of Physical Education and Sports Management</i> , 0, , mcs.a006247.	0.5	0
3869	A haplotype-resolved genome assembly of the Nile rat facilitates exploration of the genetic basis of diabetes. <i>BMC Biology</i> , 2022, 20, .	1.7	8
3870	Case report: Genomic screening for inherited cardiac conditions in Ecuadorian mestizo relatives: Improving familial diagnose. <i>Frontiers in Cardiovascular Medicine</i> , 0, 9, .	1.1	2
3871	Two new Scianna variants causing loss of high prevalence antigens: <i>ERMAP</i> model and 3D analysis of the antigens. <i>Transfusion</i> , 2023, 63, 230-238.	0.8	2
3872	Multivariate analysis of a missense variant in <i>CREBRF</i> reveals associations with measures of adiposity in people of Polynesian ancestries. <i>Genetic Epidemiology</i> , 0, , .	0.6	0
3873	Combined germline pathogenic variants in <i>FLCN</i> and <i>TP53</i> are associated with early onset renal cell carcinoma and brain tumors. <i>Molecular Genetics & Genomic Medicine</i> , 0, , .	0.6	2

#	ARTICLE	IF	CITATIONS
3874	Genome-wide identification of exon extension/shrinkage events induced by splice-site-creating mutations. <i>RNA Biology</i> , 2022, 19, 1143-1152.	1.5	0
3876	Syntaxin 4 is essential for hearing in human and zebrafish. <i>Human Molecular Genetics</i> , 2023, 32, 1184-1192.	1.4	1
3878	FAVOR: functional annotation of variants online resource and annotator for variation across the human genome. <i>Nucleic Acids Research</i> , 2023, 51, D1300-D1311.	6.5	39
3879	Copy Number Variation and Structural Genomic Findings in 116 Cases of Sudden Unexplained Death between 1 and 28 Months of Age. <i>Genetics & Genomics Next</i> , 2023, 4, .	0.8	1
3880	The complex, dynamic SpliceOme of the small GTPase transcripts altered by technique, sex, genetics, tissue specificity, and RNA base editing. <i>Frontiers in Cell and Developmental Biology</i> , 0, 10, .	1.8	2
3882	Role of genetic testing in young patients with idiopathic atrioventricular conduction disease. <i>Europace</i> , 2023, 25, 643-650.	0.7	4
3883	Damaging missense variants in IGF1R implicate a role for IGF-1 resistance in the etiology of type 2 diabetes. <i>Cell Genomics</i> , 2022, 2, 100208.	3.0	12
3884	Deciphering the impact of genetic variation on human polyadenylation using APARENT2. <i>Genome Biology</i> , 2022, 23, .	3.8	12
3886	Analysis of 1276 Haplotype-Resolved Genomes Allows Characterization of Cis- and Trans-Abundant Genes. <i>Methods in Molecular Biology</i> , 2023, , 237-272.	0.4	0
3887	Genetics of Kidney Disease: The Unexpected Role of Rare Disorders. <i>Annual Review of Medicine</i> , 2023, 74, 353-367.	5.0	3
3888	Identification of nine novel variants across <i>PAX3</i> , <i>SOX10</i> , <i>EDNRB</i> , and <i>MITF</i> genes in Waardenburg syndrome with next-generation sequencing. <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, .	0.6	5
3889	Variant Enrichment Analysis to Explore Pathways Disruption in a Necropsy Series of Asbestos-Exposed Shipyard Workers. <i>International Journal of Molecular Sciences</i> , 2022, 23, 13628.	1.8	1
3890	Ehlers-Danlos: A Literature Review and Case Report in a Colombian Woman with Multiple Comorbidities. <i>Genes</i> , 2022, 13, 2118.	1.0	2
3891	Targeted S5 RNA sequencing assay for the identification and direct association of common body fluids with DNA donors in mixtures. <i>International Journal of Legal Medicine</i> , 2023, 137, 13-32.	1.2	3
3892	A novel splice-affecting HNF1A variant with large population impact on diabetes in Greenland. <i>Lancet Regional Health - Europe</i> , The, 2023, 24, 100529.	3.0	3
3893	Meta-analysis fine-mapping is often miscalibrated at single-variant resolution. <i>Cell Genomics</i> , 2022, 2, 100210.	3.0	30
3894	Translational opportunities emerge from genetic influences on health. <i>Trends in Molecular Medicine</i> , 2022, 28, 1028-1029.	3.5	0
3896	A Heterozygous Gain-of-Function Variant in IKBKB Associated with Autoimmunity and Autoinflammation. <i>Journal of Clinical Immunology</i> , 2023, 43, 512-520.	2.0	2

#	ARTICLE	IF	CITATIONS
3897	Profiling human pathogenic repeat expansion regions by synergistic and multi-level impacts on molecular connections. <i>Human Genetics</i> , 0, , .	1.8	0
3898	Genome-Wide Sequencing Identified Rare Genetic Variants for Childhood-Onset Monogenic Lupus. <i>Journal of Rheumatology</i> , 2023, 50, 671-675.	1.0	3
3899	Maternal heterozygosity of <i>Slc6a19</i> causes metabolic perturbation and congenital NAD deficiency disorder in mice. <i>DMM Disease Models and Mechanisms</i> , 2023, 16, .	1.2	3
3901	A Clinician's Guide to Bioinformatics for Next-Generation Sequencing. <i>Journal of Thoracic Oncology</i> , 2023, 18, 143-157.	0.5	9
3903	Inherited Cancer Susceptibility Gene Sequence Variations Among Patients With Appendix Cancer. <i>JAMA Oncology</i> , 2023, 9, 95.	3.4	8
3904	The prevalence of germline pathogenic variants in Estonian colorectal cancer patients: results from routine clinical setting 2016–2021. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1
3905	Further Evidence That <i>ARIH1</i> Rare Variants Predispose to Thoracic Aortic Disease. <i>Circulation Genomic and Precision Medicine</i> , 0, , .	1.6	0
3906	RBPM2 Is a Myocardial-Enriched Splicing Regulator Required for Cardiac Function. <i>Circulation Research</i> , 2022, 131, 980-1000.	2.0	9
3907	Overall survival with circulating tumor DNA-guided therapy in advanced non-small-cell lung cancer. <i>Nature Medicine</i> , 2022, 28, 2353-2363.	15.2	41
3908	Whole exome sequencing of FFPE samples—expanding the horizon of forensic molecular autopsies. <i>International Journal of Legal Medicine</i> , 0, , .	1.2	0
3910	Ethnic-specificity, evolution origin and deleteriousness of Asian <i>BRCA</i> variation revealed by over 7500 <i>BRCA</i> variants derived from Asian population. <i>International Journal of Cancer</i> , 2023, 152, 1159-1173.	2.3	6
3911	Microdeletions at 19p13.11p12 in five individuals with neurodevelopmental delay. <i>European Journal of Medical Genetics</i> , 2023, 66, 104669.	0.7	0
3912	Human genetics uncovers <i>MAP3K15</i> as an obesity-independent therapeutic target for diabetes. <i>Science Advances</i> , 2022, 8, .	4.7	11
3913	Acral lamellar ichthyosis with amino acid substitution in the C-terminus of keratin 2. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2023, 37, 817-822.	1.3	0
3915	A novel splicing mutation in 5'UTR of <i>GJB1</i> causes X-linked Charcot-Marie tooth disease. <i>Molecular Genetics & Genomic Medicine</i> , 2023, 11, .	0.6	2
3916	Genetics of BAG3: A Paradigm for Developing Precision Therapies for Dilated Cardiomyopathies. <i>Journal of the American Heart Association</i> , 2022, 11, .	1.6	8
3917	Identification of PCSK9-like human gene knockouts using metabolomics, proteomics, and whole-genome sequencing in a consanguineous population. <i>Cell Genomics</i> , 2023, 3, 100218.	3.0	4
3918	Carrier frequency and incidence estimation of RPE65-associated inherited retinal diseases in East Asian population by population database-based analysis. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, .	1.2	1

#	ARTICLE	IF	CITATIONS
3919	Single-cell genome-wide association reveals that a nonsynonymous variant in ERAP1 confers increased susceptibility to influenza virus. <i>Cell Genomics</i> , 2022, 2, 100207.	3.0	2
3920	Rare tandem repeat expansions associate with genes involved in synaptic and neuronal signaling functions in schizophrenia. <i>Molecular Psychiatry</i> , 2023, 28, 475-482.	4.1	10
3921	Deep learning-assisted genome-wide characterization of massively parallel reporter assays. <i>Nucleic Acids Research</i> , 0, , .	6.5	1
3922	Persistent Müllerian duct syndrome associated with genetic defects in the regulatory subunit of myosin phosphatase. <i>Human Reproduction</i> , 2022, 37, 2952-2959.	0.4	2
3923	Functional genomics of OCTN2 variants informs protein-specific variant effect predictor for Carnitine Transporter Deficiency. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	6
3924	Annotation of structural variants with reported allele frequencies and related metrics from multiple datasets using SVAFotate. <i>BMC Bioinformatics</i> , 2022, 23, .	1.2	2
3925	Structural basis of organic cation transporter-3 inhibition. <i>Nature Communications</i> , 2022, 13, .	5.8	26
3927	Characterization of HCl-EC-23 a novel estrogen- and progesterone-responsive endometrial cancer cell line. <i>Scientific Reports</i> , 2022, 12, .	1.6	0
3928	Genetic Ancestry Inference from Cancer-Derived Molecular Data across Genomic and Transcriptomic Platforms. <i>Cancer Research</i> , 2023, 83, 49-58.	0.4	5
3930	Carrier frequency and incidence estimation of familial hemophagocytic lymphohistiocytosis in East Asian populations by genome aggregation database (gnomAD) based analysis. <i>Frontiers in Pediatrics</i> , 0, 10, .	0.9	1
3931	Diagnostic yield of a multigene sequencing approach in children classified as idiopathic short stature. <i>Endocrine Connections</i> , 2022, 11, .	0.8	4
3932	Distinctive Nested Glomoid Neoplasm. <i>American Journal of Surgical Pathology</i> , 2023, 47, 12-24.	2.1	16
3933	Novel genotyping algorithms for rare variants significantly improve the accuracy of Applied Biosystems [®] , [®] Axiom [®] , [®] array genotyping calls: Retrospective evaluation of UK Biobank array data. <i>PLoS ONE</i> , 2022, 17, e0277680.	1.1	2
3934	The next-generation Open Targets Platform: reimagined, redesigned, rebuilt. <i>Nucleic Acids Research</i> , 2023, 51, D1353-D1359.	6.5	88
3936	Utility of genetic testing in pediatric epilepsy: Experience from a low to middle-income country. <i>Epilepsy and Behavior Reports</i> , 2022, 20, 100575.	0.5	1
3938	Neopeptides prediction strategies: an integration of cancer genomics and immunoinformatics approaches. <i>Briefings in Functional Genomics</i> , 0, , .	1.3	1
3939	Transposable element-mediated rearrangements are prevalent in human genomes. <i>Nature Communications</i> , 2022, 13, .	5.8	17
3942	The current understanding of germline predisposition in non-syndromic sagittal craniosynostosis: a systematic review. <i>Child's Nervous System</i> , 0, , .	0.6	1

#	ARTICLE	IF	CITATIONS
3943	Osteoporosis related to WNT1 variants: a not infrequent cause of osteoporosis. Osteoporosis International, 2023, 34, 405-411.	1.3	2
3946	<i>GTF3A</i> mutations predispose to herpes simplex encephalitis by disrupting biogenesis of the host-derived RIG-I ligand <i>RNA5SP141</i>. Science Immunology, 2022, 7, .	5.6	4
3947	A 9.8Âmb deletion at 7q31.2q31.31 downstream of <i>FOXP2</i> in an individual with speech and language impairment suggests a possible positional effect. Clinical Case Reports (discontinued), 2022, 10, .	0.2	0
3948	Pharmacogenomics of GLP-1 receptor agonists: a genome-wide analysis of observational data and large randomised controlled trials. Lancet Diabetes and Endocrinology,the, 2023, 11, 33-41.	5.5	29
3949	New approaches to genetic counseling. , 2024, , 173-195.		0
3950	Novel variants in GABAA receptor subunits: A possible association with benzodiazepine resistance in patients with drug-resistant epilepsy. Epilepsy Research, 2023, 189, 107056.	0.8	2
3951	Toward reporting standards for the pathogenicity of variant combinations involved in multilocus/oligogenic diseases. Human Genetics and Genomics Advances, 2023, 4, 100165.	1.0	3
3952	Mapping the Constrained Coding Regions in the Human Genome to Their Corresponding Proteins. Journal of Molecular Biology, 2023, 435, 167892.	2.0	0
3953	Inherited rare variants in homologous recombination and neurodevelopmental genes are associated with increased risk of neuroblastoma. EBioMedicine, 2023, 87, 104395.	2.7	4
3954	A recessive form of craniodiaphyseal dysplasia caused by a homozygous missense variant in SP7/Osterix. Bone, 2023, 167, 116633.	1.4	1
3955	The Human Genome. , 2022, , .		0
3956	Ashkenazi Jewish and Other White APC I1307K Carriers Are at Higher Risk for Multiple Cancers. Cancers, 2022, 14, 5875.	1.7	2
3959	Targeted Resequencing of Otosclerosis Patients from Different Populations Replicates Results from a Previous Genome-Wide Association Study. Journal of Clinical Medicine, 2022, 11, 6978.	1.0	0
3960	Single amino acid variation in MAB21L1 is dominantly associated with congenital eye defects. Journal of Medical Genetics, 0, , jmedgenet-2022-108506.	1.5	1
3961	Whole-exome sequencing study identifies rare variants and genes associated with intraocular pressure and glaucoma. Nature Communications, 2022, 13, .	5.8	6
3963	Multi-objective prioritization of genes for high-throughput functional assays towards improved clinical variant classification. , 2022, , .		0
3964	Systematic assays and resources for the functional annotation of non-coding variants. Medizinische Genetik, 2022, 34, 275-286.	0.1	1
3965	Hyperammonemia in Russia Due to Carbonic Anhydrase VA Deficiency Caused by Homozygous Mutation p.Lys185Lys (c.555G>A) of the CA5A Gene. International Journal of Molecular Sciences, 2022, 23, 15026.	1.8	1

#	ARTICLE	IF	CITATIONS
3966	<i>PRDM10</i> directs <i>FLCN</i> expression in a novel disorder overlapping with Birt-Hogg-Dubé syndrome and familial lipomatosis. <i>Human Molecular Genetics</i> , 2023, 32, 1223-1235.	1.4	5
3967	Identification of prolactin receptor variants with diverse effects on receptor signalling. <i>Journal of Molecular Endocrinology</i> , 2023, 70, .	1.1	2
3968	Mutational analysis of ribosomal proteins in a cohort of pediatric patients with T-cell acute lymphoblastic leukemia reveals Q123R, a novel mutation in <i>RPL10</i> . <i>Frontiers in Genetics</i> , 0, 13, .	1.1	3
3969	Genome-Wide DNA Methylation Profiling Solves Uncertainty in Classifying <i>NSD1</i> Variants. <i>Genes</i> , 2022, 13, 2163.	1.0	4
3970	Do Semaphorins Play a Role in Development of Fibrosis in Patients with Nonalcoholic Fatty Liver Disease?. <i>Biomedicines</i> , 2022, 10, 3014.	1.4	1
3971	An Approach to Identifying and Quantifying Bias in Biomedical Data. , 2022, , .		0
3972	Millennium-old pathogenic Mendelian mutation discovery for multiple osteochondromas from a Gaelic Medieval graveyard. <i>European Journal of Human Genetics</i> , 2023, 31, 248-251.	1.4	2
3974	Predicting and Understanding the Pathology of Single Nucleotide Variants in Human <i>COQ</i> Genes. <i>Antioxidants</i> , 2022, 11, 2308.	2.2	3
3975	Genomic Scar Score: A robust model predicting homologous recombination deficiency based on genomic instability. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , 2022, 129, 14-22.	1.1	5
3976	Genomic signature of Fanconi anaemia DNA repair pathway deficiency in cancer. <i>Nature</i> , 2022, 612, 495-502.	13.7	28
3977	Exome sequencing identifies rare damaging variants in <i>ATP8B4</i> and <i>ABCA1</i> as risk factors for Alzheimer's disease. <i>Nature Genetics</i> , 2022, 54, 1786-1794.	9.4	51
3978	Insights on variant analysis in silico tools for pathogenicity prediction. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	17
3981	Analyzing the Korean reference genome with meta-imputation increased the imputation accuracy and spectrum of rare variants in the Korean population. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	0
3982	Arrhythmogenic cardiomyopathy is under-recognized in end-stage pediatric heart failure: A 36-year single-center experience. <i>Pediatric Transplantation</i> , 2023, 27, .	0.5	2
3983	A novel combination of OHVIRA syndrome and likely causal variant in <i>UMOD</i> gene. <i>CEN Case Reports</i> , 2023, 12, 249-253.	0.5	1
3984	Retinopathy and optic atrophy in a case of <i>COQ2</i> -related primary coenzyme Q ₁₀ deficiency. <i>Ophthalmic Genetics</i> , 2023, 44, 486-490.	0.5	2
3986	GhostKnockoff inference empowers identification of putative causal variants in genome-wide association studies. <i>Nature Communications</i> , 2022, 13, .	5.8	1
3990	FarGen: Elucidating the distribution of coding variants in the isolated population of the Faroe Islands. <i>European Journal of Human Genetics</i> , 2023, 31, 329-337.	1.4	3

#	ARTICLE	IF	CITATIONS
3991	Angiopoietin-like 2 is essential to aortic valve development in mice. <i>Communications Biology</i> , 2022, 5, .	2.0	2
3992	Exome-based gene panel analysis in a cohort of acute juvenile ischemic stroke patients:relevance of NOTCH3 and GLA variants. <i>Journal of Neurology</i> , 2023, 270, 1501-1511.	1.8	5
3993	From Samples to Germline and Somatic Sequence Variation: A Focus on Next-Generation Sequencing in Melanoma Research. <i>Life</i> , 2022, 12, 1939.	1.1	1
3995	Greater genetic diversity is needed in human pluripotent stem cell models. <i>Nature Communications</i> , 2022, 13, .	5.8	13
3997	Using coding and non-coding rare variants to target candidate genes in patients with severe tinnitus. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	8
3998	Analysis of 363 Genetic Variants in F5 via an Interactive Web Database Reveals New Insights into FV Deficiency and FV Leiden. <i>TH Open</i> , 2023, 07, e30-e41.	0.7	2
4000	Genetic and phenotypic spectrum in the <sc><i>NONO</i></sc>â€associated syndromic disorder. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 469-478.	0.7	5
4001	Common and rare variant associations with clonal haematopoiesis phenotypes. <i>Nature</i> , 2022, 612, 301-309.	13.7	74
4002	Analysis of ProP1 Gene in a Cohort of Tunisian Patients with Congenital Combined Pituitary Hormone Deficiency. <i>Journal of Clinical Medicine</i> , 2022, 11, 7525.	1.0	2
4003	Rare copy number variants in males and females with childhood attention-deficit/hyperactivity disorder. <i>Molecular Psychiatry</i> , 2023, 28, 1240-1247.	4.1	1
4006	Estimation of ENPP1 deficiency genetic prevalence using a comprehensive literature review and population databases. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, .	1.2	6
4007	Evaluation of in silico predictors on short nucleotide variants in HBA1, HBA2, and HBB associated with haemoglobinopathies. <i>ELife</i> , 0, 11, .	2.8	6
4008	5q35 duplication syndrome: Narrowing the critical region on the distal side and further evidence of intrafamilial variability and expression. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	0
4009	Characterization of a novel nonâ€canonical splice site variant (c.886â€T>A) in <i>NBAS</i> and description of the associated phenotype. <i>Molecular Genetics & Genomic Medicine</i> , 0, , .	0.6	1
4011	Novel compound heterozygote variants: c.4193_4206delinsG (p.Leu1398Argfs*25), c.793C>A (p.Pro265Thr), in the CPS1 gene (NM_001875.4) causing late onset carbamoyl phosphate synthetase 1 deficiencyâ€Lessons learned. <i>Molecular Genetics and Metabolism Reports</i> , 2022, 33, 100942.	0.4	2
4012	Genetic landscape of human neutrophil antigen variants in India from populationâ€scale genomes. <i>Hla</i> , 0, , .	0.4	0
4013	Recurrent RNA edits in human preimplantation potentially enhance maternal mRNA clearance. <i>Communications Biology</i> , 2022, 5, .	2.0	0
4014	Mutagenesis studies of TRPV1 subunit interfaces informed by genomic variant analysis. <i>Biophysical Journal</i> , 2023, 122, 322-332.	0.2	0

#	ARTICLE	IF	CITATIONS
4015	Loss of surface transport is a main cellular pathomechanism of CRB2 variants causing podocytopathies. <i>Life Science Alliance</i> , 2023, 6, e202201649.	1.3	0
4017	Complex molecular profile of DNA repair genes in epithelial ovarian carcinoma patients with different sensitivity to platinum-based therapy. <i>Frontiers in Oncology</i> , 0, 12, .	1.3	2
4018	Abundant copathologies of polyglucosan bodies, frontotemporal lobar degeneration with TDP43 inclusions and ageing-related tau astroglipathy in a family with a <i>GBE1</i> mutation. <i>Neuropathology and Applied Neurobiology</i> , 2023, 49, .	1.8	3
4019	Hiding in plain sight: genetics of childhood steroid-resistant nephrotic syndrome in Sub-Saharan Africa. <i>Pediatric Nephrology</i> , 0, , .	0.9	2
4020	Deep Intronic <i>FGF14</i> GAA Repeat Expansion in Late-Onset Cerebellar Ataxia. <i>New England Journal of Medicine</i> , 2023, 388, 128-141.	13.9	70
4021	STRling: a k-mer counting approach that detects short tandem repeat expansions at known and novel loci. <i>Genome Biology</i> , 2022, 23, .	3.8	17
4023	Evaluating Genetic Disorders in the Neonate: The Role of Exome Sequencing in the NICU. <i>NeoReviews</i> , 2022, 23, e829-e840.	0.4	1
4025	A robust pipeline for ranking carrier frequencies of autosomal recessive and X-linked Mendelian disorders. <i>Npj Genomic Medicine</i> , 2022, 7, .	1.7	3
4027	ARHGAP35 is a novel factor disrupted in human developmental eye phenotypes. <i>European Journal of Human Genetics</i> , 2023, 31, 363-367.	1.4	4
4028	Arginase deficiency in Bulgaria: first cases and potential endemic region for the disorder. <i>Journal of Genetics</i> , 2023, 102, .	0.4	0
4029	Tackling hypo and hyper sensory processing heterogeneity in autism: From clinical stratification to genetic pathways. <i>Autism Research</i> , 0, , .	2.1	4
4030	Heterozygous and homozygous variants in <i>STX1A</i> cause a neurodevelopmental disorder with or without epilepsy. <i>European Journal of Human Genetics</i> , 2023, 31, 345-352.	1.4	5
4031	Maternal Copy Number Imbalances in Non-Invasive Prenatal Testing: Do They Matter?. <i>Diagnostics</i> , 2022, 12, 3056.	1.3	0
4032	Continuous Bayesian variant interpretation accounts for incomplete penetrance among Mendelian cardiac channelopathies. <i>Genetics in Medicine</i> , 2023, 25, 100355.	1.1	4
4033	Clinical characteristics and survival analysis of Chinese ovarian cancer patients with <i>RAD51D</i> germline mutations. <i>BMC Cancer</i> , 2022, 22, .	1.1	2
4034	Craniosynostosis, inner ear, and renal anomalies in a child with complete loss of <i>SPRY1</i> (sprouty) Tj ETQq1 1 0.784314 ggBT /Over	1.5	2
4035	Prevalence and Penetrance of Rare Pathogenic Variants in Neurodevelopmental Psychiatric Genes in a Health Care System Population. <i>American Journal of Psychiatry</i> , 2023, 180, 65-72.	4.0	7
4036	Prevalence, mutational spectrum and clinical implications of clonal hematopoiesis of indeterminate potential in plasma cell dyscrasias. <i>Seminars in Oncology</i> , 2022, 49, 465-475.	0.8	5

#	ARTICLE	IF	CITATIONS
4037	An ELF4 hypomorphic variant results in NK cell deficiency. <i>JCI Insight</i> , 2022, 7, .	2.3	3
4038	Disruption of the HIF-1 pathway in individuals with Ollier disease and Maffucci syndrome. <i>PLoS Genetics</i> , 2022, 18, e1010504.	1.5	2
4039	Diverse monogenic subforms of human spermatogenic failure. <i>Nature Communications</i> , 2022, 13, .	5.8	17
4041	Biobanking as a Tool for Genomic Research: From Allele Frequencies to Cross-Ancestry Association Studies. <i>Journal of Personalized Medicine</i> , 2022, 12, 2040.	1.1	3
4042	Integrated exome and transcriptome analysis prioritizes MAP4K4 de novo frameshift variants in autism spectrum disorder as a novel disease-associated gene association. <i>Human Genetics</i> , 2023, 142, 343-350.	1.8	2
4043	Reconciling Mouse and Human Immunology at the Altar of Genetics. <i>Annual Review of Immunology</i> , 2023, 41, 39-71.	9.5	10
4044	A minimal role for synonymous variation in human disease. <i>American Journal of Human Genetics</i> , 2022, 109, 2105-2109.	2.6	13
4046	Estimation of intrafamilial DNA contamination in family trio genome sequencing using deviation from Mendelian inheritance. <i>Genome Research</i> , 0, , .	2.4	1
4047	Human genetic diversity alters off-target outcomes of therapeutic gene editing. <i>Nature Genetics</i> , 2023, 55, 34-43.	9.4	28
4048	Expansion of the phenotypic and molecular spectrum of <i>CWF19L1</i> -related disorder. <i>Clinical Genetics</i> , 2023, 103, 566-573.	1.0	1
4049	Discovery and systematic characterization of risk variants and genes for coronary artery disease in over a million participants. <i>Nature Genetics</i> , 2022, 54, 1803-1815.	9.4	150
4050	The structure of the human LACTB filament reveals the mechanisms of assembly and membrane binding. <i>PLoS Biology</i> , 2022, 20, e3001899.	2.6	4
4051	A Missense Variant in PDK1 Associated with Severe Neurodevelopmental Delay and Epilepsy. <i>Biomedicines</i> , 2022, 10, 3171.	1.4	1
4052	<i>KMT2A</i> pathogenicity, prevalence, and variation according to a population database. <i>Cancer Medicine</i> , 2023, 12, 7234-7245.	1.3	1
4053	A reference human induced pluripotent stem cell line for large-scale collaborative studies. <i>Cell Stem Cell</i> , 2022, 29, 1685-1702.e22.	5.2	59
4054	Targeting de novo loss-of-function variants in constrained disease genes improves diagnostic rates in the 100,000 Genomes Project. <i>Human Genetics</i> , 2023, 142, 351-362.	1.8	7
4056	Genome-wide analysis of copy-number variation in humans with cleft lip and/or cleft palate identifies COBLL1, RIC1, and ARHGEF38 as clefting genes. <i>American Journal of Human Genetics</i> , 2023, 110, 71-91.	2.6	4
4057	Efficient identification of trait-associated loss-of-function variants in the UK Biobank cohort by exome-sequencing based genotype imputation. <i>Genetic Epidemiology</i> , 2023, 47, 121-134.	0.6	3

#	ARTICLE	IF	CITATIONS
4058	Whole-Exome Sequencing Study of Consanguineous Parkinson's Disease Families and Related Phenotypes: Report of Twelve Novel Variants. <i>Journal of Molecular Neuroscience</i> , 0, , .	1.1	0
4059	Myoclonic-Atonic Epilepsy Caused by a Novel de Novo Heterozygous Missense Variant in the SLC6A1 Gene: Brief Discussion of the Literature and Detailed Case Description of a Severely Intellectually Disabled Adult Male Patient. <i>International Medical Case Reports Journal</i> , 0, Volume 15, 753-759.	0.3	4
4060	<i>CHD8</i> suppression impacts on histone H3 lysine 36 trimethylation and alters RNA alternative splicing. <i>Nucleic Acids Research</i> , 2022, 50, 12809-12828.	6.5	9
4062	A method to build extended sequence context models of point mutations and indels. <i>Nature Communications</i> , 2022, 13, .	5.8	4
4063	Genome-wide data from medieval German Jews show that the Ashkenazi founder event pre-dated the 14th century. <i>Cell</i> , 2022, 185, 4703-4716.e16.	13.5	12
4064	Optical genome mapping and revisiting short-read genome sequencing data reveal previously overlooked structural variants disrupting retinal disease-associated genes. <i>Genetics in Medicine</i> , 2023, 25, 100345.	1.1	9
4065	Clustered variants in the 5' coding region of <i>TRA2B</i> cause a distinctive neurodevelopmental syndrome. <i>Genetics in Medicine</i> , 2023, 25, 100003.	1.1	1
4066	Histopathologic and proteogenomic heterogeneity reveals features of clear cell renal cell carcinoma aggressiveness. <i>Cancer Cell</i> , 2023, 41, 139-163.e17.	7.7	43
4067	Three-tiered EGFR domain risk stratification for individualized NOTCH3-small vessel disease prediction. <i>Brain</i> , 2023, 146, 2913-2927.	3.7	6
4068	A novel <i>PLS1</i> c.981+1G variant causes autosomal dominant hereditary hearing loss in a family. <i>Clinical Genetics</i> , 2023, 103, 413-423.	1.0	1
4069	Evaluating gap junction variants for a role in pediatric cataract: an overview of the genetic landscape and clinical classification of variants in the <i>GJA3</i> and <i>GJA8</i> genes. <i>Expert Review of Ophthalmology</i> , 2023, 18, 71-95.	0.3	1
4070	A recurrent de novo splice site variant involving <i>DNM1</i> exon 10a causes developmental and epileptic encephalopathy through a dominant-negative mechanism. <i>American Journal of Human Genetics</i> , 2022, 109, 2253-2269.	2.6	16
4071	Identification of de novo variants in nonsyndromic cleft lip with/without cleft palate patients with low polygenic risk scores. <i>Molecular Genetics & Genomic Medicine</i> , 0, , .	0.6	2
4072	What Is the True HbA1c? A HbA1c Peak in the Absence of HbA in an Adult Patient without Sickle Cell Disease. <i>Journal of Applied Laboratory Medicine</i> , The, 0, , .	0.6	0
4073	Brain metastatic outgrowth and osimertinib resistance are potentiated by RhoA in EGFR-mutant lung cancer. <i>Nature Communications</i> , 2022, 13, .	5.8	6
4074	Variants in <i>CLDN5</i> cause a syndrome characterized by seizures, microcephaly and brain calcifications. <i>Brain</i> , 2023, 146, 2285-2297.	3.7	4
4076	The different clinical facets of <i>SYN1</i> -related neurodevelopmental disorders. <i>Frontiers in Cell and Developmental Biology</i> , 0, 10, .	1.8	7
4077	The structure, binding and function of a Notch transcription complex involving RBPJ and the epigenetic reader protein L3MBTL3. <i>Nucleic Acids Research</i> , 0, , .	6.5	1

#	ARTICLE	IF	CITATIONS
4078	Exploring biomarkers for prognosis and neoadjuvant chemosensitivity in rectal cancer: Multi-omics and ctDNA sequencing collaboration. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	2
4079	Will a hyperactive classical complement pathway exacerbate autoimmune diseases?. <i>Autoimmunity Reviews</i> , 2022, , 103241.	2.5	0
4081	Optimising clinical care through <i>CDH1</i> -specific germline variant curation: improvement of clinical assertions and updated curation guidelines. <i>Journal of Medical Genetics</i> , 2023, 60, 568-575.	1.5	5
4083	DNA methylation epesignatures: insight into copy number variation. <i>Epigenomics</i> , 2022, 14, 1373-1388.	1.0	7
4084	Mediation of the Same Epigenetic and Transcriptional Effect by Independent Osteoarthritis Risk-Conferring Alleles on a Shared Target Gene, <i>COLGALT2</i> . <i>Arthritis and Rheumatology</i> , 2023, 75, 910-922.	2.9	5
4085	Language and Communication Deficits in Chromosome 16p11.2 Deletion Syndrome. <i>Journal of Speech, Language, and Hearing Research</i> , 2022, 65, 4724-4740.	0.7	3
4086	Genomic, transcriptomic and RNA editing analysis of human MM1 and VV2 sporadic Creutzfeldt-Jakob disease. <i>Acta Neuropathologica Communications</i> , 2022, 10, .	2.4	0
4087	Genotype-phenotype characterisation of long survivors with motor neuron disease in Scotland. <i>Journal of Neurology</i> , 2023, 270, 1702-1712.	1.8	4
4088	Tobacco Smoking-Related Mutational Signatures in Classifying Smoking-Associated and Nonsmoking-Associated NSCLC. <i>Journal of Thoracic Oncology</i> , 2023, 18, 487-498.	0.5	14
4089	MUG: A mutation overview of GPCR subfamily A17 receptors. <i>Computational and Structural Biotechnology Journal</i> , 2023, 21, 586-600.	1.9	0
4090	Approach to Cohort-Wide Re-Analysis of Exome Data in 1000 Individuals with Neurodevelopmental Disorders. <i>Genes</i> , 2023, 14, 30.	1.0	2
4091	Precision medicine for developmental and epileptic encephalopathies in Africa- strategies for a resource-limited setting. <i>Genetics in Medicine</i> , 2023, 25, 100333.	1.1	0
4092	Aging is associated with a systemic length-associated transcriptome imbalance. <i>Nature Aging</i> , 2022, 2, 1191-1206.	5.3	28
4093	Analysis of miRNA rare variants in amyotrophic lateral sclerosis and in silico prediction of their biological effects. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1
4095	Predicting response to immune checkpoint blockade in NSCLC with tumour-only RNA-seq. <i>British Journal of Cancer</i> , 2023, 128, 1148-1154.	2.9	2
4097	TogoVar: A comprehensive Japanese genetic variation database. <i>Human Genome Variation</i> , 2022, 9, .	0.4	4
4099	Dynamic spatiotemporal determinants modulate GPCR:G protein coupling selectivity and promiscuity. <i>Nature Communications</i> , 2022, 13, .	5.8	25
4100	A multiple sclerosis-protective coding variant reveals an essential role for HDAC7 in regulatory T cells. <i>Science Translational Medicine</i> , 2022, 14, .	5.8	8

#	ARTICLE	IF	CITATIONS
4101	Two unique BAP1 pathogenic variants identified in the same family by panel cascade testing. <i>Familial Cancer</i> , 0, , .	0.9	0
4102	UQCRC1 variants in early-onset and familial Parkinson's disease in a Taiwanese cohort. <i>Frontiers in Neurology</i> , 0, 13, .	1.1	1
4103	Calculating variant penetrance from family history of disease and average family size in population-scale data. <i>Genome Medicine</i> , 2022, 14, .	3.6	3
4104	HNF1B Alters an Evolutionarily Conserved Nephrogenic Program of Target Genes. <i>Journal of the American Society of Nephrology: JASN</i> , 2023, 34, 412-432.	3.0	5
4105	Whole exome sequencing in dense families suggests genetic pleiotropy amongst Mendelian and complex neuropsychiatric syndromes. <i>Scientific Reports</i> , 2022, 12, .	1.6	2
4106	High molecular diagnostic yields and novel phenotypic expansions involving syndromic anorectal malformations. <i>European Journal of Human Genetics</i> , 0, , .	1.4	4
4108	Identification and drug metabolic characterization of four new CYP2C9 variants CYP2C9*72-*75 in the Chinese Han population. <i>Frontiers in Pharmacology</i> , 0, 13, .	1.6	3
4110	Determination of regulatory motifs and pathogenicity of intronic variants of GNPTAB, GNPTG, and NAGPA genes in individuals with stuttering. <i>Bulletin of the National Research Centre</i> , 2022, 46, .	0.7	0
4111	Rare Variants in Genes of the Cholesterol Pathway Are Present in 60% of Patients with Acute Myocardial Infarction. <i>International Journal of Molecular Sciences</i> , 2022, 23, 16127.	1.8	2
4112	Dual Molecular Diagnoses of Recessive Disorders in a Child from Consanguineous Parents: Case Report and Literature Review. <i>Genes</i> , 2022, 13, 2377.	1.0	1
4113	A novel compound heterozygous BEST1 gene mutation in two siblings causing autosomal recessive bestrophinopathy. <i>BMC Ophthalmology</i> , 2022, 22, .	0.6	0
4114	Genetic architecture and evolution of color variation in American black bears. <i>Current Biology</i> , 2023, 33, 86-97.e10.	1.8	9
4115	A biallelic loss of function variant in <i>HORMAD1</i> within a large consanguineous Turkish family is associated with spermatogenic arrest. <i>Human Reproduction</i> , 0, , .	0.4	2
4117	Population-based analysis of <i>POT1</i> variants in a cutaneous melanoma caseâ€“control cohort. <i>Journal of Medical Genetics</i> , 2023, 60, 692-696.	1.5	4
4118	Wholeâ€“exome sequencing of a Saudi epilepsy cohort reveals association signals in known and potentially novel loci. <i>Human Genomics</i> , 2022, 16, .	1.4	0
4122	Biallelic variants in OGDH encoding oxoglutarate dehydrogenase lead to a neurodevelopmental disorder characterized by global developmental delay, movement disorder, and metabolic abnormalities. <i>Genetics in Medicine</i> , 2023, 25, 100332.	1.1	4
4124	Perspectives on the future of dysmorphology. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 659-671.	0.7	9
4127	Genotype-Phenotype Correlation of Distal 2q37 Deletions. <i>Cytogenetic and Genome Research</i> , 0, , 1-7.	0.6	0

#	ARTICLE	IF	CITATIONS
4128	A generalizable deep learning framework for inferring fine-scale germline mutation rate maps. <i>Nature Machine Intelligence</i> , 2022, 4, 1209-1223.	8.3	5
4129	A Comprehensive Biomarker Analysis of Microsatellite Unstable/Mismatch Repair Deficient Colorectal Cancer Cohort Treated with Immunotherapy. <i>International Journal of Molecular Sciences</i> , 2023, 24, 118.	1.8	2
4130	Application of Long-Read Nanopore Sequencing to the Search for Mutations in Hypertrophic Cardiomyopathy. <i>International Journal of Molecular Sciences</i> , 2022, 23, 15845.	1.8	4
4131	De novo mutation hotspots in homologous protein domains identify function-altering mutations in neurodevelopmental disorders. <i>American Journal of Human Genetics</i> , 2023, 110, 92-104.	2.6	3
4132	<sc>RosettaDDGPrediction</sc> for high-throughput mutational scans: From stability to binding. <i>Protein Science</i> , 2023, 32, .	3.1	15
4133	Genetic risk factors of food allergy: a review of genome-wide studies. <i>Russian Journal of Allergy</i> , 0, , .	0.1	0
4134	<sc>ZDHHC15</sc> as a candidate gene for autism spectrum disorder. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 941-947.	0.7	2
4135	Calibration of computational tools for missense variant pathogenicity classification and ClinGen recommendations for PP3/BP4 criteria. <i>American Journal of Human Genetics</i> , 2022, 109, 2163-2177.	2.6	124
4136	Rapid Targeted Sequencing Using Dried Blood Spot Samples for Patients With Suspected Actionable Genetic Diseases. <i>Annals of Laboratory Medicine</i> , 2023, 43, 280-289.	1.2	4
4137	Functional genomics for curation of variants in telomere biology disorder associated genes: A systematic review. <i>Genetics in Medicine</i> , 2023, 25, 100354.	1.1	2
4138	Rescue of Misfolded Organic Cation Transporter 3 Variants. <i>Cells</i> , 2023, 12, 39.	1.8	1
4139	Exome-wide association analysis of CT imaging-derived hepatic fat in a medical biobank. <i>Cell Reports Medicine</i> , 2022, 3, 100855.	3.3	3
4140	Genome-wide assessment reveals a significant association between <sc>ACSS3</sc> and physical activity. <i>Genes, Brain and Behavior</i> , 2023, 22, .	1.1	1
4142	VPatho: a deep learning-based two-stage approach for accurate prediction of gain-of-function and loss-of-function variants. <i>Briefings in Bioinformatics</i> , 2023, 24, .	3.2	5
4143	Progressive multifocal leukoencephalopathy genetic risk variants for pharmacovigilance of immunosuppressant therapies. <i>Frontiers in Neurology</i> , 0, 13, .	1.1	7
4144	A method for multiplexed full-length single-molecule sequencing of the human mitochondrial genome. <i>Nature Communications</i> , 2022, 13, .	5.8	12
4145	Loss of the Immunomodulatory Transcription Factor BATF2 in Humans Is Associated with a Neurological Phenotype. <i>Cells</i> , 2023, 12, 227.	1.8	2
4146	Heterozygous mutations in SOX2 may cause idiopathic hypogonadotropic hypogonadism via dominant-negative mechanisms. <i>JCI Insight</i> , 2023, 8, .	2.3	0

#	ARTICLE	IF	CITATIONS
4149	X-linked C1GALT1C1 mutation causes atypical hemolytic uremic syndrome. <i>European Journal of Human Genetics</i> , 2023, 31, 1101-1107.	1.4	3
4150	The frequency of somatic mutations in cancer predicts the phenotypic relevance of germline mutations. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1
4151	Deep learning predicts the impact of regulatory variants on cell-type specific enhancers in the brain. <i>Bioinformatics Advances</i> , 0, , .	0.9	0
4152	Characterizing asparagine synthetase deficiency variants in lymphoblastoid cell lines. <i>JIMD Reports</i> , 2023, 64, 167-179.	0.7	4
4153	TREM2 has a significant, gender-specific, effect on human obesity. <i>Scientific Reports</i> , 2023, 13, .	1.6	6
4154	Expanded genetic testing of GIST patients identifies high proportion of non-syndromic patients with germline alterations. <i>Npj Precision Oncology</i> , 2023, 7, .	2.3	9
4155	RNA editing: Expanding the potential of RNA therapeutics. <i>Molecular Therapy</i> , 2023, 31, 1533-1549.	3.7	19
4156	Fast and reliable detection of repeat expansions in spinocerebellar ataxia using exomes. <i>Journal of Medical Genetics</i> , 2023, 60, 717-721.	1.5	5
4157	Deletion of first noncoding exon in <i>ANKRD11</i> leads to <i>KBG</i> syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 1044-1049.	0.7	1
4158	Quantitative differentiation of benign and misfolded glaucoma-causing myocilin variants on the basis of protein thermal stability. <i>DMM Disease Models and Mechanisms</i> , 2023, 16, .	1.2	3
4159	BMP2 Variants Underlie Nonsyndromic Oligodontia. <i>International Journal of Molecular Sciences</i> , 2023, 24, 1648.	1.8	0
4160	<i>AQP5</i> , a second gene at play with <i>CFTR</i> in aquagenic palmoplantar keratoderma. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2023, 37, .	1.3	2
4161	PHIP-associated Chung-Jansen syndrome: Report of 23 new individuals. <i>Frontiers in Cell and Developmental Biology</i> , 0, 10, .	1.8	6
4162	Identifying biomarkers of differential chemotherapy response in TNBC patient-derived xenografts with a CTD/WGCNA approach. <i>IScience</i> , 2023, 26, 105799.	1.9	12
4163	Mutations of TP53 and genes related to homologous recombination repair in breast cancer with germline BRCA1/2 mutations. <i>Human Genomics</i> , 2023, 17, .	1.4	3
4164	Biallelic loss of function variant in the <i>NRCAM</i> gene is associated with motor-predominant axonal polyneuropathy; the second report. <i>Molecular Genetics & Genomic Medicine</i> , 0, , .	0.6	1
4166	Genome-wide screen of otosclerosis in population biobanks: 27 loci and shared associations with skeletal structure. <i>Nature Communications</i> , 2023, 14, .	5.8	2
4167	Deleterious synonymous mutation identification based on selective ensemble strategy. <i>Briefings in Bioinformatics</i> , 0, , .	3.2	0

#	ARTICLE	IF	CITATIONS
4168	A Unique Role for Protocadherin β 3 in Promoting Dendrite Arborization through an Axin1-Dependent Mechanism. <i>Journal of Neuroscience</i> , 2023, 43, 918-935.	1.7	9
4169	Expanding the phenotypic spectrum of KCNK4: From syndromic neurodevelopmental disorder to rolandic epilepsy. <i>Frontiers in Molecular Neuroscience</i> , 0, 15, .	1.4	4
4171	Identification and characterization of novel compound heterozygous variants in FSHR causing primary ovarian insufficiency with resistant ovary syndrome. <i>Frontiers in Endocrinology</i> , 0, 13, .	1.5	1
4172	Genomic analysis as a tool to infer disparate phylogenetic origins of dysembryoplastic neuroepithelial tumors and their satellite lesions. <i>Scientific Reports</i> , 2023, 13, .	1.6	1
4173	Gene–Folic Acid Interactions and Risk of Conotruncal Heart Defects: Results from the National Birth Defects Prevention Study. <i>Genes</i> , 2023, 14, 180.	1.0	2
4174	Systematic errors in annotations of truncations, loss-of-function and synonymous variants. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	4
4175	Skeletal anomaly and opisthotonus in early-onset epileptic encephalopathy with KCNQ2 abnormality. <i>Brain and Development</i> , 2023, 45, 231-236.	0.6	1
4176	Detection of pathogenic variants in Alzheimer’s disease related genes in Bulgarian patients by pooled whole-exome sequencing. <i>Biotechnology and Biotechnological Equipment</i> , 2023, 37, 74-78.	0.5	0
4177	Transcriptomic profiling and genomic rearrangement landscape of Nigerian prostate cancer. <i>Prostate</i> , 0, , .	1.2	0
4178	Diagnostic Yield of Genetic Testing for Ocular and Oculocutaneous Albinism in a Diverse United States Pediatric Population. <i>Genes</i> , 2023, 14, 135.	1.0	2
4179	A novel NONO variant that causes developmental delay and cardiac phenotypes. <i>Scientific Reports</i> , 2023, 13, .	1.6	1
4180	Structure of SALL4 zinc finger domain reveals link between AT-rich DNA binding and Okhiro syndrome. <i>Life Science Alliance</i> , 2023, 6, e202201588.	1.3	2
4182	Myofilament-associated proteins with intrinsic disorder (MAPIDs) and their resolution by computational modeling. <i>Quarterly Reviews of Biophysics</i> , 2023, 56, .	2.4	3
4184	MYB/MYBL1::QKI fusion-positive diffuse glioma. <i>Journal of Neuropathology and Experimental Neurology</i> , 2023, 82, 250-260.	0.9	3
4186	Genetic adaptation to pathogens and increased risk of inflammatory disorders in post-Neolithic Europe. <i>Cell Genomics</i> , 2023, 3, 100248.	3.0	19
4188	Genotype first: Clinical genomics research through a reverse phenotyping approach. <i>American Journal of Human Genetics</i> , 2023, 110, 3-12.	2.6	15
4189	Capturing the conversion of the pathogenic alpha-1-antitrypsin fold by ATF6 enhanced proteostasis. <i>Cell Chemical Biology</i> , 2023, 30, 22-42.e5.	2.5	6
4190	The impact of single nucleotide polymorphisms on return-to-work after taxane-based chemotherapy in breast cancer. <i>Cancer Chemotherapy and Pharmacology</i> , 0, , .	1.1	0

#	ARTICLE	IF	CITATIONS
4191	Familial multiple discoid fibromas is linked to a locus on chromosome 5 including the FNIP1 gene. <i>Journal of Human Genetics</i> , 2023, 68, 273-279.	1.1	3
4192	Inherited mutations in Chinese patients with upper tract urothelial carcinoma. <i>Cell Reports Medicine</i> , 2023, 4, 100883.	3.3	0
4193	Association between HLA DNA Variants and Long-Term Response to Anti-TNF Drugs in a Spanish Pediatric Inflammatory Bowel Disease Cohort. <i>International Journal of Molecular Sciences</i> , 2023, 24, 1797.	1.8	6
4194	Cargo selection in endoplasmic reticulum to Golgi transport and relevant diseases. <i>Journal of Clinical Investigation</i> , 2023, 133, .	3.9	7
4195	Genome-wide association study of varicose veins identifies a protective missense variant in GJD3 enriched in the Finnish population. <i>Communications Biology</i> , 2023, 6, .	2.0	4
4196	Bi-allelic variants in NAE1 cause intellectual disability, ischiopubic hypoplasia, stress-mediated lymphopenia and neurodegeneration. <i>American Journal of Human Genetics</i> , 2023, 110, 146-160.	2.6	1
4197	Interpreting the molecular mechanisms of disease variants in human transmembrane proteins. <i>Biophysical Journal</i> , 2023, 122, 2176-2191.	0.2	7
4199	Association of Genetic Diagnoses for Childhood-Onset Hearing Loss With Cochlear Implant Outcomes. <i>JAMA Otolaryngology - Head and Neck Surgery</i> , 2023, 149, 212.	1.2	8
4200	Functional Characterization of a Spectrum of Novel Romano-Ward Syndrome KCNQ1 Variants. <i>International Journal of Molecular Sciences</i> , 2023, 24, 1350.	1.8	2
4201	A Lower Frequency of Spliceosome Mutations Distinguishes Clonal Cytopenias of Undetermined Significance From Low-Risk Myelodysplastic Syndromes, Despite Inherent Similarities in Genomic, Laboratory, and Clinical Features. <i>Modern Pathology</i> , 2023, 36, 100068.	2.9	0
4202	Genetic predictors of lifelong medication-use patterns in cardiometabolic diseases. <i>Nature Medicine</i> , 2023, 29, 209-218.	15.2	7
4203	Calmodulin Mutations in Human Disease. <i>Channels</i> , 2023, 17, .	1.5	13
4204	Control-independent mosaic single nucleotide variant detection with DeepMosaic. <i>Nature Biotechnology</i> , 2023, 41, 870-877.	9.4	13
4205	Genetic Variant in GRM1 Underlies Congenital Cerebellar Ataxia with No Obvious Intellectual Disability. <i>International Journal of Molecular Sciences</i> , 2023, 24, 1551.	1.8	3
4206	Relating pathogenic loss-of-function mutations in humans to their evolutionary fitness costs. <i>ELife</i> , 0, 12, .	2.8	12
4207	Random allelic expression in the adult human body. <i>Cell Reports</i> , 2023, 42, 111945.	2.9	10
4209	New insights from the last decade of research in psychiatric genetics: discoveries, challenges and clinical implications. <i>World Psychiatry</i> , 2023, 22, 4-24.	4.8	38
4210	DeepSom: a CNN-based approach to somatic variant calling in WGS samples without a matched normal. <i>Bioinformatics</i> , 2023, 39, .	1.8	1

#	ARTICLE	IF	CITATIONS
4212	A novel frameshift mutation in TRPV6 is associated with hereditary pancreatitis. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1
4213	Biochemical characterization of two novel mutations in the human high-affinity choline transporter 1 identified in a patient with congenital myasthenic syndrome. <i>Human Molecular Genetics</i> , 0, , .	1.4	0
4215	Rare variants in IMPDH2 cause autosomal dominant dystonia in Chinese population. <i>Journal of Neurology</i> , 2023, 270, 2197-2203.	1.8	3
4216	Allelic prevalence and geographic distribution of cerebrotendinous xanthomatosis. <i>Orphanet Journal of Rare Diseases</i> , 2023, 18, .	1.2	8
4217	The global prevalence and ethnic heterogeneity of iron-refractory iron deficiency anaemia. <i>Orphanet Journal of Rare Diseases</i> , 2023, 18, .	1.2	2
4218	Allogenic Adipose-Derived Stem Cells in Diabetic Foot Ulcer Treatment: Clinical Effectiveness, Safety, Survival in the Wound Site, and Proteomic Impact. <i>International Journal of Molecular Sciences</i> , 2023, 24, 1472.	1.8	2
4219	Common pathophysiology for <sc>ANXA11</sc> disorders caused by aspartate 40 variants. <i>Annals of Clinical and Translational Neurology</i> , 0, , .	1.7	0
4222	Annotation of uORFs in the OMIM genes allows to reveal pathogenic variants in 5â€²UTRs. <i>Nucleic Acids Research</i> , 2023, 51, 1229-1244.	6.5	3
4223	Functional genomics provide key insights to improve the diagnostic yield of hereditary ataxia. <i>Brain</i> , 2023, 146, 2869-2884.	3.7	4
4224	Functional Impact of a Cancer-Related Variant in Human Î” ¹ -Pyrroline-5-Carboxylate Reductase 1. <i>ACS Omega</i> , 0, , .	1.6	0
4225	Classification of <sc>GBA1</sc> Variants in Parkinson's Disease: The <sc>GBA1</sc> Browser. <i>Movement Disorders</i> , 2023, 38, 489-495.	2.2	18
4226	Enrichment of titin-truncating variants in exon 327 in dilated cardiomyopathy and its relevance to reduced nonsense-mediated mRNA decay efficiency. <i>Frontiers in Genetics</i> , 0, 13, .	1.1	1
4227	Distinct Roles of Histone Lysine Demethylases and Methyltransferases in Developmental Eye Disease. <i>Genes</i> , 2023, 14, 216.	1.0	1
4228	Paroxysmal nocturnal hemoglobinuria: Where we stand. <i>American Journal of Hematology</i> , 2023, 98, .	2.0	3
4229	Karyopherin Î± deficiency contributes to human preimplantation embryo arrest. <i>Journal of Clinical Investigation</i> , 2023, 133, .	3.9	13
4230	Identifying somatic changes in drug transporters using whole genome and transcriptome sequencing data of advanced tumors. <i>Biomedicine and Pharmacotherapy</i> , 2023, 159, 114210.	2.5	0
4231	Primary failure of eruption: From molecular diagnosis to therapeutic management. <i>Journal of Oral Biology and Craniofacial Research</i> , 2023, 13, 169-176.	0.8	3
4232	Biochemical, Clinical, and Genetic Characteristics of Mexican Patients with Primary Hypertriglyceridemia, Including the First Case of Hyperchylomicronemia Syndrome Due to GPIHBP1 Deficiency. <i>International Journal of Molecular Sciences</i> , 2023, 24, 465.	1.8	0

#	ARTICLE	IF	CITATIONS
4233	Genome-Wide Association and Inheritance-Based Analyses Implicate Unconventional Myosin Genes in Hypoplastic Left Heart Syndrome. <i>Circulation Genomic and Precision Medicine</i> , 0, , .	1.6	0
4235	High-Throughput Prediction of the Impact of Genetic Variability on Drug Sensitivity and Resistance Patterns for Clinically Relevant Epidermal Growth Factor Receptor Mutations from Atomistic Simulations. <i>Journal of Chemical Information and Modeling</i> , 2023, 63, 321-334.	2.5	1
4236	Variants Tagging LGALS-3 Haplotype Block in Association with First Myocardial Infarction and Plasma Galectin-3 Six Months after the Acute Event. <i>Genes</i> , 2023, 14, 109.	1.0	2
4237	Whole-Exome Sequencing Identifies Genetic Variants for Severe Adolescent Idiopathic Scoliosis in a Taiwanese Population. <i>Journal of Personalized Medicine</i> , 2023, 13, 32.	1.1	3
4238	Novel genes linked to Class II Division 1 malocclusion with mandibular micrognathism. <i>American Journal of Orthodontics and Dentofacial Orthopedics</i> , 2022, , .	0.8	1
4239	Investigation of Rare Non-Coding Variants in Familial Multiple Myeloma. <i>Cells</i> , 2023, 12, 96.	1.8	2
4240	<i>UGT2B7 c.-161C>>T</i> polymorphism frequency in Croatian population. <i>Arhiv Za Higijenu Rada I Toksikologiju</i> , 2022, 73, 303-307.	0.4	0
4241	Low Frequency of Cancer-Predisposition Gene Mutations in Liver Transplant Candidates with Hepatocellular Carcinoma. <i>Cancers</i> , 2023, 15, 201.	1.7	2
4242	Gingival Overgrowths Revealing PTEN Hamartoma Tumor Syndrome: Report of Novel PTEN Pathogenic Variants. <i>Biomedicines</i> , 2023, 11, 81.	1.4	1
4243	Identification of <i>TCF3</i> germline variants in pediatric B-cell acute lymphoblastic leukemia. <i>Blood Advances</i> , 2023, 7, 2177-2180.	2.5	3
4244	Reader Response: Use of Whole-Genome Sequencing for Mitochondrial Disease Diagnosis. <i>Neurology</i> , 2023, 100, 49-50.	1.5	0
4246	Variant Location Is a Novel Risk Factor for Individuals With Arrhythmogenic Cardiomyopathy Due to a Desmoplakin (<i>DSP</i>) Truncating Variant. <i>Circulation Genomic and Precision Medicine</i> , 2023, 16, .	1.6	6
4247	Severe high-molecular-weight kininogen deficiency: clinical characteristics, deficiencyâ€‘causing KNG1 variants, and estimated prevalence. <i>Journal of Thrombosis and Haemostasis</i> , 2023, 21, 237-254.	1.9	3
4248	The MAOA rs979605 Genetic Polymorphism Is Differentially Associated with Clinical Improvement Following Antidepressant Treatment between Male and Female Depressed Patients. <i>International Journal of Molecular Sciences</i> , 2023, 24, 497.	1.8	4
4249	Exome Sequencing of a Clinical Population for Autosomal Dominant Polycystic Kidney Disease. <i>JAMA - Journal of the American Medical Association</i> , 2022, 328, 2412.	3.8	21
4250	The role of genetic testing in diagnosis and care of inherited cardiac conditions in a specialised multidisciplinary clinic. <i>Genome Medicine</i> , 2022, 14, .	3.6	4
4251	Haplotype Structures and Protein Levels of TGFBI in HPV Infection and Cervical Lesion: A Case-Control Study. <i>Cells</i> , 2023, 12, 84.	1.8	1
4252	A De Novo Missense Variant in TUBG2 in a Child with Global Developmental Delay, Microcephaly, Refractory Epilepsy and Perisylvian Polymicrogyria. <i>Genes</i> , 2023, 14, 108.	1.0	1

#	ARTICLE	IF	CITATIONS
4253	FinnGen provides genetic insights from a well-phenotyped isolated population. <i>Nature</i> , 2023, 613, 508-518.	13.7	612
4254	The Mutation Spectrum of Rare Variants in the Gene of Adenosine Triphosphate (ATP)-Binding Cassette Subfamily C Member 8 in Patients with a MODY Phenotype in Western Siberia. <i>Journal of Personalized Medicine</i> , 2023, 13, 172.	1.1	1
4255	The Singapore National Precision Medicine Strategy. <i>Nature Genetics</i> , 2023, 55, 178-186.	9.4	9
4256	Understanding cancer predisposition in Singapore: What's next. <i>Singapore Medical Journal</i> , 2023, 64, 37.	0.3	2
4257	Cross-ancestry genome-wide analysis of atrial fibrillation unveils disease biology and enables cardioembolic risk prediction. <i>Nature Genetics</i> , 2023, 55, 187-197.	9.4	19
4259	INPP4A-related genetic and phenotypic spectrum and functional relevance of subcellular targeting of INPP4A isoforms. <i>Neurogenetics</i> , 2023, 24, 79-93.	0.7	1
4260	Harnessing the Power of Electronic Health Records and Genomics for Drug Discovery. <i>Annual Review of Pharmacology and Toxicology</i> , 2023, 63, 65-76.	4.2	4
4261	Case report: Adult-onset limb girdle muscular dystrophy in sibling pair due to novel homozygous LAMA2 missense variant. <i>Frontiers in Neurology</i> , 0, 14, .	1.1	0
4262	Mono- and biallelic variant effects on disease at biobank scale. <i>Nature</i> , 2023, 613, 519-525.	13.7	24
4263	Genetic determinants of type 1 diabetes in individuals with weak evidence of islet autoimmunity at disease onset. <i>Diabetologia</i> , 2023, 66, 695-708.	2.9	0
4265	<i>SPTSSA</i> variants alter sphingolipid synthesis and cause a complex hereditary spastic paraplegia. <i>Brain</i> , 2023, 146, 1420-1435.	3.7	9
4266	On the q.t. no more: Exposing the arrhythmic risks of dietary supplements. <i>Heart Rhythm</i> , 2023, 20, 587-588.	0.3	0
4267	APOL1 and APOL1-Associated Kidney Disease: A Common Disease, an Unusual Disease Gene â€“ Proceedings of the Henry Shavelle Professorship. <i>Glomerular Diseases</i> , 0, , 75-87.	0.2	5
4269	The relationship between caseâ€“control differential gene expression from brain tissue and genetic associations in schizophrenia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2023, 192, 85-92.	1.1	2
4272	A review of ancestry and admixture in Latin America and the caribbean focusing on native American and African descendant populations. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	6
4273	Health inequity in genomic personalized medicine in underrepresented populations: a look at the current evidence. <i>Functional and Integrative Genomics</i> , 2023, 23, .	1.4	2
4274	Case report: Pylorus-preserving pancreatoduodenectomy for focal congenital hyperinsulinism in a 5-month-old baby. <i>Frontiers in Surgery</i> , 0, 9, .	0.6	1
4275	Cardiomyopathy prevalence exceeds 30% in individuals with TTN variants and early atrial fibrillation. <i>Genetics in Medicine</i> , 2023, 25, 100012.	1.1	7

#	ARTICLE	IF	CITATIONS
4276	How does precursor RNA structure influence RNA processing and gene expression?. <i>Bioscience Reports</i> , 2023, 43, .	1.1	1
4278	Evaluating the role of CHEK2 p.(Asp438Tyr) allele in inherited breast cancer predisposition. <i>Familial Cancer</i> , 0, , .	0.9	0
4279	Understanding Insulin in the Age of Precision Medicine and Big Data: Under-Explored Nature of Genomics. <i>Biomolecules</i> , 2023, 13, 257.	1.8	1
4280	Identifying rare genetic variants in 21 highly multiplex autism families: the role of diagnosis and autistic traits. <i>Molecular Psychiatry</i> , 2023, 28, 2148-2157.	4.1	4
4281	De Novo Variant in the KCNJ9 Gene as a Possible Cause of Neonatal Seizures. <i>Genes</i> , 2023, 14, 366.	1.0	1
4282	TMEM161B regulates cerebral cortical gyration, Sonic Hedgehog signaling, and ciliary structure in the developing central nervous system. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2023, 120, .	3.3	7
4283	Electrophysiological Characterization of a <sc> <i>MYH7</i> </sc> Variant With Tremor Phenotype. <i>Movement Disorders Clinical Practice</i> , 0, , .	0.8	0
4285	Base editing screens map mutations affecting interferon- β signaling in cancer. <i>Cancer Cell</i> , 2023, 41, 288-303.e6.	7.7	14
4287	Biallelic Variants in TULP1 Are Associated with Heterogeneous Phenotypes of Retinal Dystrophy. <i>International Journal of Molecular Sciences</i> , 2023, 24, 2709.	1.8	1
4288	DSP-Related Cardiomyopathy as a Distinct Clinical Entity? Emerging Evidence from an Italian Cohort. <i>International Journal of Molecular Sciences</i> , 2023, 24, 2490.	1.8	8
4289	The Spectrum of MORC2-Related Disorders: A Potential Link to Cockayne Syndrome. <i>Pediatric Neurology</i> , 2023, 141, 79-86.	1.0	3
4290	Benefits, harms, and costs of newborn genetic screening for hypertrophic cardiomyopathy: Estimates from the PreEMPT model. <i>Genetics in Medicine</i> , 2023, 25, 100797.	1.1	1
4291	A Comprehensive Analysis of Cutaneous Melanoma Patients in Greece Based on Multi-Omic Data. <i>Cancers</i> , 2023, 15, 815.	1.7	0
4292	Heritable defects in telomere and mitotic function selectively predispose to sarcomas. <i>Science</i> , 2023, 379, 253-260.	6.0	13
4293	Not to Miss: Intronic Variants, Treatment, and Review of the Phenotypic Spectrum in VPS13D-Related Disorder. <i>International Journal of Molecular Sciences</i> , 2023, 24, 1874.	1.8	5
4294	Estimated Prevalence and Clinical Manifestations of <i>UBA1</i> Variants Associated With VEXAS Syndrome in a Clinical Population. <i>JAMA - Journal of the American Medical Association</i> , 2023, 329, 318.	3.8	69
4295	Why do humans need thrombospondin-1?. <i>Journal of Cell Communication and Signaling</i> , 2023, 17, 485-493.	1.8	4
4296	<i>PPM1K</i> defects cause mild maple syrup urine disease: The second case in the literature. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 1360-1365.	0.7	1

#	ARTICLE	IF	CITATIONS
4297	The association between a genetic variant in the SULF2 gene, metabolic parameters and vascular disease in patients at high cardiovascular risk. <i>Cardiovascular Endocrinology and Metabolism</i> , 2023, 12, e0278.	0.5	1
4298	Infancy-onset diabetes caused by de-regulated <sc>AMPylation</sc> of the human endoplasmic reticulum chaperone <sc>BiP</sc>. <i>EMBO Molecular Medicine</i> , 2023, 15, .	3.3	4
4300	Molecular Genetic Characteristics of FANCI, a Proposed New Ovarian Cancer Predisposing Gene. <i>Genes</i> , 2023, 14, 277.	1.0	3
4301	Pseudocoloboma-like maculopathy with biallelic <i>RDH12</i> missense mutations. <i>Journal of Medical Genetics</i> , 2023, 60, 859-865.	1.5	1
4302	Protein interaction studies in human induced neurons indicate convergent biology underlying autism spectrum disorders. <i>Cell Genomics</i> , 2023, 3, 100250.	3.0	12
4303	TIVAN-indel: a computational framework for annotating and predicting non-coding regulatory small insertions and deletions. <i>Bioinformatics</i> , 2023, 39, .	1.8	2
4304	Utilizing Large Functional and Population Genomics Resources for CRISPR/Cas Perturbation Experiment Design. <i>Methods in Molecular Biology</i> , 2023, , 63-73.	0.4	0
4305	LoFTK: a framework for fully automated calculation of predicted Loss-of-Function variants and genes. <i>BioData Mining</i> , 2023, 16, .	2.2	1
4306	Computer-Assisted Interpretation of Cancer-Predisposing Variants. , 2023, , 117-129.		0
4307	Integrative genetic and single cell RNA sequencing analysis provides new clues to the amyotrophic lateral sclerosis neurodegeneration. <i>Frontiers in Neuroscience</i> , 0, 17, .	1.4	2
4308	Impact of Copy Number Variants and Polygenic Risk Scores on Psychopathology in the UK Biobank. <i>Biological Psychiatry</i> , 2023, 94, 591-600.	0.7	3
4309	MeCP2 regulates Gdf11, a dosage-sensitive gene critical for neurological function. <i>ELife</i> , 0, 12, .	2.8	4
4310	SLC26A1 is a major determinant of sulfate homeostasis in humans. <i>Journal of Clinical Investigation</i> , 2023, 133, .	3.9	5
4312	Alternative polyadenylation alters protein dosage by switching between intronic and 3' UTR sites. <i>Science Advances</i> , 2023, 9, .	4.7	12
4315	Altered striatal actin dynamics drives behavioral inflexibility in a mouse model of fragile X syndrome. <i>Neuron</i> , 2023, 111, 1760-1775.e8.	3.8	6
4317	Venous malformation may be a feature of EXT1-related hereditary multiple exostoses: A report of two unrelated probands. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	1
4318	dbCNV: deleteriousness-based model to predict pathogenicity of copy number variations. <i>BMC Genomics</i> , 2023, 24, .	1.2	1
4319	The conserved histone chaperone Spt6 is strongly required for DNA replication and genome stability. <i>Cell Reports</i> , 2023, 42, 112264.	2.9	4

#	ARTICLE	IF	CITATIONS
4320	Rare variant analyses in large-scale cohorts identified SLC13A1 associated with chronic pain. <i>Pain</i> , 2023, 164, 1841-1851.	2.0	3
4323	A Palindrome-Like Structure on 16p13.3 Is Associated with the Formation of Complex Structural Variations and SRRM2 Haploinsufficiency. <i>Human Mutation</i> , 2023, 2023, 1-9.	1.1	1
4324	Identifying individuals at extreme risk of venous thromboembolism using polygenic risk scores. <i>Nature Genetics</i> , 2023, 55, 358-360.	9.4	0
4325	Clinically relevant combined effect of polygenic background, rare pathogenic germline variants, and family history on colorectal cancer incidence. <i>BMC Medical Genomics</i> , 2023, 16, .	0.7	9
4327	Relevance of Coding Variation in FLG And DOCK8 in Finnish Pediatric Patients with Early-Onset Moderate-To-Severe Atopic Dermatitis. <i>JID Innovations</i> , 2023, 3, 100203.	1.2	1
4329	Identifying the genetic causes of phenotypically diagnosed Pakistani mucopolysaccharidoses patients by whole genome sequencing. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	2
4330	Increased clonal hematopoiesis involving DNA damage response genes in patients undergoing lung transplantation. <i>JCI Insight</i> , 2023, 8, .	2.3	2
4331	Ultra-rare complement factor 8 coding variants in families with age-related macular degeneration. <i>IScience</i> , 2023, 26, 106417.	1.9	3
4332	Exploring genotype-phenotype correlations in glutaric aciduria type 1. <i>Journal of Inherited Metabolic Disease</i> , 2023, 46, 371-390.	1.7	8
4333	<i>PAX9</i> mutations and genetic synergism in familial tooth agenesis. <i>Annals of the New York Academy of Sciences</i> , 2023, 1524, 87-96.	1.8	2
4334	<i>Drosophila</i> Models Reveal Properties of Mutant Lamins That Give Rise to Distinct Diseases. <i>Cells</i> , 2023, 12, 1142.	1.8	0
4335	Cohort profile: SUPER-Finland – the Finnish study for hereditary mechanisms of psychotic disorders. <i>BMJ Open</i> , 2023, 13, e070710.	0.8	1
4336	Clinical, technical, and environmental biases influencing equitable access to clinical genetics/genomics testing: A points to consider statement of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2023, 25, 100812.	1.1	6
4337	Characterization of genome-wide STR variation in 6487 human genomes. <i>Nature Communications</i> , 2023, 14, .	5.8	15
4338	Pathway-driven rare germline variants associated with transplant-associated thrombotic microangiopathy (TA-TMA). <i>Thrombosis Research</i> , 2023, 225, 39-46.	0.8	0
4339	Cohort-driven variant burden analysis and pathogenicity identification in monogenic autoinflammatory disorders. <i>Journal of Allergy and Clinical Immunology</i> , 2023, 152, 517-527.	1.5	1
4340	A heterozygous missense variant in DLX3 leads to uterine leiomyomas and pregnancy losses in a consanguineous Iranian family. <i>Gene</i> , 2023, 865, 147292.	1.0	0
4341	Dissecting the 22q13 region to explore the genetic and phenotypic diversity of patients with Phelan-McDermid syndrome. <i>European Journal of Medical Genetics</i> , 2023, 66, 104732.	0.7	10

#	ARTICLE	IF	CITATIONS
4342	Noninvasive genomic profiling of somatic mutations in oral cavity cancers. <i>Oral Oncology</i> , 2023, 140, 106372.	0.8	1
4343	Structural and genomic analysis of single nucleotide polymorphisms in human host factor endothelial protein C receptor (EPCR) reveals complex interplay with malaria parasites. <i>Infection, Genetics and Evolution</i> , 2023, 110, 105413.	1.0	3
4344	Identification of pathogenic GCK variants in patients with common type 2 diabetes can lead to discontinuation of pharmacological treatment. <i>Molecular Genetics and Metabolism Reports</i> , 2023, 35, 100972.	0.4	0
4345	A novel FLNC variation associated with restrictive cardiomyopathy with an unusually long clinical course – A case report. <i>Gene Reports</i> , 2023, 31, 101769.	0.4	0
4346	Genetics of Chronic Kidney Disease in Low-Resource Settings. <i>Seminars in Nephrology</i> , 2022, 42, 151314.	0.6	0
4347	Pathogenic variants in the <i>SPTLC1</i> gene cause hyperkeratosis lenticularis perstans. <i>British Journal of Dermatology</i> , 2023, 188, 94-99.	1.4	3
4349	Population Frequency of Undiagnosed Fabry Disease in the General Population. <i>Kidney International Reports</i> , 2023, 8, 1373-1379.	0.4	1
4350	A deep intronic TCTN2 variant activating a cryptic exon predicted by SpliceRover in a patient with Joubert syndrome. <i>Journal of Human Genetics</i> , 2023, 68, 499-505.	1.1	2
4352	Explanations for the discrepancy between variant frequency and homozygous disease occurrence: Lessons from Ashkenazi Jewish data. <i>European Journal of Medical Genetics</i> , 2023, 66, 104765.	0.7	0
4355	Functional interpretation, cataloging, and analysis of 1,341 glucose-6-phosphate dehydrogenase variants. <i>American Journal of Human Genetics</i> , 2023, 110, 228-239.	2.6	6
4356	Biallelic pathogenic variants in the mitochondrial form of phosphoenolpyruvate carboxykinase cause peripheral neuropathy. <i>Human Genetics and Genomics Advances</i> , 2023, 4, 100182.	1.0	0
4358	Inflated expectations: Rare-variant association analysis using public controls. <i>PLoS ONE</i> , 2023, 18, e0280951.	1.1	2
4361	aRgus: Multilevel visualization of non-synonymous single nucleotide variants & advanced pathogenicity score modeling for genetic vulnerability assessment. <i>Computational and Structural Biotechnology Journal</i> , 2023, 21, 1077-1083.	1.9	7
4363	A molecular phenotypic map of malignant pleural mesothelioma. <i>GigaScience</i> , 2022, 12, .	3.3	4
4364	Germline rare deleterious variant load alters cancer risk, age of onset and tumor characteristics. <i>Npj Precision Oncology</i> , 2023, 7, .	2.3	1
4365	Clinical and functional characterisation of a recurrent KCNQ1 variant in the Belgian population. <i>Orphanet Journal of Rare Diseases</i> , 2023, 18, .	1.2	1
4366	Chromosomal Aberrations Accumulate during Metastasis of Virus-Negative Merkel Cell Carcinoma. <i>Journal of Investigative Dermatology</i> , 2023, 143, 1168-1177.e2.	0.3	1
4367	Differential haplotype expression in class I MHC genes during SARS-CoV-2 infection of human lung cell lines. <i>Frontiers in Immunology</i> , 0, 13, .	2.2	3

#	ARTICLE	IF	CITATIONS
4368	The in-silico evaluation of important GLUT9 residue for uric acid transport based on renal hypouricemia type 2. <i>Chemico-Biological Interactions</i> , 2023, 373, 110378.	1.7	1
4369	Association of Rare Protein-Truncating DNA Variants in <i>APOB</i> or <i>PCSK9</i> With Low-density Lipoprotein Cholesterol Level and Risk of Coronary Heart Disease. <i>JAMA Cardiology</i> , 2023, 8, 258.	3.0	10
4371	Human IRF1 governs macrophagic IFN- β immunity to mycobacteria. <i>Cell</i> , 2023, 186, 621-645.e33.	13.5	25
4372	North and East African mitochondrial genetic variation needs further characterization towards precision medicine. <i>Journal of Advanced Research</i> , 2023, , .	4.4	0
4373	Landscape of pathogenic mutations in premature ovarian insufficiency. <i>Nature Medicine</i> , 2023, 29, 483-492.	15.2	34
4374	Discovery of a Missense Mutation (Q222K) of the APOE Gene from the Australian Imaging, Biomarker and Lifestyle Study. <i>Journal of Alzheimer's Disease Reports</i> , 2023, 7, 165-172.	1.2	0
4375	Genome-wide association study reveals <i>BET1L</i> associated with survival time in the 137,693 Japanese individuals. <i>Communications Biology</i> , 2023, 6, .	2.0	0
4376	Case Report: Prenatal Recurrent Microcephaly and Corpus Callosum Abnormalities in a Chinese Family with Novel Biallelic <i>SASS6</i> Mutations. <i>Fetal Diagnosis and Therapy</i> , 2023, 50, 84-91.	0.6	1
4377	<i>UNC13A</i> in amyotrophic lateral sclerosis: from genetic association to therapeutic target. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2023, 94, 649-656.	0.9	7
4378	Genetic scores for predicting longevity in the Croatian oldest-old population. <i>PLoS ONE</i> , 2023, 18, e0279971.	1.1	3
4382	Spatially constrained gene regulation identifies key genetic contributions of preeclampsia, hypertension, and proteinuria. <i>Biology of Reproduction</i> , 2023, 108, 659-670.	1.2	2
4383	Dystrophin (DMD) Missense Variant in Cats with Becker-Type Muscular Dystrophy. <i>International Journal of Molecular Sciences</i> , 2023, 24, 3192.	1.8	2
4384	WGS Data Collections: How Do Genomic Databases Transform Medicine?. <i>International Journal of Molecular Sciences</i> , 2023, 24, 3031.	1.8	2
4385	Spinal cord extracts of amyotrophic lateral sclerosis spread TDP-43 pathology in cerebral organoids. <i>PLoS Genetics</i> , 2023, 19, e1010606.	1.5	15
4387	Homologous recombination inquiry through ovarian malignancy investigations: JGOG3025 Study. <i>Cancer Science</i> , 2023, 114, 2515-2523.	1.7	3
4388	Mutation Screening of <i>MED27</i> in a Large Dystonia Cohort. <i>Acta Neurologica Scandinavica</i> , 2023, 2023, 1-6.	1.0	0
4389	Rare variant aggregation in 148,508 exomes identifies genes associated with proxy dementia. <i>Scientific Reports</i> , 2023, 13, .	1.6	3
4392	Using species richness calculations to model the global profile of unsampled pathogenic variants: Examples from <i>BRCA1</i> and <i>BRCA2</i> . <i>PLoS ONE</i> , 2023, 18, e0278010.	1.1	1

#	ARTICLE	IF	CITATIONS
4393	Type I Interferonopathy due to a Homozygous Loss-of-Inhibitory Function Mutation in STAT2. <i>Journal of Clinical Immunology</i> , 2023, 43, 808-818.	2.0	1
4394	Phenotypic and genotypic landscape of PROKR2 in neuroendocrine disorders. <i>Frontiers in Endocrinology</i> , 0, 14, .	1.5	4
4395	Modeling Reduced Contractility and Stiffness Using iPSC-Derived Cardiomyocytes Generated From Female Becker Muscular Dystrophy Carrier. <i>JACC Basic To Translational Science</i> , 2023, 8, 599-613.	1.9	1
4396	Polygenic architecture of rare coding variation across 394,783 exomes. <i>Nature</i> , 2023, 614, 492-499.	13.7	46
4398	The human inactive X chromosome modulates expression of the active X chromosome. <i>Cell Genomics</i> , 2023, 3, 100259.	3.0	20
4399	A method for an unbiased estimate of cross-ancestry genetic correlation using individual-level data. <i>Nature Communications</i> , 2023, 14, .	5.8	4
4400	Marked intrafamilial variability of clinical and neuroimaging manifestations in <i>NFIB</i> -related developmental disorder. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 1395-1400.	0.7	0
4401	The cancer-risk variant frequency among Polish population reported by the first national whole-genome sequencing study. <i>Frontiers in Oncology</i> , 0, 13, .	1.3	0
4402	Systemic artery to pulmonary artery aneurysm malformations associated with variants at <i>MCF2L</i> . <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 1250-1260.	0.7	0
4403	Telomere Length as a New Risk Marker of Early-Onset Colorectal Cancer. <i>International Journal of Molecular Sciences</i> , 2023, 24, 3526.	1.8	1
4404	Spatial genomic diversity associated with <i>APOBEC</i> mutagenesis in squamous cell carcinoma arising from ovarian teratoma. <i>Cancer Science</i> , 2023, 114, 2145-2157.	1.7	4
4406	De novo intronic GATA1 mutation leads to diamond-blackfan anemia like disease. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	0
4407	AIP gene germline variants in adult Polish patients with apparently sporadic pituitary macroadenomas. <i>Frontiers in Endocrinology</i> , 0, 14, .	1.5	2
4408	Fumarate Hydratase Variants and their Association with Paraganglioma/Pheochromocytoma. <i>Urology</i> , 2023, , .	0.5	0
4409	Inherited mutations affecting the SRCAP complex are central in moderate-penetrance predisposition to uterine leiomyomas. <i>American Journal of Human Genetics</i> , 2023, 110, 460-474.	2.6	4
4410	SpliceAI-visual: a free online tool to improve SpliceAI splicing variant interpretation. <i>Human Genomics</i> , 2023, 17, .	1.4	15
4412	Systematic Assessment of Protein C-Termini Mutated in Human Disorders. <i>Biomolecules</i> , 2023, 13, 355.	1.8	0
4413	CJB4 variants linked to skin disease exhibit a trafficking deficiency en route to gap junction formation that can be restored by co-expression of select connexins. <i>Frontiers in Cell and Developmental Biology</i> , 0, 11, .	1.8	1

#	ARTICLE	IF	CITATIONS
4414	The first genetic landscape of inherited retinal dystrophies in Portuguese patients identifies recurrent homozygous mutations as a frequent cause of pathogenesis. , 2023, 2, .		4
4415	Sensitive Detection of Cell-Free Tumour DNA Using Optimised Targeted Sequencing Can Predict Prognosis in Gastro-Oesophageal Cancer. <i>Cancers</i> , 2023, 15, 1160.	1.7	6
4416	Genomic and Transcriptomic Characteristics of Metastatic Thyroid Cancers with Exceptional Responses to Radioactive Iodine Therapy. <i>Clinical Cancer Research</i> , 2023, 29, 1620-1630.	3.2	10
4417	Functional Characterization of Six <i>SLCO1B1</i> (<i>OATP1B1</i>) Variants Observed in Finnish Individuals with a Psychotic Disorder. <i>Molecular Pharmaceutics</i> , 2023, 20, 1500-1508.	2.3	1
4418	PipeIT2: A tumor-only somatic variant calling workflow for molecular diagnostic Ion Torrent sequencing data. <i>Genomics</i> , 2023, 115, 110587.	1.3	1
4419	Ultra-low-coverage genome-wide association studyâ€”insights into gestational age using 17,844 embryo samples with preimplantation genetic testing. <i>Genome Medicine</i> , 2023, 15, .	3.6	4
4421	Atypical ATMs: Broadening the phenotypic spectrum of ATM-associated hereditary cancer. <i>Frontiers in Oncology</i> , 0, 13, .	1.3	1
4422	Patterns of mutations in nine cancer-related genes and PAF development among smoking male patients diagnosed with bladder cancer. <i>Tumor Biology</i> , 2023, 45, 1-14.	0.8	0
4424	Modifications of the endosomal compartment in fibroblasts from sporadic Alzheimerâ€™s disease patients are associated with cognitive impairment. <i>Translational Psychiatry</i> , 2023, 13, .	2.4	3
4426	Ultrarare Missense Variants Implicated in Utah Pedigrees Multiply Affected With Schizophrenia. <i>Biological Psychiatry Global Open Science</i> , 2023, 3, 797-802.	1.0	0
4427	Clinical and Histopathologic Characteristics and Template of the TGFBI p.(His626Arg) Missense Variant Lattice Corneal Dystrophy. <i>Cornea</i> , 2023, 42, 1124-1132.	0.9	1
4429	An In-Depth Single-Gene Worldwide Carrier Frequency and Genetic Prevalence Analysis of <i>CYP4V2</i> as the Cause of Bietti Crystalline Dystrophy. <i>Translational Vision Science and Technology</i> , 2023, 12, 27.	1.1	2
4430	Genomic and Glycolytic Entropy Are Reliable Radiogenomic Heterogeneity Biomarkers for Non-Small Cell Lung Cancer. <i>International Journal of Molecular Sciences</i> , 2023, 24, 3988.	1.8	2
4431	Molecular Dynamic Simulation Analysis of a Novel Missense Variant in <i>CYB5R3</i> Gene in Patients with Methemoglobinemia. <i>Medicina (Lithuania)</i> , 2023, 59, 379.	0.8	5
4432	Functional characterization of all missense variants in <i>LEPR</i> , <i>PCSK1</i> , and <i>POMC</i> genes arising from single-nucleotide variants. <i>Expert Review of Endocrinology and Metabolism</i> , 2023, 18, 209-219.	1.2	1
4433	Whole-genome sequencing analysis in families with recurrent pregnancy loss: A pilot study. <i>PLoS ONE</i> , 2023, 18, e0281934.	1.1	3
4434	Pathogenic gene variants in <i>CCDC39</i> , <i>CCDC40</i> , <i>RSPH1</i> , <i>RSPH9</i> , <i>HYDIN</i> , and <i>SPEF2</i> cause defects of sperm flagella composition and male infertility. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	2
4435	Variants in <i>DTNA</i> cause a mild, dominantly inherited muscular dystrophy. <i>Acta Neuropathologica</i> , 2023, 145, 479-496.	3.9	0

#	ARTICLE	IF	CITATIONS
4437	The most common European HINT1 neuropathy variant phenotype and its case studies. <i>Frontiers in Neurology</i> , 0, 14, .	1.1	1
4438	<i>C9orf72</i> expansions are the most common cause of genetic frontotemporal dementia in a Southeast Asian cohort. <i>Annals of Clinical and Translational Neurology</i> , 2023, 10, 568-578.	1.7	0
4439	Unexpected frequency of the pathogenic <i>AR</i> CAG repeat expansion in the general population. <i>Brain</i> , 2023, 146, 2723-2729.	3.7	6
4442	KidneyNetwork: using kidney-derived gene expression data to predict and prioritize novel genes involved in kidney disease. <i>European Journal of Human Genetics</i> , 2023, 31, 1300-1308.	1.4	4
4443	Multivariate genomic architecture of cortical thickness and surface area at multiple levels of analysis. <i>Nature Communications</i> , 2023, 14, .	5.8	11
4444	Genetic Interaction of tRNA-Dependent Mistranslation with Fused in Sarcoma Protein Aggregates. <i>Genes</i> , 2023, 14, 518.	1.0	0
4446	A common genetic variation in GZMB may associate with cancer risk in patients with Lynch syndrome. <i>Frontiers in Oncology</i> , 0, 13, .	1.3	0
4447	Mendelian inheritance revisited: dominance and recessiveness in medical genetics. <i>Nature Reviews Genetics</i> , 2023, 24, 442-463.	7.7	16
4448	Association of African Ancestry-Specific <i>APOE</i> Missense Variant R145C With Risk of Alzheimer Disease. <i>JAMA - Journal of the American Medical Association</i> , 2023, 329, 551.	3.8	12
4449	A Comprehensive Genetic Analysis of Slovenian Families with Multiple Cases of Orofacial Clefts Reveals Novel Variants in the Genes <i>IRF6</i> , <i>GRHL3</i> , and <i>TBX22</i> . <i>International Journal of Molecular Sciences</i> , 2023, 24, 4262.	1.8	0
4450	Novel Calcium-Sensing Receptor (<i>CASR</i>) Mutation in a Family with Autosomal Dominant Hypocalcemia Type 1 (<i>ADH1</i>): Genetic Study over Three Generations and Clinical Characteristics. <i>Hormone Research in Paediatrics</i> , 2023, 96, 473-482.	0.8	1
4451	Head Size in Phelan-McDermid Syndrome: A Literature Review and Pooled Analysis of 198 Patients Identifies Candidate Genes on 22q13. <i>Genes</i> , 2023, 14, 540.	1.0	0
4452	<i>GABRG2</i> Variants Associated with Febrile Seizures. <i>Biomolecules</i> , 2023, 13, 414.	1.8	4
4453	Multi-ancestry genome-wide association study of 4069 children with glioma identifies 9p21.3 risk locus. <i>Neuro-Oncology</i> , 2023, 25, 1709-1720.	0.6	2
4454	De novo variants in <i>MAST4</i> related to neurodevelopmental disorders with developmental delay and infantile spasms: Genotype-phenotype association. <i>Frontiers in Molecular Neuroscience</i> , 0, 16, .	1.4	4
4461	Rare and Common Variants in <i>GALNT3</i> May Affect Bone Mass Independently of Phosphate Metabolism. <i>Journal of Bone and Mineral Research</i> , 2020, 38, 678-691.	3.1	0
4462	Loss of function of <i>ADNP</i> by an intragenic inversion. <i>European Journal of Human Genetics</i> , 2023, 31, 967-970.	1.4	4
4465	Genome-wide association study and identification of systemic comorbidities in development of age-related macular degeneration in a hospital-based cohort of Han Chinese. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	1

#	ARTICLE	IF	CITATIONS
4467	Probabilistic Mixture Models Improve Calibration of Panel-derived Tumor Mutational Burden in the Context of both Tumor-normal and Tumor-only Sequencing. <i>Cancer Research Communications</i> , 2023, 3, 501-509.	0.7	1
4470	Genetic Heterogeneity of X-Linked Ichthyosis in the Republic of North Ossetia-Alania, Case Series Report. <i>International Journal of Molecular Sciences</i> , 2023, 24, 4515.	1.8	0
4471	New SNCA mutation and structures of α -synuclein filaments from juvenile-onset synucleinopathy. <i>Acta Neuropathologica</i> , 2023, 145, 561-572.	3.9	8
4472	The evolution of the human DNA replication timing program. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2023, 120, .	3.3	2
4473	Computational and mitochondrial functional studies of novel compound heterozygous variants in SPATA5 gene support a causal link with epileptogenic encephalopathy. <i>Human Genomics</i> , 2023, 17, .	1.4	1
4474	The prevalence of pharmacogenetic variants of vitamin K epoxide reductase complex subunit 1 gene (rs9923231), cytochrome P450 family 2 subfamily C member 9 gene (rs1799853) and cytochrome P450 family 3 subfamily-A member-5 gene (rs776746) among 13 ethnic groups of Pakistan. <i>Molecular Biology Reports</i> , 0, .	1.0	0
4475	Chung's Jansen syndrome can mimic Cornelia de Lange syndrome: Another player among chromatinopathies?. <i>American Journal of Medical Genetics, Part A</i> , 0, .	0.7	0
4476	Nicotinamide riboside supplementation is not associated with altered methylation homeostasis in Parkinson's disease. <i>IScience</i> , 2023, 26, 106278.	1.9	4
4477	Unified views on variant impact across many diseases. <i>Trends in Genetics</i> , 2023, 39, 442-450.	2.9	3
4478	Genomic Strategies in Mitochondrial Diagnostics. <i>Methods in Molecular Biology</i> , 2023, , 397-425.	0.4	0
4479	Global distribution of functionally important CYP2C9 alleles and their inferred metabolic consequences. <i>Human Genomics</i> , 2023, 17, .	1.4	11
4481	Germline Variants in Childhood Cutaneous Melanoma. <i>Journal of Investigative Dermatology</i> , 2023, 143, 1610-1613.	0.3	0
4485	Characterizing the genotypic spectrum of retinitis pigmentosa in East Asian populations: a systematic review. <i>Ophthalmic Genetics</i> , 2023, 44, 109-118.	0.5	1
4486	Connective tissue presentation in two families expands the phenotypic spectrum of <i>PYROXD1</i> disorders. <i>Human Molecular Genetics</i> , 0, .	1.4	0
4488	Whole-genome sequencing reveals a complex African population demographic history and signatures of local adaptation. <i>Cell</i> , 2023, 186, 923-939.e14.	13.5	34
4489	The structure, function, and pharmacology of MRGPRs. <i>Trends in Pharmacological Sciences</i> , 2023, 44, 237-251.	4.0	7
4490	Structural and signaling proteins in the Z-disk and their role in cardiomyopathies. <i>Frontiers in Physiology</i> , 0, 14, .	1.3	5
4491	Association of rs9939609 in FTO with BMI among Polynesian peoples living in Aotearoa New Zealand and other Pacific nations. <i>Journal of Human Genetics</i> , 0, .	1.1	0

#	ARTICLE	IF	CITATIONS
4492	Congenital hydrocephalus: new Mendelian mutations and evidence for oligogenic inheritance. <i>Human Genomics</i> , 2023, 17, .	1.4	4
4493	Evaluation of in silico pathogenicity prediction tools for the classification of small in-frame indels. <i>BMC Medical Genomics</i> , 2023, 16, .	0.7	1
4494	Genetics of psychotic disorders with focus on early-onset psychosis. , 2023, , 51-80.		0
4496	De novo mutations disturb early brain development more frequently than common variants in schizophrenia. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2023, 192, 62-70.	1.1	0
4497	PRRC2 proteins impact translation initiation by promoting leaky scanning. <i>Nucleic Acids Research</i> , 2023, 51, 3391-3409.	6.5	6
4498	<i>DIS3</i> Variants are Associated With Primary Ovarian Insufficiency: Importance of Transcription/Translation in Oogenesis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 0, , .	1.8	0
4500	RNF130 Regulates LDLR Availability and Plasma LDL Cholesterol Levels. <i>Circulation Research</i> , 2023, 132, 849-863.	2.0	2
4502	Multiomic analyses implicate a neurodevelopmental program in the pathogenesis of cerebral arachnoid cysts. <i>Nature Medicine</i> , 2023, 29, 667-678.	15.2	5
4503	Genome-wide significant risk loci for mood disorders in the Old Order Amish founder population. <i>Molecular Psychiatry</i> , 0, , .	4.1	3
4505	Somatic mutation landscape in a cohort of meningiomas that have undergone grade progression. <i>BMC Cancer</i> , 2023, 23, .	1.1	0
4507	Insights from Spatial Measures of Intolerance to Identifying Pathogenic Variants in Developmental and Epileptic Encephalopathies. <i>International Journal of Molecular Sciences</i> , 2023, 24, 5114.	1.8	0
4509	The Severity of Congenital Hypothyroidism With Gland-In-Situ Predicts Molecular Yield by Targeted Next-Generation Sequencing. <i>Journal of Clinical Endocrinology and Metabolism</i> , 0, , .	1.8	0
4510	Identification of Genetic Alterations in Rapid Progressive Glioblastoma by Use of Whole Exome Sequencing. <i>Diagnostics</i> , 2023, 13, 1017.	1.3	3
4511	Genetic analyses of DNA repair pathway associated genes implicate new candidate cancer predisposing genes in ancestrally defined ovarian cancer cases. <i>Frontiers in Oncology</i> , 0, 13, .	1.3	1
4514	CHEK2 Alterations in Pediatric Malignancy: A Single-Institution Experience. <i>Cancers</i> , 2023, 15, 1649.	1.7	2
4515	Association of HLA-class II alleles with risk of relapse in myeloperoxidase-antineutrophil cytoplasmic antibody positive vasculitis in the Japanese population. <i>Frontiers in Immunology</i> , 0, 14, .	2.2	1
4516	Pan-cancer association of DNA repair deficiencies with whole-genome mutational patterns. <i>ELife</i> , 0, 12, .	2.8	0
4518	Comprehensive laboratory diagnosis of Fanconi anaemia: comparison of cellular and molecular analysis. <i>Journal of Medical Genetics</i> , 2023, 60, 801-809.	1.5	1

#	ARTICLE	IF	CITATIONS
4519	In Silico-Based Structural Evaluation to Categorize the Pathogenicity of Mutations Identified in the RAD Class of Proteins. <i>ACS Omega</i> , 2023, 8, 10266-10277.	1.6	0
4520	A De Novo Sequence Variant in Barrier-to-Autointegration Factor Is Associated with Dominant Motor Neuronopathy. <i>Cells</i> , 2023, 12, 847.	1.8	0
4521	Convergent coexpression of autism-associated genes suggests some novel risk genes may not be detectable in large-scale genetic studies. <i>Cell Genomics</i> , 2023, 3, 100277.	3.0	2
4523	Imputation-powered whole-exome analysis identifies genes associated with kidney function and disease in the UK Biobank. <i>Nature Communications</i> , 2023, 14, .	5.8	3
4524	A crowdsourcing database for the copy-number variation of the Spanish population. <i>Human Genomics</i> , 2023, 17, .	1.4	2
4525	Al-Gazali Skeletal Dysplasia Constitutes the Lethal End of ADAMTSL2-Related Disorders. <i>Journal of Bone and Mineral Research</i> , 2020, 38, 692-706.	3.1	3
4527	A cryptic pathogenic <i>NDUFV1</i> variant identified by RNA-seq in a patient with normal complex I activity in muscle and transient magnetic resonance imaging changes. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 1599-1606.	0.7	2
4529	Essential genes: a cross-species perspective. <i>Mammalian Genome</i> , 2023, 34, 357-363.	1.0	2
4531	Association of FAT1 with focal epilepsy and correlation between seizure relapse and gene expression stage. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2023, , .	0.9	6
4532	Developmental disorder and spastic paraparesis in two sisters with a <i>TCF7L2</i> truncating variant inherited from a mosaic mother. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 1658-1663.	0.7	0
4534	Revisiting mutagenesis at non-B DNA motifs in the human genome. <i>Nature Structural and Molecular Biology</i> , 2023, 30, 417-424.	3.6	8
4535	The Driverless Triple-Wild-Type (BRAF, RAS, KIT) Cutaneous Melanoma: Whole Genome Sequencing Discoveries. <i>Cancers</i> , 2023, 15, 1712.	1.7	3
4536	dbAQP-SNP: a database of missense single-nucleotide polymorphisms in human aquaporins. <i>Database: the Journal of Biological Databases and Curation</i> , 2023, 2023, .	1.4	2
4537	vaRHC: an R package for semi-automation of variant classification in hereditary cancer genes according to ACMG/AMP and gene-specific ClinGen guidelines. <i>Bioinformatics</i> , 2023, 39, .	1.8	1
4538	Expanding the Phenotypic Spectrum of Kenny-Caffey Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2023, 108, e754-e768.	1.8	0
4540	High Performance of a Dominant/X-Linked Gene Panel in Patients with Neurodevelopmental Disorders. <i>Genes</i> , 2023, 14, 708.	1.0	3
4541	Compound Heterozygous PLD1 Variants in Right-Sided Heart Malformations. <i>Congenital Heart Disease</i> , 2023, 18, 213-218.	0.0	0
4543	Whole exome sequencing study identifies candidate loss of function variants and locus heterogeneity in familial cholesteatoma. <i>PLoS ONE</i> , 2023, 18, e0272174.	1.1	3

#	ARTICLE	IF	CITATIONS
4544	Diet-induced loss of adipose hexokinase 2 correlates with hyperglycemia. <i>ELife</i> , 0, 12, .	2.8	5
4545	Whole-genome doubling drives oncogenic loss of chromatin segregation. <i>Nature</i> , 2023, 615, 925-933.	13.7	10
4546	Mutational Spectrum of the ABCA12 Gene and Genotypeâ€“Phenotype Correlation in a Cohort of 64 Patients with Autosomal Recessive Congenital Ichthyosis. <i>Genes</i> , 2023, 14, 717.	1.0	3
4547	Genetic association analysis of 77,539 genomes reveals rare disease etiologies. <i>Nature Medicine</i> , 2023, 29, 679-688.	15.2	18
4548	Clinical case study meets population cohort: identification of a BRCA1 pathogenic founder variant in Orcadians. <i>European Journal of Human Genetics</i> , 2023, 31, 588-595.	1.4	1
4549	A Phase II Trial of Guadecitabine plus Atezolizumab in Metastatic Urothelial Carcinoma Progressing after Initial Immune Checkpoint Inhibitor Therapy. <i>Clinical Cancer Research</i> , 2023, 29, 2052-2065.	3.2	8
4550	FGF9-Associated Multiple Synostoses Syndrome Type 3 in a Multigenerational Family. <i>Genes</i> , 2023, 14, 724.	1.0	1
4551	ATP binding cassette member A3 (ABCA3): coming of age. <i>Thorax</i> , 2023, 78, 533-534.	2.7	0
4552	Molecular genetics in diagnosis of Coats disease: combination of oligogenic variants associated with different forms of hereditary retinal dystrophy. <i>Vestnik Oftalmologii</i> , 2023, 139, 69.	0.1	0
4553	Reflexive standardization and the resolution of uncertainty in the genomics clinic. <i>Social Studies of Science</i> , 2023, 53, 358-378.	1.5	3
4556	Pathogenic Variants Associated with Rare Monogenic Diseases Established in Ancient Neanderthal and Denisovan Genome-Wide Data. <i>Genes</i> , 2023, 14, 727.	1.0	0
4557	POSTRE: a tool to predict the pathological effects of human structural variants. <i>Nucleic Acids Research</i> , 2023, 51, e54-e54.	6.5	3
4558	Psychedelics: preclinical insights provide directions for future research. <i>Neuropsychopharmacology</i> , 2024, 49, 119-127.	2.8	7
4559	Mutation screening of AOPEP variants in a large dystonia cohort. <i>Journal of Neurology</i> , 2023, 270, 3225-3233.	1.8	3
4561	Pioneer factor ASCL1 cooperates with the mSWI/SNF complex at distal regulatory elements to regulate human neural differentiation. <i>Genes and Development</i> , 2023, 37, 218-242.	2.7	13
4568	The Genomics of Diabetic Neuropathy. <i>Contemporary Diabetes</i> , 2023, , 239-251.	0.0	0
4569	Variations in the poly-histidine repeat motif of HOXA1 contribute to bicuspid aortic valve in mouse and zebrafish. <i>Nature Communications</i> , 2023, 14, .	5.8	3
4571	Clinical and genetic associations of deep learning-derived cardiac magnetic resonance-based left ventricular mass. <i>Nature Communications</i> , 2023, 14, .	5.8	8

#	ARTICLE	IF	CITATIONS
4572	Exome-wide assessment of isolated biliary atresia: A report from the <sc>National Birth Defects Prevention Study</sc> using child-parent trios and a case-control design to identify novel rare variants. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 1546-1556.	0.7	2
4573	Chromatin remodeler Activity-Dependent Neuroprotective Protein (ADNP) contributes to syndromic autism. <i>Clinical Epigenetics</i> , 2023, 15, .	1.8	11
4574	Improving Hereditary Hemorrhagic Telangiectasia Molecular Diagnosis: A Referral Center Experience. <i>Genes</i> , 2023, 14, 772.	1.0	1
4576	Nasal Construction in Congenital Arhinia Due to Novel SMCHD1 Gene Variant. <i>Journal of Craniofacial Surgery</i> , 2023, 34, 849-854.	0.3	1
4577	An Assessment of Quaternary Structure Functionality in Homomer Protein Complexes. <i>Molecular Biology and Evolution</i> , 2023, 40, .	3.5	2
4578	Zonule-Associated Gene Variants in Isolated Ectopia Lentis and Glaucoma. <i>Journal of Glaucoma</i> , 2023, 32, e80-e89.	0.8	0
4579	A study of somatic <i>BRCA</i> variants and their putative effect on protein properties in malignant mesothelioma. <i>Pleura and Peritoneum</i> , 2023, 8, 19-25.	0.5	0
4580	Primate protein-ligand interfaces exhibit significant conservation and unveil human-specific evolutionary drivers. <i>PLoS Computational Biology</i> , 2023, 19, e1010966.	1.5	0
4581	Adjusting for common variant polygenic scores improves yield in rare variant association analyses. <i>Nature Genetics</i> , 2023, 55, 544-548.	9.4	9
4582	A Primer in Precision Nephrology: Optimizing Outcomes in Kidney Health and Disease via Data-Driven Medicine. <i>Kidney360</i> , 2023, Publish Ahead of Print, .	0.9	2
4583	Genetic Loss of Sucrase-Isomaltase Function: Mechanisms, Implications, and Future Perspectives. <i>The Application of Clinical Genetics</i> , 0, Volume 16, 31-39.	1.4	4
4584	Voretigene Neparvovec for the Treatment of RPE65-associated Retinal Dystrophy: Consensus and Recommendations from the Korea RPE65-IRD Consensus Paper Committee. <i>Korean Journal of Ophthalmology: KJO</i> , 2023, 37, 166-186.	0.5	1
4585	Genomic profile of two Brazilian choroid plexus tumors by whole-exome sequencing. <i>Journal of Physical Education and Sports Management</i> , 2023, 9, a006245.	0.5	0
4586	Genetic risk for Alzheimer's disease and adherence to the Mediterranean diet: results from the HELIAD study. <i>Nutritional Neuroscience</i> , 2024, 27, 289-299.	1.5	0
4587	N-Type Ca Channel in Epileptic Syndromes and Epilepsy: A Systematic Review of Its Genetic Variants. <i>International Journal of Molecular Sciences</i> , 2023, 24, 6100.	1.8	0
4588	Children with Early-Onset Psychosis Have Increased Burden of Rare GRIN2A Variants. <i>Genes</i> , 2023, 14, 779.	1.0	2
4589	Missense3D-PPI: A Web Resource to Predict the Impact of Missense Variants at Protein Interfaces Using 3D Structural Data. <i>Journal of Molecular Biology</i> , 2023, 435, 168060.	2.0	4
4591	Clinical exome sequencing findings in 1589 patients. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	0.7	0

#	ARTICLE	IF	CITATIONS
4593	A common variant in <i>AAK1</i> reduces risk of noise-induced hearing loss. <i>National Science Review</i> , 2023, 10, .	4.6	1
4594	Family-based analysis of the contribution of rare and common genetic variants to school performance in schizophrenia. <i>Molecular Psychiatry</i> , 2023, 28, 2081-2087.	4.1	6
4596	A hybrid approach to assess the structural impact of long noncoding RNA mutations uncovers key <i>NEAT1</i> interactions in colorectal cancer. <i>IUBMB Life</i> , 2023, 75, 566-579.	1.5	3
4598	Allele-dependent interaction of LRRK2 and NOD2 in leprosy. <i>PLoS Pathogens</i> , 2023, 19, e1011260.	2.1	3
4599	Genome-wide identification of tandem repeats associated with splicing variation across 49 tissues in humans. <i>Genome Research</i> , 2023, 33, 435-447.	2.4	10
4602	Recurrent mutation in the ancestry of a rare variant. <i>Genetics</i> , 2023, 224, .	1.2	4
4604	Identification of a heterogeneous and dynamic cilium during embryonic development and cell differentiation. <i>Development (Cambridge)</i> , 2023, 150, .	1.2	4
4605	CERT1 mutations perturb human development by disrupting sphingolipid homeostasis. <i>Journal of Clinical Investigation</i> , 2023, 133, .	3.9	6
4606	Single Nucleotide Polymorphism rs9277336 Controls the Nuclear Alpha Actinin 4 Human Leukocyte Antigen- β PA1 Axis and Pulmonary Endothelial Pathophenotypes in Pulmonary Arterial Hypertension. <i>Journal of the American Heart Association</i> , 2023, 12, .	1.6	1
4607	Post-identifiability in changing sociotechnological genomic data environments. <i>BioSocieties</i> , 0, , .	0.8	1
4608	Rare Catechol-O-methyltransferase Missense Variants Are Structurally Unstable Proteasome Targets. <i>Biochemistry</i> , 2023, 62, 1394-1405.	1.2	1
4609	YWHAE loss of function causes a rare neurodevelopmental disease with brain abnormalities in human and mouse. <i>Genetics in Medicine</i> , 2023, 25, 100835.	1.1	1
4610	A retrospective analysis of phosphatase catalytic subunit gene variants in patients with rare disorders identifies novel candidate neurodevelopmental disease genes. <i>Frontiers in Cell and Developmental Biology</i> , 0, 11, .	1.8	1
4611	The phenotype of the most common human <i>ADAR1</i> p150 mutation Δ ± mutation <i>P193A</i> in mice is partially penetrant. <i>EMBO Reports</i> , 2023, 24, .	2.0	8
4612	Mutations in plasticity-related-gene-1 (PRG-1) protein contribute to hippocampal seizure susceptibility and modify epileptic phenotype. <i>Cerebral Cortex</i> , 0, , .	1.6	0
4614	Clonal hematopoiesis detection in patients with cancer using cell-free DNA sequencing. <i>Science Translational Medicine</i> , 2023, 15, .	5.8	6
4616	<i>H4C5</i> missense variant leads to a neurodevelopmental phenotype overlapping with Angelman syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2023, 191, 1911-1916.	0.7	0
4618	Somatic and germline aberrations in homologous recombination repair genes in Chinese prostate cancer patients. <i>Frontiers in Oncology</i> , 0, 13, .	1.3	0

#	ARTICLE	IF	CITATIONS
4619	Rare Single Nucleotide and Copy Number Variants and the Etiology of Congenital Obstructive Uropathy: Implications for Genetic Diagnosis. <i>Journal of the American Society of Nephrology: JASN</i> , 2023, Publish Ahead of Print, .	3.0	2
4620	Whole-exome screening for primary congenital glaucoma in Lebanon. <i>Ophthalmic Genetics</i> , 2023, 44, 234-245.	0.5	1
4621	The EN-TE _x resource of multi-tissue personal epigenomes& variant-impact models. <i>Cell</i> , 2023, 186, 1493-1511.e40.	13.5	13
4623	Genetic and Molecular Quality Control of Genetically Engineered Mice. <i>Methods in Molecular Biology</i> , 2023, , 53-101.	0.4	0
4624	The Expanding Phenotypical Spectrum of WARS2-Related Disorder: Four Novel Cases with a Common Recurrent Variant. <i>Genes</i> , 2023, 14, 822.	1.0	0
4625	A Complex Intrachromosomal Rearrangement Disrupting IRF6 in a Family with Popliteal Pterygium and Van der Woude Syndromes. <i>Genes</i> , 2023, 14, 849.	1.0	0
4626	Estimating the Prevalence of LAMA2 Congenital Muscular Dystrophy using Population Genetic Databases. <i>Journal of Neuromuscular Diseases</i> , 2023, , 1-7.	1.1	1
4628	Pathogenic genomic alterations in Chinese pancreatic cancer patients and their therapeutical implications. <i>Cancer Medicine</i> , 2023, 12, 11672-11685.	1.3	5
4629	Assessment of pathogenic variation in gynecologic cancer genes in a national cohort. <i>Scientific Reports</i> , 2023, 13, .	1.6	0
4630	Novel <i>SERAC1</i> Variant Presenting With Adult-Onset Extrapyramidal Dystonia-Parkinsonism Phenotype. <i>Neurology: Genetics</i> , 2023, 9, e200067.	0.9	0
4632	A Novel Alu Element Insertion in ATM Induces Exon Skipping in Suspected HBOC Patients. <i>Human Mutation</i> , 2023, 2023, 1-10.	1.1	0
4633	Multiple Germline Events Contribute to Cancer Development in Patients with Li-Fraumeni Syndrome. <i>Cancer Research Communications</i> , 2023, 3, 738-754.	0.7	1
4634	NAGS, CPS1, and SLC25A13 (Citrin) at the Crossroads of Arginine and Pyrimidines Metabolism in Tumor Cells. <i>International Journal of Molecular Sciences</i> , 2023, 24, 6754.	1.8	1
4636	Germline (epi)genetics reveals high predisposition in females: a 5-year, nationwide, prospective Wilms tumour cohort. <i>Journal of Medical Genetics</i> , 2023, 60, 842-849.	1.5	2
4637	Leveraging Unique Chromosomal Microarray Probes to Accurately Detect Copy Number at the Highly Homologous 15q15.3 Deafness-Infertility Syndrome Locus. <i>Clinical Chemistry</i> , 0, , .	1.5	1
4638	CHCHD2 and CHCHD10-related neurodegeneration: molecular pathogenesis and the path to precision therapy. <i>Biochemical Society Transactions</i> , 2023, 51, 797-809.	1.6	5
4639	Amplification is the primary mode of gene-by-sex interaction in complex human traits. <i>Cell Genomics</i> , 2023, 3, 100297.	3.0	14
4640	Genetic variant in the <i>BRAF</i> gene compatible with Noonan spectrum disorders in an adult Fontan patient with refractory protein losing enteropathy: a follow-up report. <i>European Heart Journal - Case Reports</i> , 2023, 7, .	0.3	1

#	ARTICLE	IF	CITATIONS
4641	Studying ultra-rare variants in STX1A uncovers a novel neurodevelopmental disorder. <i>European Journal of Human Genetics</i> , 0, , .	1.4	1
4642	ABCC8-Related Monogenic Diabetes Presenting Like Type 1 Diabetes in an Adolescent. <i>ACE Clinical Case Reports</i> , 2023, 9, 101-103.	0.4	2
4643	First description of Portuguese patients with cardiac amyloidosis and p.Val142Ile: more evidence of an African variant in Caucasians. <i>Scandinavian Cardiovascular Journal</i> , 2023, 57, .	0.4	0
4644	Predicting response to enzalutamide and abiraterone in metastatic prostate cancer using whole-omics machine learning. <i>Nature Communications</i> , 2023, 14, .	5.8	4
4645	Identification of Rare Variants Involved in High Myopia Unraveled by Whole Genome Sequencing. <i>Ophthalmology Science</i> , 2023, 3, 100303.	1.0	0
4646	Identification of Compound Heterozygous EVC2 Gene Variants in Two Mexican Families with Ellis-van Creveld Syndrome. <i>Genes</i> , 2023, 14, 887.	1.0	0
4647	Tools to differentiate between Filamin C and Titin truncating variant carriers: value of MRI. <i>European Journal of Human Genetics</i> , 2023, 31, 1323-1332.	1.4	4
4649	Revisiting Genetic Epidemiology with a Refined Targeted Gene Panel for Hereditary Hearing Impairment in the Taiwanese Population. <i>Genes</i> , 2023, 14, 880.	1.0	0
4650	Analysis of glaucoma genes in Finnish patients with juvenile open-angle glaucoma. <i>Acta Ophthalmologica</i> , 2023, 101, 797-806.	0.6	0
4651	Genetic variants in genes involved in creatine biosynthesis in patients with severe obesity or anorexia nervosa. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	1
4652	Prevalence, clinical features and complications of common forms of Maturity Onset Diabetes of the Young (MODY) seen at a tertiary diabetes centre in south India. <i>Primary Care Diabetes</i> , 2023, 17, 401-407.	0.9	1
4653	LHX2 haploinsufficiency causes a variable neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2023, 25, 100839.	1.1	4
4654	De novo missense variants in RRAGC lead to a fatal mTORopathy of early childhood. <i>Genetics in Medicine</i> , 2023, 25, 100838.	1.1	0
4656	PredinID: Predicting Pathogenic Inframe Indels in Human Through Graph Convolution Neural Network With Graph Sampling Technique. <i>IEEE/ACM Transactions on Computational Biology and Bioinformatics</i> , 2023, 20, 3226-3233.	1.9	0
4657	Genetic variability in sporadic amyotrophic lateral sclerosis. <i>Brain</i> , 2023, 146, 3760-3769.	3.7	8
4658	Prenatal <i>CFAP53</i> -related laterality defect: case report and review of the literature. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2023, 36, .	0.7	0
4659	Breast cancer polygenic risk scores derived in White European populations are not calibrated for women of Ashkenazi Jewish descent. <i>Genetics in Medicine</i> , 2023, 25, 100846.	1.1	1
4660	Germline multigene panel testing of patients with endometrial cancer. <i>Oncology Letters</i> , 2023, 25, .	0.8	1

#	ARTICLE	IF	CITATIONS
4661	A Novel Variant in VPS13B Underlying Cohen Syndrome. <i>BioMed Research International</i> , 2023, 2023, 1-7.	0.9	0
4662	Bi-allelic variants in INTS11 are associated with a complex neurological disorder. <i>American Journal of Human Genetics</i> , 2023, 110, 774-789.	2.6	8
4663	Associations of minor histocompatibility antigens with outcomes following allogeneic hematopoietic cell transplantation. <i>American Journal of Hematology</i> , 0, , .	2.0	0
4664	Structure-Function of the Human WAC Protein in GABAergic Neurons: Towards an Understanding of Autosomal Dominant DeSantoâ€“Shinawi Syndrome. <i>Biology</i> , 2023, 12, 589.	1.3	0
4665	The origins and functional effects of postzygotic mutations throughout the human life span. <i>Science</i> , 2023, 380, .	6.0	6
4666	Contribution of <i>APOE</i> Genetic Variants to Dyslipidemia. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 0, , .	1.1	1
4667	High Frequency of <sc><i>LRRK2</i> R1441C</sc> Pathogenic Variant Among <sc>Palestinianâ€“Arabs</sc>. <i>Movement Disorders</i> , 2023, 38, 1362-1363.	2.2	0
4668	DrOGA: an artificial intelligence solution for driver-status prediction of genomics mutations in precision cancer medicine. <i>IEEE Access</i> , 2023, , 1-1.	2.6	0
4670	Structural and Pathogenic Impacts of ABCA4 Variants in Retinal Degenerationsâ€“An In-Silico Study. <i>International Journal of Molecular Sciences</i> , 2023, 24, 7280.	1.8	1
4671	Variant calling and benchmarking in an era of complete human genome sequences. <i>Nature Reviews Genetics</i> , 2023, 24, 464-483.	7.7	26
4672	Genetics of Inborn Errors of Immunity in highly consanguineous Middle Eastern and North African populations. <i>Seminars in Immunology</i> , 2023, 67, 101763.	2.7	1
4673	<i><sc>KCNT2</sc></i>â€“Related Disorders: Phenotypes, Functional, and Pharmacological Properties. <i>Annals of Neurology</i> , 2023, 94, 332-349.	2.8	4
4674	Comparing Genomic Landscapes of Oral and Cutaneous Squamous Cell Carcinoma of the Head and Neck: Quest for Novel Diagnostic Markers. <i>Modern Pathology</i> , 2023, 36, 100190.	2.9	1
4675	Inherited human ZNF341 deficiency. <i>Current Opinion in Immunology</i> , 2023, 82, 102326.	2.4	4
4676	Functional Analysis of a Novel, Non-Canonical RPGR Splice Variant Causing X-Linked Retinitis Pigmentosa. <i>Genes</i> , 2023, 14, 934.	1.0	1
4677	POLR1A variants underlie phenotypic heterogeneity in craniofacial, neural, and cardiac anomalies. <i>American Journal of Human Genetics</i> , 2023, 110, 809-825.	2.6	4
4679	Genetic Etiology of Nonsyndromic Hearing Loss in Hungarian Patients. <i>International Journal of Molecular Sciences</i> , 2023, 24, 7401.	1.8	1
4681	Genotype-by-environment interactions in chronic back pain. <i>Spine Journal</i> , 2023, , .	0.6	1

#	ARTICLE	IF	CITATIONS
4682	Transcriptome analyses of murine right and left maxilla-mandibular complex. <i>Orthodontics and Craniofacial Research</i> , 2023, 26, 39-47.	1.2	1
4683	Position statement of the International Society for Gastrointestinal Hereditary Tumours (InSiGHT) on APC11307K and cancer risk. <i>Journal of Medical Genetics</i> , 2023, 60, 1035-1043.	1.5	1
4684	A nomogram based on genotypic and clinicopathologic factors to predict the non-sentinel lymph node metastasis in Chinese women breast cancer patients. <i>Frontiers in Oncology</i> , 0, 13, .	1.3	0
4685	Using brain cell-type-specific protein interactomes to interpret neurodevelopmental genetic signals in schizophrenia. <i>IScience</i> , 2023, 26, 106701.	1.9	2
4687	DeMAG predicts the effects of variants in clinically actionable genes by integrating structural and evolutionary epistatic features. <i>Nature Communications</i> , 2023, 14, .	5.8	2
4688	Persistent Molecular Disease in Adult Patients With AML Evaluated With Whole-Exome and Targeted Error-Corrected DNA Sequencing. <i>JCO Precision Oncology</i> , 2023, , .	1.5	2
4689	Variants in PSMB9 and FGR differentially affect Parkinson's disease risk in GBA and LRRK2 mutation carriers. <i>Parkinsonism and Related Disorders</i> , 2023, 111, 105398.	1.1	1
4690	Mosaic variants detectable in blood extend the clinicogenetic spectrum of GLI3-related hypothalamic hamartoma. , 2023, 1, 100810.		0
4691	Forensic biogeographical ancestry inference: recent insights and current trends. <i>Genes and Genomics</i> , 2023, 45, 1229-1238.	0.5	4
4692	A Comprehensive Analysis of 21 Actionable Pharmacogenes in the Spanish Population: From Genetic Characterisation to Clinical Impact. <i>Pharmaceutics</i> , 2023, 15, 1286.	2.0	2
4693	Molecular diagnostic results of a nephropathy gene panel in patients with suspected hereditary kidney disease. <i>Laboratory Medicine</i> , 2024, 55, 13-19.	0.8	0
4694	De novo variants in CNOT9 cause a neurodevelopmental disorder with or without epilepsy. <i>Genetics in Medicine</i> , 2023, , 100859.	1.1	0
4695	Prevalence of Monogenic Bone Disorders in a Dutch Cohort of Atypical Femur Fracture Patients. <i>Journal of Bone and Mineral Research</i> , 2020, 38, 896-906.	3.1	1
4697	Classification of PRSS1 variants responsible for chronic pancreatitis: An expert perspective from the Franco-Chinese GREPAN Study Group. <i>Pancreatology</i> , 2023, 23, 491-506.	0.5	5
4698	Dominant-negative variants in CBX1 cause a neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2023, 25, 100861.	1.1	1
4699	Pharmacogenomic profile of a central European urban random population-Czech population. <i>PLoS ONE</i> , 2023, 18, e0284386.	1.1	0
4700	Rare CIDEC coding variants enriched in age-related macular degeneration patients with small low-luminance deficit cause lipid droplet and fat storage defects. <i>PLoS ONE</i> , 2023, 18, e0280484.	1.1	1
4702	Single-cell genomics meets human genetics. <i>Nature Reviews Genetics</i> , 2023, 24, 535-549.	7.7	18

#	ARTICLE	IF	CITATIONS
4703	Predicting the pathogenicity of missense variants using features derived from AlphaFold2. <i>Bioinformatics</i> , 2023, 39, .	1.8	13
4704	PRDM1 DNA-binding zinc finger domain is required for normal limb development and is disrupted in split hand/foot malformation. <i>DMM Disease Models and Mechanisms</i> , 2023, 16, .	1.2	5
4705	Systematic elucidation of genetic mechanisms underlying cholesterol uptake. <i>Cell Genomics</i> , 2023, , 100304.	3.0	2
4706	Functional analyses of rare germline BRCA1 variants by transcriptional activation and homologous recombination repair assays. <i>BMC Cancer</i> , 2023, 23, .	1.1	2
4707	eyeVarP: A computational framework for the identification of pathogenic variants specific to eye disease. <i>Genetics in Medicine</i> , 2023, 25, 100862.	1.1	2
4708	Epigenetic Alteration of H3K27me3 as a Possible Oncogenic Mechanism of Central Neurocytoma. <i>Laboratory Investigation</i> , 2023, 103, 100159.	1.7	1
4709	Encephalitis and poor neuronal deathâ€‘mediated control of herpes simplex virus in human inherited RIPK3 deficiency. <i>Science Immunology</i> , 2023, 8, .	5.6	8
4710	H2A monoubiquitination: insights from human genetics and animal models. <i>Human Genetics</i> , 0, , .	1.8	0
4741	Inborn Errors of Immunity in Hidradenitis Suppurativa Pathogenesis and Disease Burden. <i>Journal of Clinical Immunology</i> , 2023, 43, 1040-1051.	2.0	4
4764	Editorial: Pharmacogenetics and pharmacogenomics in Latin America: ethnic variability, new insights in advances and perspectives: a RELIVAF-CYTED initiative, Volume II. <i>Frontiers in Pharmacology</i> , 0, 14, .	1.6	1
4879	Case report: marfan syndrome (MFS) mimicking cutaneous vasculitis. <i>Frontiers in Pediatrics</i> , 0, 11, .	0.9	0
4897	Primary ciliary dyskinesia. , 2023, , 118-134.		1
4979	Case report: A rare variant m.4135T>C in the MT-ND1 gene leads to Leber hereditary optic neuropathy and altered respiratory chain supercomplexes. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	2
5092	EMQN: Recommendations for genetic testing in inherited cardiomyopathies and arrhythmias. <i>European Journal of Human Genetics</i> , 2023, 31, 1003-1009.	1.4	2
5097	Integrating Computational Approaches to Predict the Effect of Genetic Variants on Protein Stability in Retinal Degenerative Disease. <i>Advances in Experimental Medicine and Biology</i> , 2023, , 157-163.	0.8	1
5138	Integrating non-mammalian model organisms in the diagnosis of rare genetic diseases in humans. <i>Nature Reviews Genetics</i> , 2024, 25, 46-60.	7.7	10
5140	Tutorial: a statistical genetics guide to identifying HLA alleles driving complex disease. <i>Nature Protocols</i> , 2023, 18, 2625-2641.	5.5	5
5144	The benefits and pitfalls of machine learning for biomarker discovery. <i>Cell and Tissue Research</i> , 2023, 394, 17-31.	1.5	7

#	ARTICLE	IF	CITATIONS
5148	Case report: Birkâ€“Landauâ€“Perez syndrome linked to the SLC30A9 geneâ€“identification of additional cases and expansion of the phenotypic spectrum. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	0
5195	Novel homozygous variants in PRORP expand the genotypic spectrum of combined oxidative phosphorylation deficiency 54. <i>European Journal of Human Genetics</i> , 2023, 31, 1190-1194.	1.4	1
5301	Familial co-segregation and the emerging role of long-read sequencing to re-classify variants of uncertain significance in inherited retinal diseases. <i>Npj Genomic Medicine</i> , 2023, 8, .	1.7	1
5365	Advancing variant effect prediction using protein language models. <i>Nature Genetics</i> , 2023, 55, 1426-1427.	9.4	1
5472	Implementation of Exome Sequencing to Identify Rare Genetic Diseases. <i>Methods in Molecular Biology</i> , 2024, , 79-98.	0.4	0
5511	Whole-exome and whole-genome sequencing in the molecular diagnostic laboratory. , 2024, , 27-38.		0
5529	Computational immunogenomic approaches to predict response to cancer immunotherapies. <i>Nature Reviews Clinical Oncology</i> , 2024, 21, 28-46.	12.5	1
5546	Context-specific functions of chromatin remodellers in development and disease. <i>Nature Reviews Genetics</i> , 0, , .	7.7	3
5582	Distal hereditary motor neuronopathy as a new phenotype associated with variants in BAG3. <i>Journal of Neurology</i> , 2024, 271, 986-994.	1.8	0
5663	Estimating the proportion of nonsense variants undergoing the newly described phenomenon of manufactured splice rescue. <i>European Journal of Human Genetics</i> , 2024, 32, 238-242.	1.4	2
5694	Case Report: Sacral Neuromodulation with Suspected Neuromuscular Blockade Secondary to Butyrylcholinesterase Variant. , 2023, 1, .		1
5701	An expanded genomic database for identifying disease-related variants. <i>Nature</i> , 0, , .	13.7	0
5707	Genetic risk variants in New Yorkers of Puerto Rican and Dominican Republic heritage with Parkinsonâ€™s disease. <i>Npj Parkinson's Disease</i> , 2023, 9, .	2.5	0
5737	Comprehensive Representation ofâ€“Variation Interpretation Data viaâ€“Conceptual Modeling. <i>Lecture Notes in Computer Science</i> , 2023, , 25-34.	1.0	0
5766	Rediscovering tandem repeat variation in schizophrenia: challenges and opportunities. <i>Translational Psychiatry</i> , 2023, 13, .	2.4	0
5776	The Human GP130 Cytokine Receptor and Its Expressionâ€“an Atlas and Functional Taxonomy of Genetic Variants. <i>Journal of Clinical Immunology</i> , 2024, 44, .	2.0	1
5810	Next-generation sequencing and bioinformatics in rare movement disorders. <i>Nature Reviews Neurology</i> , 2024, 20, 114-126.	4.9	0
5822	Characterizing the pathogenicity of genetic variants: the consequences of context. <i>Npj Genomic Medicine</i> , 2024, 9, .	1.7	1

#	ARTICLE	IF	CITATIONS
5828	The expanding diagnostic toolbox for rare genetic diseases. <i>Nature Reviews Genetics</i> , 0, , .	7.7	0
5838	Heterogeneities in Hereditary Cancer Genes as Revealed by a Large-Scale Genome Analysis. , 2023, , 59-78.		0
5895	Pediatric Pancytopenia and Monosomy 7: A Case Report of SAMD9L-Associated Disease. <i>Journal of Clinical Immunology</i> , 2024, 44, .	2.0	0
5901	Prediction of Non-coding Driver Mutations Using Ensemble Learning. , 2023, , .		0
5948	Case report: Identification of a novel variant p.Gly215Arg in the CHN1 gene causing Moebius syndrome. <i>Frontiers in Genetics</i> , 0, 15, .	1.1	0
6008	Genetics and etiology of congenital heart disease. <i>Current Topics in Developmental Biology</i> , 2024, , 297-331.	1.0	0
6012	Sequencing and characterizing short tandem repeats in the human genome. <i>Nature Reviews Genetics</i> , 0, , .	7.7	0
6133	Dissecting the Immune System through Gene Regulation. <i>Advances in Experimental Medicine and Biology</i> , 2024, , 219-235.	0.8	0