Eric W Klee

List of Publications by Year
in descending order

158 papers	5,104 citations	117453 34 h-index	118652 62 g-index
164 all docs	164 docs citations	164 times ranked	10279 citing authors

#	Article	IF	Citations
1	Standards and Guidelines for Validating Next-Generation Sequencing Bioinformatics Pipelines. Journal of Molecular Diagnostics, 2018, 20, 4-27.	1.2	341
2	Integrated Genomic Characterization Reveals Novel, Therapeutically Relevant Drug Targets in FGFR and EGFR Pathways in Sporadic Intrahepatic Cholangiocarcinoma. PLoS Genetics, 2014, 10, e1004135.	1.5	292
3	Preemptive Genotyping for Personalized Medicine: Design of the Right Drug, Right Dose, Right Timeâ€"Using Genomic Data to Individualize Treatment Protocol. Mayo Clinic Proceedings, 2014, 89, 25-33.	1.4	250
4	Preemptive Pharmacogenomic Testing for Precision Medicine. Journal of Molecular Diagnostics, 2016, 18, 438-445.	1.2	171
5	Genome-wide detection of tandem DNA repeats that are expanded in autism. Nature, 2020, 586, 80-86.	13.7	155
6	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	5.8	150
7	Adrenomedullin is Up-regulated in Patients With Pancreatic Cancer and Causes Insulin Resistance in Î² Cells and Mice. Gastroenterology, 2012, 143, 1510-1517.e1.	0.6	145
8	Sentieon DNASeq Variant Calling Workflow Demonstrates Strong Computational Performance and Accuracy. Frontiers in Genetics, 2019, 10, 736.	1.1	131
9	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	3.8	121
10	Clinical spectrum and genotypeâ€"phenotype associations of KCNA2-related encephalopathies. Brain, 2017, 140, 2337-2354.	3.7	117
11	Bioinformatics for Clinical Next Generation Sequencing. Clinical Chemistry, 2015, 61, 124-135.	1.5	114
12	Disruption of the ATXN1â€"CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. Nature Genetics, 2017, 49, 527-536.	9.4	113
13	Confirming Variants in Next-Generation Sequencing Panel Testing by Sanger Sequencing. Journal of Molecular Diagnostics, 2015, 17, 456-461.	1.2	109
14	Bi-allelic Alterations in AEBP1 Lead to Defective Collagen Assembly and Connective Tissue Structure Resulting in a Variant of Ehlers-Danlos Syndrome. American Journal of Human Genetics, 2018, 102, 696-705.	2.6	105
15	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	2.6	87
16	Outcome of Whole Exome Sequencing for Diagnostic Odyssey Cases of an Individualized Medicine Clinic. Mayo Clinic Proceedings, 2016, 91, 297-307.	1.4	83
17	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	3.9	68
18	Evaluating eukaryotic secreted protein prediction. BMC Bioinformatics, 2005, 6, 256.	1.2	67

#	Article	IF	Citations
19	Genome-Wide Reverse Genetics Framework to Identify Novel Functions of the Vertebrate Secretome. PLoS ONE, 2006, 1, e104.	1.1	67
20	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. Npj Genomic Medicine, 2020, 5, 47.	1.7	67
21	Arterial tortuosity syndrome: 40 new families and literature review. Genetics in Medicine, 2018, 20, 1236-1245.	1.1	66
22	Zebrafish for the Study of the Biological Effects of Nicotine. Nicotine and Tobacco Research, 2011, 13, 301-312.	1.4	61
23	Candidate Serum Biomarkers for Prostate Adenocarcinoma Identified by mRNA Differences in Prostate Tissue and Verified with Protein Measurements in Tissue and Blood. Clinical Chemistry, 2012, 58, 599-609.	1.5	61
24	TREAT: a bioinformatics tool for variant annotations and visualizations in targeted and exome sequencing data. Bioinformatics, 2012, 28, 277-278.	1.8	59
25	Implementing individualized medicine into the medical practice. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 15-23.	0.7	58
26	Experience with precision genomics and tumor board, indicates frequent target identification, but barriers to delivery. Oncotarget, 2017, 8, 27145-27154.	0.8	55
27	RNA sequencing shows transcriptomic changes in rectosigmoid mucosa in patients with irritable bowel syndrome-diarrhea: a pilot case-control study. American Journal of Physiology - Renal Physiology, 2014, 306, G1089-G1098.	1.6	52
28	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	1.1	48
29	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
30	Larval Zebrafish Model for FDA-Approved Drug Repositioning for Tobacco Dependence Treatment. PLoS ONE, 2014, 9, e90467.	1.1	48
31	Expanding DNA diagnostic panel testing: is more better?. Expert Review of Molecular Diagnostics, 2011, 11, 703-709.	1.5	46
32	Identifying secretomes in people, pufferfish and pigs. Nucleic Acids Research, 2004, 32, 1414-1421.	6.5	44
33	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	9.4	44
34	De novo DDX3X missense variants in males appear viable and contribute to syndromic intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 570-578.	0.7	42
35	Zebrafish approaches enhance the translational research tackle box. Translational Research, 2014, 163, 65-78.	2.2	40
36	RINT1 Bi-allelic Variations Cause Infantile-Onset Recurrent Acute Liver Failure and Skeletal Abnormalities. American Journal of Human Genetics, 2019, 105, 108-121.	2.6	39

#	Article	IF	Citations
37	LeafCutterMD: an algorithm for outlier splicing detection in rare diseases. Bioinformatics, 2020, 36, 4609-4615.	1.8	38
38	Development and Verification of an RNA Sequencing (RNA-Seq) Assay for the Detection of Gene Fusions in Tumors. Journal of Molecular Diagnostics, 2018, 20, 495-511.	1.2	36
39	Novel NR2F1 variants likely disrupt DNA binding: molecular modeling in two cases, review of published cases, genotypeâ€“phenotype correlation, and phenotypic expansion of the Boschâ€“Boonstraâ€“Schaaf optic atrophy syndrome. Journal of Physical Education and Sports Management, 2017, 3, a002162.	0.5	33
40	De novo variants in FBXO11 cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. European Journal of Human Genetics, 2019, 27, 738-746.	1.4	32
41	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	2.6	31
42	Computational classification of classically secreted proteins. Drug Discovery Today, 2007, 12, 234-240.	3.2	30
43	Pharmacogenomic findings from clinical whole exome sequencing of diagnostic odyssey patients. Molecular Genetics & Genomic Medicine, 2017, 5, 269-279.	0.6	30
44	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
45	Impact of sample acquisition and linear amplification on gene expression profiling of lung adenocarcinoma: laser capture micro-dissection cell-sampling versus bulk tissue-sampling. BMC Medical Genomics, 2009, 2, 13.	0.7	29
46	â€œBig Dataâ€•in Laboratory Medicine. Clinical Chemistry, 2015, 61, 1433-1440.	1.5	29
47	Utility of DNA, RNA, Protein, and Functional Approaches to Solve Cryptic Immunodeficiencies. Journal of Clinical Immunology, 2018, 38, 307-319.	2.0	29
48	Next Generation Sequencing of Sporadic Vestibular Schwannoma: Necessity of Biallelic NF2 Inactivation and Implications of Accessory Non-NF2 Variants. Otology and Neurotology, 2018, 39, e860-e871.	0.7	29
49	Kinase Genotype Analysis of Gastric Gastrointestinal Stromal Tumor Cytology Samples Using Targeted Next-Generation Sequencing. Clinical Gastroenterology and Hepatology, 2015, 13, 202-206.	2.4	28
50	Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency: Unique Presenting Laboratory Values and a Review of Biochemical and Clinical Features. JIMD Reports, 2017, 40, 63-69.	0.7	27
51	A tailored approach to fusion transcript identification increases diagnosis of rare inherited disease. PLoS ONE, 2019, 14, e0223337.	1.1	27
52	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. European Journal of Human Genetics, 2020, 28, 770-782.	1.4	27
53	Pilot study of small bowel mucosal gene expression in patients with irritable bowel syndrome with diarrhea. American Journal of Physiology - Renal Physiology, 2016, 311, G365-G376.	1.6	25
54	Recommendations for performance optimizations when using GATK3.8 and GATK4. BMC Bioinformatics, 2019, 20, 557.	1.2	25

#	Article	IF	Citations
55	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. European Journal of Human Genetics, 2020, 28, 1422-1431.	1.4	25
56	Impact of integrated translational research on clinical exome sequencing. Genetics in Medicine, 2021, 23, 498-507.	1.1	24
57	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila. American Journal of Human Genetics, 2021, 108, 1669-1691.	2.6	23
58	Novel de novo variant in EBF3 is likely to impact DNA binding in a patient with a neurodevelopmental disorder and expanded phenotypes: patient report, in silico functional assessment, and review of published cases. Journal of Physical Education and Sports Management, 2017, 3, a001743.	0.5	22
59	PCNT point mutations and familial intracranial aneurysms. Neurology, 2018, 91, e2170-e2181.	1.5	22
60	Whole Exome Sequencing Implicates an INO80D Mutation in a Syndrome of Aortic Hypoplasia, Premature Atherosclerosis, and Arterial Stiffness. Circulation: Cardiovascular Genetics, 2014, 7, 607-614.	5.1	21
61	Silent Tyrosinemia Type I Without Elevated Tyrosine or Succinylacetone Associated with Liver Cirrhosis and Hepatocellular Carcinoma. Human Mutation, 2016, 37, 1097-1105.	1.1	21
62	Clinical Applications and Utility of a Precision Medicine Approach for Patients With Unexplained Cytopenias. Mayo Clinic Proceedings, 2019, 94, 1753-1768.	1.4	21
63	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	1.1	21
64	Clinical Correlates and Treatment Outcomes for Patients With Short Telomere Syndromes. Mayo Clinic Proceedings, 2018, 93, 834-839.	1.4	20
65	COVID-19 Mortality Prediction From Deep Learning in a Large Multistate Electronic Health Record and Laboratory Information System Data Set: Algorithm Development and Validation. Journal of Medical Internet Research, 2021, 23, e30157.	2.1	20
66	Bioinformatics Methods for Prioritizing Serum Biomarker Candidates. Clinical Chemistry, 2006, 52, 2162-2164.	1.5	19
67	Quantitating tissue specificity of human genes to facilitate biomarker discovery. Bioinformatics, 2007, 23, 1348-1355.	1.8	19
68	Data Mining for Biomarker Development: A Review of Tissue Specificity Analysis. Clinics in Laboratory Medicine, 2008, 28, 127-143.	0.7	19
69	Characterization of three ciliopathy pedigrees expands the phenotype associated with biallelic C2CD3 variants. European Journal of Human Genetics, 2018, 26, 1797-1809.	1.4	19
70	Molecular modeling and molecular dynamic simulation of the effects of variants in the TGFBR2 kinase domain as a paradigm for interpretation of variants obtained by next generation sequencing. PLoS ONE, 2017, 12, e0170822.	1.1	19
71	A Novel Kleefstra Syndrome-associated Variant That Affects the Conserved TPLX Motif within the Ankyrin Repeat of EHMT1 Leads to Abnormal Protein Folding. Journal of Biological Chemistry, 2017, 292, 3866-3876.	1.6	18
72	Proteinâ€elongating mutations inMYH11are implicated in a dominantly inherited smooth muscle dysmotility syndrome with severe esophageal, gastric, and intestinal disease. Human Mutation, 2020, 41, 973-982.	1.1	18

#	Article	IF	Citations
73	SPECC1L regulates palate development downstream of IRF6. Human Molecular Genetics, 2020, 29, 845-858.	1.4	18
74	The prevalence of diseases caused by lysosome-related genes in a cohort of undiagnosed patients. Molecular Genetics and Metabolism Reports, 2017, 13, 46-51.	0.4	17
75	RNAâ€Seq detects a SAMD12â€EXT1 fusion transcript and leads to the discovery of an EXT1 deletion in a child with multiple osteochondromas. Molecular Genetics & Genomic Medicine, 2019, 7, e00560.	0.6	17
76	The Zebrafish Secretome. Zebrafish, 2008, 5, 131-138.	0.5	16
77	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	2.6	16
78	Variants in DOCK3 cause developmental delay and hypotonia. European Journal of Human Genetics, 2019, 27, 1225-1234.	1.4	15
79	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	2.6	15
80	A novel ANO3 variant identified in a 53-year-old woman presenting with hyperkinetic dysarthria, blepharospasm, hyperkinesias, and complex motorÂtics. BMC Medical Genetics, 2016, 17, 93.	2.1	14
81	Comparative analysis of de novo assemblers for variation discovery in personal genomes. Briefings in Bioinformatics, 2018, 19, 893-904.	3.2	14
82	A case of YY1â€associated syndromic learning disability or Gabrieleâ€de Vries syndrome with myasthenia gravis. American Journal of Medical Genetics, Part A, 2018, 176, 2846-2849.	0.7	14
83	Molecular characterization of known and novel ACVR1 variants in phenotypes of aberrant ossification. American Journal of Medical Genetics, Part A, 2019, 179, 1764-1777.	0.7	13
84	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	2.6	13
85	Genomics Integration Into Nephrology Practice. Kidney Medicine, 2021, 3, 785-798.	1.0	13
86	De novo coding variants in the AGO1 gene cause a neurodevelopmental disorder with intellectual disability. Journal of Medical Genetics, 2022, 59, 965-975.	1.5	13
87	Assessing Human Genetic Variations in Glucose Transporter SLC2A10 and Their Role in Altering Structural and Functional Properties. Frontiers in Genetics, 2018, 9, 276.	1.1	12
88	Haploinsufficiency as a disease mechanism in GNB1â€associated neurodevelopmental disorder. Molecular Genetics & Genomic Medicine, 2020, 8, e1477.	0.6	12
89	De novo variants of NR4A2 are associated with neurodevelopmental disorder and epilepsy. Genetics in Medicine, 2020, 22, 1413-1417.	1.1	12
90	AMOD: a morpholino oligonucleotide selection tool. Nucleic Acids Research, 2005, 33, W506-W511.	6.5	11

#	Article	IF	Citations
91	Multigenerational pedigree with STAR syndrome: A novel FAM58A variant and expansion of the phenotype. , 2017, 173, 1328-1333.		11
92	Functional Analysis of the SIM1 Variant p.G715V in 2 Patients With Obesity. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 355-361.	1.8	11
93	Pathogenic mutations in the chromokinesin KIF22 disrupt anaphase chromosome segregation. ELife, 0, 11, .	2.8	11
94	Target selection forDanio rerio functional genomics. Genesis, 2001, 30, 123-125.	0.8	10
95	Variability in assigning pathogenicity to incidental findings: insights from LDLR sequence linked to the electronic health record in 1013 individuals. European Journal of Human Genetics, 2017, 25, 410-415.	1.4	10
96	Clinical characteristics and platelet phenotype in a family withRUNX1mutated thrombocytopenia. Leukemia and Lymphoma, 2017, 58, 1963-1967.	0.6	10
97	Molecular modeling of LDLR aids interpretation of genomic variants. Journal of Molecular Medicine, 2019, 97, 533-540.	1.7	10
98	<scp>TSPEAR</scp> variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study. American Journal of Medical Genetics, Part A, 2021, 185, 2417-2433.	0.7	10
99	Autosomal Recessive Cerebellar Atrophy and Spastic Ataxia in Patients With Pathogenic Biallelic Variants in GEMIN5. Frontiers in Cell and Developmental Biology, 2022, 10, 783762.	1.8	10
100	Familial Creutzfeldt-Jakob Disease: Case report and role of genetic counseling in post mortem testing. Prion, 2016, 10, 502-506.	0.9	9
101	Pathogenic Variant in ACTB, p.Arg183Trp, Causes Juvenile-Onset Dystonia, Hearing Loss, and Developmental Delay without Midline Malformation. Case Reports in Genetics, 2017, 2017, 1-4.	0.1	9
102	Novel germline missense DDX41 variant in a patient with an adult-onset myeloid neoplasm with excess blasts without dysplasia. Leukemia and Lymphoma, 2019, 60, 1337-1339.	0.6	9
103	Aetiology and outcomes of secondary myelofibrosis occurring in the context of inherited platelet disorders: A single institutional study of four patients. British Journal of Haematology, 2020, 190, e316-e320.	1.2	9
104	SeekFusion - A Clinically Validated Fusion Transcript Detection Pipeline for PCR-Based Next-Generation Sequencing of RNA. Frontiers in Genetics, 2021, 12, 739054.	1.1	9
105	A novel de novo frameshift deletion in <scp>EHMT</scp>1 in a patient with Kleefstra Syndrome results in decreased H3K9 dimethylation. Molecular Genetics & Genomic Medicine, 2017, 5, 141-146.	0.6	8
106	Familial chronic megacolon presenting in childhood or adulthood: Seeking the presumed gene association. Neurogastroenterology and Motility, 2019, 31, e13550.	1.6	8
107	Variable expressivity of syndromic BMP4-related eye, brain, and digital anomalies: A review of the literature and description of three new cases. European Journal of Human Genetics, 2019, 27, 1379-1388.	1.4	8
108	Frequency of mitogen-activated protein kinase and phosphoinositide 3-kinase signaling pathway pathogenic alterations in EUS-FNA sampled malignant lymph nodes in rectal cancer with theranostic potential. Gastrointestinal Endoscopy, 2015, 82, 550-556.e1.	0.5	7

#	Article	IF	Citations
109	Functional validation reveals the novel missense V419L variant in TGFBR2 associated with Loeysâ€“Dietz syndrome (LDS) impairs canonical TGF-Î² signaling. Journal of Physical Education and Sports Management, 2017, 3, a001727.	0.5	7
110	Earlyâ€onset limbâ€girdle muscular dystrophyâ€2L in a female athlete. Muscle and Nerve, 2017, 55, E19-E21.	1.0	7
111	Extension of the mutational and clinical spectrum of SOX2 related disorders: Description of six new cases and a novel association with suprasellar teratoma. American Journal of Medical Genetics, Part A, 2018, 176, 2710-2719.	0.7	7
112	Co-occurrence of a maternally inherited DNMT3A duplication and a paternally inherited pathogenic variant in EZH2 in a child with growth retardation and severe short stature: atypical Weaver syndrome or evidence of a DNMT3A dosage effect?. Journal of Physical Education and Sports Management, 2018, 4, a002899.	0.5	7
113	Computational Detection of Known Pathogenic Gene Fusions in a Normal Tissue Database and Implications for Genetic Disease Research. Frontiers in Genetics, 2020, 11, 173.	1.1	7
114	Expansion of the Genotypic and Phenotypic Spectrum of WASF1-Related Neurodevelopmental Disorder. Brain Sciences, 2021, 11, 931.	1.1	7
115	Design considerations for workflow management systems use in production genomics research and the clinic. Scientific Reports, 2021, 11, 21680.	1.6	7
116	A Systematic Method for Selection of Promising Serum Protein Biomarkers to Improve Prostate Cancer (PCa1) Detection. Clinical Chemistry, 2006, 52, 2159-2162.	1.5	6
117	Developmental delay, coarse facial features, and epilepsy in a patient with EXT2 gene variants. Clinical Case Reports (discontinued), 2019, 7, 632-637.	0.2	6
118	X-Linked Lymphoproliferative Syndrome Presenting as Adult-Onset Multi-Infarct Dementia. Journal of Neuropathology and Experimental Neurology, 2019, 78, 460-466.	0.9	6
119	Exome sequencing confirms diagnosis of kabuki syndrome in an-adult with hodgkin lymphoma and unusually severe multisystem phenotype. Clinical Immunology, 2019, 207, 55-57.	1.4	6
120	Genetic variants in DGAT1 cause diverse clinical presentations of malnutrition through a specific molecular mechanism. European Journal of Medical Genetics, 2020, 63, 103817.	0.7	6
121	Novel loss-of-function variants in TRIO are associated with neurodevelopmental disorder: case report. BMC Medical Genetics, 2020, 21, 219.	2.1	6
122	Nail-patella-like renal disease masquerading as Fabry disease on kidney biopsy: a case report. BMC Nephrology, 2020, 21, 341.	0.8	6
123	Next-Generation Sequencing of CYP2C19 in Stent Thrombosis: Implications for Clopidogrel Pharmacogenomics. Cardiovascular Drugs and Therapy, 2021, 35, 549-559.	1.3	6
124	De novo <scp>PBX1</scp> variant in a patient with glaucoma, kidney anomalies, and developmental delay: An expansion of the <scp>CAKUTHED</scp> phenotype. American Journal of Medical Genetics, Part A, 2022, 188, 919-925.	0.7	6
125	Gain and loss of TASK3 channel function and its regulation by novel variation cause KCNK9 imprinting syndrome. Genome Medicine, 2022, 14, .	3.6	6
126	Three rare disease diagnoses in one patient through exome sequencing. Journal of Physical Education and Sports Management, 2019, 5, a004390.	0.5	5

#	Article	IF	Citations
127	An intragenic duplication of TRPS1 leading to abnormal transcripts and causing trichorhinophalangeal syndrome type I. Journal of Physical Education and Sports Management, 2019, 5, a004655.	0.5	5
128	Novel biallelic variants in MSTO1 associated with mitochondrial myopathy. Journal of Physical Education and Sports Management, 2019, 5, a004309.	0.5	5
129	Interpretation challenges of novel dualâ€class missense and spliceâ€impacting variant in POLR3Aâ€related lateâ€onset hereditary spastic ataxia. Molecular Genetics & Genomic Medicine, 2020, 8, e1341.	0.6	5
130	HELLO: improved neural network architectures and methodologies for small variant calling. BMC Bioinformatics, 2021, 22, 404.	1.2	5
131	Improved Characterization of Complex Î²-Globin Gene Cluster Structural Variants Using Long-Read Sequencing. Journal of Molecular Diagnostics, 2021, 23, 1732-1740.	1.2	5
132	Novel Pathogenic Variant in TGFBR2 Confirmed by Molecular Modeling Is a Rare Cause of Loeys-Dietz Syndrome. Case Reports in Genetics, 2017, 2017, 1-4.	0.1	4
133	GFAP canonical transcript may not be suitable for the diagnosis of adult-onset Alexander disease. Acta Neuropathologica Communications, 2018, 6, 112.	2.4	4
134	Expansion of PURA-Related Phenotypes and Discovery of a Novel PURA Variant: A Case Report. Child Neurology Open, 2020, 7, 2329048X2095500.	0.5	4
135	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395.	1.1	4
136	LPCAT1-TERT fusions are uniquely recurrent in epithelioid trophoblastic tumors and positively regulate cell growth. PLoS ONE, 2021, 16, e0250518.	1.1	4
137	â€œThe moleculeâ€™s the thing:â€•the promise of molecular modeling and dynamic simulations in aiding the prioritization and interpretation of genomic testing results. F1000Research, 2016, 5, 766.	0.8	4
138	Heterozygous variants in PRPF8 are associated with neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2022, 188, 2750-2759.	0.7	4
139	Functional characterization of a <scp>GFAP</scp> variant of uncertain significance in an Alexander disease case within the setting of an individualized medicine clinic. Clinical Case Reports (discontinued), 2016, 4, 885-895.	0.2	3
140	Preemptive sequencing in the genomic medicine era. Expert Review of Precision Medicine and Drug Development, 2017, 2, 91-98.	0.4	3
141	Improving Single-Nucleotide Polymorphism-Based Fetal Fraction Estimation of Maternal Plasma Circulating Cell-Free DNA Using Bayesian Hierarchical Models. Journal of Computational Biology, 2018, 25, 1040-1049.	0.8	3
142	An activating germline IDH1 variant associated with a tumor entity characterized by unilateral and bilateral chondrosarcoma of the mastoid. Human Genetics and Genomics Advances, 2020, 1, 100006.	1.0	3
143	Jumonji domain containing 1C (JMJD1C) sequence variants in seven patients with autism spectrum disorder, intellectual disability and seizures. European Journal of Medical Genetics, 2020, 63, 103850.	0.7	3
144	Epigenetic alteration contributes to the transcriptional reprogramming in T-cell prolymphocytic leukemia. Scientific Reports, 2021, 11, 8318.	1.6	3

#	Article	IF	Citations
145	Functional validation of TERT and TERC variants of uncertain significance in patients with short telomere syndromes. Blood Cancer Journal, 2020, 10, 120.	2.8	2
146	Recurrent ganglioneuroma in <scp>PTPN11</scp>â€associated Noonan syndrome: A case report and literature review. American Journal of Medical Genetics, Part A, 2021, 185, 1883-1887.	0.7	2
147	Late onset asymptomatic pancreatic neuroendocrine tumor â€“ A case report on the phenotypic expansion for MEN1. Hereditary Cancer in Clinical Practice, 2017, 15, 10.	0.6	1
148	Proposal for Modification of Cahan's Criteria Utilizing Molecular Genetic Analyses for Cases without Baseline Histopathology: A Unique Method Applicable to Primary Radiosurgery. Journal of Neurological Surgery, Part B: Skull Base, 2019, 80, 010-017.	0.4	1
149	Biallelic variants in PROZ as a cause of hypercoagulability and livedo racemosa. Thrombosis Research, 2020, 195, 187-189.	0.8	1
150	Successful Treatment of Skewed Lyonization Associated with X-Linked CGD in a Female Presenting with Recalcitrant Crohnâ€™s Disease. Journal of Clinical Immunology, 2020, 40, 1056-1061.	2.0	1
151	A novel missense variant and multiexon deletion causing a delayed presentation of xeroderma pigmentosum, group C. Journal of Physical Education and Sports Management, 2020, 6, a005165.	0.5	1
152	P2T2: Protein Panoramic annoTation Tool for the interpretation of protein coding genetic variants. JAMIA Open, 2021, 4, ooab065.	1.0	1
153	Long Range Sequencing Shows Improved Resolution in the Detection of Beta Globin Cluster Variants. Blood, 2019, 134, 3548-3548.	0.6	1
154	Genomic Profiling Reveals Molecular Heterogeneity in Patients with Richter's Syndrome (RS) and Progressive Chronic Lymphocytic Leukemia (CLL). Blood, 2020, 136, 16-17.	0.6	1
155	Functional validation of a novel AAAS variant in an atypical presentation of Allgrove syndrome. Molecular Genetics & Genomic Medicine, 2022, , e1966.	0.6	1
156	Whole Exome Sequencing Leading to the Diagnosis of Dysferlinopathy with a Novel Missense Mutation (c.959G>C). Case Reports in Genetics, 2016, 2016, 1-4.	0.1	0
157	Interpretation of Dihydrorhodamine-1,2,3 Flow Cytometry in Chronic Granulomatous Disease: an Atypical Exemplar. Journal of Clinical Immunology, 2022, , 1.	2.0	0
158	Editorial: Clinical Genome Sequencing: Bioinformatics Challenges and Key Considerations. Frontiers in Genetics, 2022, 13, 896032.	1.1	0