Richard A Gibbs

List of Publications by Year
in descending order

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388	91,418	2093	373
papers	citations	100 h-index	281 g-index

406	406	406	112679
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Initial sequencing and analysis of the human genome. Nature, 2001, 409, 860-921.	13.7	21,074
2	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	13.7	13,998
3	A map of human genome variation from population-scale sequencing. Nature, 2010, 467, 1061-1073.	13.7	7,209
4	Genomic analyses identify molecular subtypes of pancreatic cancer. Nature, 2016, 531, 47-52.	13.7	2,700
5	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	13.7	1,994
6	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. Nature, 2004, 428, 493-521.	13.7	1,943
7	Pancreatic cancer genomes reveal aberrations in axon guidance pathway genes. Nature, 2012, 491, 399-405.	13.7	1,741
8	Cell-of-Origin Patterns Dominate the Molecular Classification of 10,000 Tumors from 33 Types of Cancer. Cell, 2018, 173, 291-304.e6.	13.5	1,718
9	Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. New England Journal of Medicine, 2013, 369, 1502-1511.	13.9	1,717
10	Temporal development of the gut microbiome in early childhood from the TEDDY study. Nature, 2018, 562, 583-588.	13.7	1,220
11	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	3.8	1,171
12	A framework for the interpretation of de novo mutation in human disease. Nature Genetics, 2014, 46, 944-950.	9.4	943
13	Mind the Gap: Upgrading Genomes with Pacific Biosciences RS Long-Read Sequencing Technology. PLoS ONE, 2012, 7, e47768.	1.1	896
14	Comparison and integration of deleteriousness prediction methods for nonsynonymous SNVs in whole exome sequencing studies. Human Molecular Genetics, 2015, 24, 2125-2137.	1.4	892
15	Whole-genome landscape of pancreatic neuroendocrine tumours. Nature, 2017, 543, 65-71.	13.7	716
16	Whole-Genome Sequencing in a Patient with Charcotâ€“Marieâ€“Tooth Neuropathy. New England Journal of Medicine, 2010, 362, 1181-1191.	13.9	698
17	The Somatic Genomic Landscape of Chromophobe Renal Cell Carcinoma. Cancer Cell, 2014, 26, 319-330.	7.7	665
18	Trans-ancestry mutational landscape of hepatocellular carcinoma genomes. Nature Genetics, 2014, 46, 1267-1273.	9.4	655

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19	The gut mycobiome of the Human Microbiome Project healthy cohort. Microbiome, 2017, 5, 153.	4.9	609
20	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
21	Natural variation in genome architecture among 205 Drosophila melanogaster Genetic Reference Panel lines. Genome Research, 2014, 24, 1193-1208.	2.4	565
22	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	13.9	565
23	Mammalian Y chromosomes retain widely expressed dosage-sensitive regulators. Nature, 2014, 508, 494-499.	13.7	546
24	Comparative and demographic analysis of orang-utan genomes. Nature, 2011, 469, 529-533.	13.7	541
25	The Cancer Genome Atlas Comprehensive Molecular Characterization of Renal Cell Carcinoma. Cell Reports, 2018, 23, 313-326.e5.	2.9	523
26	Mutational Landscape of Aggressive Cutaneous Squamous Cell Carcinoma. Clinical Cancer Research, 2014, 20, 6582-6592.	3.2	493
27	The sheep genome illuminates biology of the rumen and lipid metabolism. Science, 2014, 344, 1168-1173.	6.0	436
28	Convergent evolution of the genomes of marine mammals. Nature Genetics, 2015, 47, 272-275.	9.4	392
29	The Matchmaker Exchange: A Platform for Rare Disease Gene Discovery. Human Mutation, 2015, 36, 915-921.	1.1	390
30	Diagnostic Yield of Clinical Tumor and Germline Whole-Exome Sequencing for Children With Solid Tumors. JAMA Oncology, 2016, 2, 616.	3.4	378
31	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	3.3	348
32	Integrative Annotation of Variants from 1092 Humans: Application to Cancer Genomics. Science, 2013, 342, 1235587.	6.0	341
33	A comprehensive transcriptional map of primate brain development. Nature, 2016, 535, 367-375.	13.7	341
34	Clan Genomics and the Complex Architecture of Human Disease. Cell, 2011, 147, 32-43.	13.5	330
35	The genomes of two key bumblebee species with primitive eusocial organization. Genome Biology, 2015, 16, 76.	3.8	330
36	Gibbon genome and the fast karyotype evolution of small apes. Nature, 2014, 513, 195-201.	13.7	320

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37	A recombination hotspot responsible for two inherited peripheral neuropathies is located near a mariner transposon-like element. Nature Genetics, 1996, 12, 288-297.	9.4	304
38	COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. Nature Genetics, 2015, 47, 654-660.	9.4	302
39	Multilevel Genomics-Based Taxonomy of Renal Cell Carcinoma. Cell Reports, 2016, 14, 2476-2489.	2.9	298
40	The house spider genome reveals an ancient whole-genome duplication during arachnid evolution. BMC Biology, 2017, 15, 62.	1.7	286
41	Structure and function of the healthy pre-adolescent pediatric gut microbiome. Microbiome, 2015, 3, 36.	4.9	283
42	Genomic profiling of SÃ©zary syndrome identifies alterations of key T cell signaling and differentiation genes. Nature Genetics, 2015, 47, 1426-1434.	9.4	276
43	Identification of mutations leading to the Lesch-Nyhan syndrome by automated direct DNA sequencing of in vitro amplified cDNA.. Proceedings of the National Academy of Sciences of the United States of America, 1989, 86, 1919-1923.	3.3	272
44	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	1.5	261
45	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	3.8	258
46	An integrative variant analysis suite for whole exome next-generation sequencing data. BMC Bioinformatics, 2012, 13, 8.	1.2	252
47	Exonuclease mutations in DNA polymerase epsilon reveal replication strand specific mutation patterns and human origins of replication. Genome Research, 2014, 24, 1740-1750.	2.4	244
48	Genome of the Asian longhorned beetle (Anoplophora glabripennis), a globally significant invasive species, reveals key functional and evolutionary innovations at the beetleâ€“plant interface. Genome Biology, 2016, 17, 227.	3.8	244
49	Identification of FMR2, a novel gene associated with the FRAXE CCG repeat and CpG island. Nature Genetics, 1996, 13, 109-113.	9.4	238
50	Genomic innovations, transcriptional plasticity and gene loss underlying the evolution and divergence of two highly polyphagous and invasive Helicoverpa pest species. BMC Biology, 2017, 15, 63.	1.7	238
51	Comparative primate genomics: emerging patterns of genome content and dynamics. Nature Reviews Genetics, 2014, 15, 347-359.	7.7	234
52	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	4.5	234
53	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713.	5.8	227
54	Hemimetabolous genomes reveal molecular basis of termite eusociality. Nature Ecology and Evolution, 2018, 2, 557-566.	3.4	223

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55	Genome-culture coevolution promotes rapid divergence of killer whale ecotypes. Nature Communications, 2016, 7, 11693.	5.8	222
56	The First Myriapod Genome Sequence Reveals Conservative Arthropod Gene Content and Genome Organisation in the Centipede Strigamia maritima. PLoS Biology, 2014, 12, e1002005.	2.6	221
57	Hemichordate genomes and deuterostome origins. Nature, 2015, 527, 459-465.	13.7	217
58	A model species for agricultural pest genomics: the genome of the Colorado potato beetle, Leptinotarsa decemlineata (Coleoptera: Chrysomelidae). Scientific Reports, 2018, 8, 1931.	1.6	215
59	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183.	2.9	211
60	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
61	Launching genomics into the cloud: deployment of Mercury, a next generation sequence analysis pipeline. BMC Bioinformatics, 2014, 15, 30.	1.2	199
62	High-depth African genomes inform human migration and health. Nature, 2020, 586, 741-748.	13.7	197
63	Targeted enrichment beyond the consensus coding DNA sequence exome reveals exons with higher variant densities. Genome Biology, 2011, 12, R68.	13.9	192
64	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulumâ€“associated degradation pathway. Genetics in Medicine, 2014, 16, 751-758.	1.1	191
65	Whole exome sequencing study identifies novel rare and common Alzheimerâ€™s-Associated variants involved in immune response and transcriptional regulation. Molecular Psychiatry, 2020, 25, 1859-1875.	4.1	191
66	Genomic Encyclopedia of Bacteria and Archaea: Sequencing a Myriad of Type Strains. PLoS Biology, 2014, 12, e1001920.	2.6	190
67	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650.	13.5	189
68	Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685.	1.1	186
69	Unique features of a global human ectoparasite identified through sequencing of the bed bug genome. Nature Communications, 2016, 7, 10165.	5.8	184
70	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
71	Novel somatic and germline mutations in intracranial germ cell tumours. Nature, 2014, 511, 241-245.	13.7	181
72	Whole-exome sequencing points to considerable genetic heterogeneity of cerebral palsy. Molecular Psychiatry, 2015, 20, 176-182.	4.1	178

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73	Sequence and analysis of chromosome 2 of Dictyostelium discoideum. Nature, 2002, 418, 79-85.	13.7	176
74	Low-frequency and rare exome chip variants associate with fasting glucose and type 2 diabetes susceptibility. Nature Communications, 2015, 6, 5897.	5.8	173
75	Germline Mutations in Shelterin Complex Genes Are Associated With Familial Glioma. Journal of the National Cancer Institute, 2015, 107, 384.	3.0	172
76	A SNP discovery method to assess variant allele probability from next-generation resequencing data. Genome Research, 2010, 20, 273-280.	2.4	168
77	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	1.7	163
78	Prospective virome analyses in young children at increased genetic risk for type 1 diabetes. Nature Medicine, 2019, 25, 1865-1872.	15.2	161
79	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
80	Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles. American Journal of Human Genetics, 2018, 103, 171-187.	2.6	160
81	Next-generation sequencing identifies rare variants associated with Noonan syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 11473-11478.	3.3	158
82	Evolutionary History of Chemosensory-Related Gene Families across the Arthropoda. Molecular Biology and Evolution, 2017, 34, 1838-1862.	3.5	157
83	Assessing structural variation in a personal genomeâ€"towards a human reference diploid genome. BMC Genomics, 2015, 16, 286.	1.2	153
84	Gene content evolution in the arthropods. Genome Biology, 2020, 21, 15.	3.8	150
85	PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. American Journal of Human Genetics, 2014, 95, 96-107.	2.6	148
86	Comparative validation of the D. melanogaster modENCODE transcriptome annotation. Genome Research, 2014, 24, 1209-1223.	2.4	147
87	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	2.6	146
88	Exome sequencing resolves apparent incidental findings and reveals further complexity of SH3TC2 variant alleles causing Charcot-Marie-Tooth neuropathy. Genome Medicine, 2013, 5, 57.	3.6	143
89	A BAC-Based Physical Map of the Major Autosomes of Drosophila melanogaster. Science, 2000, 287, 2271-2274.	6.0	142
90	Transmission event of SARS-CoV-2 delta variant reveals multiple vaccine breakthrough infections. BMC Medicine, 2021, 19, 255.	2.3	137

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91	Duplication of a gene-rich cluster between 16p11.1 and Xq28: a novel pericentromeric-directed mechanism for paralogous genome evolution. Human Molecular Genetics, 1996, 5, 899-912.	1.4	136
92	The whole genome sequence of the Mediterranean fruit fly, Ceratitis capitata (Wiedemann), reveals insights into the biology and adaptive evolution of a highly invasive pest species. Genome Biology, 2016, 17, 192.	3.8	130
93	Diversity and evolution of the transposable element repertoire in arthropods with particular reference to insects. Bmc Ecology and Evolution, 2019, 19, 11.	0.7	129
94	De novo truncating mutations in ASXL3 are associated with a novel clinical phenotype with similarities to Bohring-Opitz syndrome. Genome Medicine, 2013, 5, 11.	3.6	128
95	Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. American Journal of Human Genetics, 2018, 102, 1126-1142.	2.6	128
96	Sheep genome functional annotation reveals proximal regulatory elements contributed to the evolution of modern breeds. Nature Communications, 2018, 9, 859.	5.8	126
97	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309.	2.6	125
98	Squamous Cell Carcinoma of the Oral Tongue in Young Non-Smokers Is Genomically Similar to Tumors in Older Smokers. Clinical Cancer Research, 2014, 20, 3842-3848.	3.2	124
99	The Earth BioGenome Project 2020: Starting the clock. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	3.3	124
100	The extent of genetic variation in the CCR5 gene. Nature Genetics, 1997, 16, 221-222.	9.4	123
101	Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. PLoS Genetics, 2014, 10, e1004258.	1.5	122
102	The comparative genomics and complex population history of Papio baboons. Science Advances, 2019, 5, eaau6947.	4.7	115
103	Molecular evolutionary trends and feeding ecology diversification in the Hemiptera, anchored by the milkweed bug genome. Genome Biology, 2019, 20, 64.	3.8	114
104	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	13.5	113
105	DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 612-622.	2.6	110
106	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	4.9	108
107	Ampullary Cancers Harbor ELF3 Tumor Suppressor Gene Mutations and Exhibit Frequent WNT Dysregulation. Cell Reports, 2016, 14, 907-919.	2.9	107
108	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. Genome Medicine, 2018, 10, 74.	3.6	105

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109	Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537.	1.1	104
110	Genome Sequencing of the Phytoseiid Predatory MiteMetaseiulus occidentalisReveals Completely AtomizedHoxGenes and Superdynamic Intron Evolution. Genome Biology and Evolution, 2016, 8, 1762-1775.	1.1	102
111	Chemistry-First Approach for Nomination of Personalized Treatment in Lung Cancer. Cell, 2018, 173, 864-878.e29.	13.5	102
112	The population genomics of rhesus macaques (Macaca mulatta) based on whole-genome sequences. Genome Research, 2016, 26, 1651-1662.	2.4	101
113	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237.	6.5	98
114	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	2.6	98
115	SYT1-associated neurodevelopmental disorder: a case series. Brain, 2018, 141, 2576-2591.	3.7	98
116	WGSA: an annotation pipeline for human genome sequencing studies. Journal of Medical Genetics, 2016, 53, 111-112.	1.5	96
117	Reproductive Longevity Predicts Mutation Rates in Primates. Current Biology, 2018, 28, 3193-3197.e5.	1.8	94
118	Analysis commons, a team approach to discovery in a big-data environment for genetic epidemiology. Nature Genetics, 2017, 49, 1560-1563.	9.4	93
119	Genetics of schizophrenia in the South African Xhosa. Science, 2020, 367, 569-573.	6.0	93
120	Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. American Journal of Human Genetics, 2014, 95, 579-583.	2.6	92
121	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811.	1.1	92
122	DVL3 Alleles Resulting in a â˜1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2016, 98, 553-561.	2.6	88
123	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	2.6	88
124	The Human Genome Project changed everything. Nature Reviews Genetics, 2020, 21, 575-576.	7.7	84
125	Molecular and phenotypic variation in patients with severe Hunter syndrome. Human Molecular Genetics, 1997, 6, 479-486.	1.4	82
126	The human COX10 gene is disrupted during homologous recombination between the 24 kb proximal and distal CMT1A-REPs. Human Molecular Genetics, 1997, 6, 1595-1603.	1.4	81

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127	Sooty mangabey genome sequence provides insight into AIDS resistance in a natural SIV host. Nature, 2018, 553, 77-81.	13.7	81
128	The transcription factor POU3F2 regulates a gene coexpression network in brain tissue from patients with psychiatric disorders. Science Translational Medicine, 2018, 10, .	5.8	81
129	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754.	2.6	80
130	Monoallelic and Biallelic Mutations in MAB21L2 Cause a Spectrum of Major Eye Malformations. American Journal of Human Genetics, 2014, 94, 915-923.	2.6	79
131	The Toxicogenome of Hyalella azteca: A Model for Sediment Ecotoxicology and Evolutionary Toxicology. Environmental Science & Technology, 2018, 52, 6009-6022.	4.6	79
132	Biallelic mutations in IRF8 impair human NK cell maturation and function. Journal of Clinical Investigation, 2016, 127, 306-320.	3.9	76
133	Enrichment of mutations in chromatin regulators in people with Rett syndrome lacking mutations in MECP2. Genetics in Medicine, 2017, 19, 13-19.	1.1	74
134	The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. American Journal of Human Genetics, 2019, 105, 132-150.	2.6	74
135	Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. Genetics in Medicine, 2017, 19, 412-420.	1.1	73
136	Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. Cell, 2019, 176, 1310-1324.e10.	13.5	73
137	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. PLoS Biology, 2020, 18, e3000954.	2.6	73
138	Whole-Exome Sequencing in Familial Parkinson Disease. JAMA Neurology, 2016, 73, 68.	4.5	71
139	Lucilia cuprina genome unlocks parasitic fly biology to underpin future interventions. Nature Communications, 2015, 6, 7344.	5.8	67
140	Monoallelic and Biallelic Variants in EMC1 Identified in Individuals with Global Developmental Delay, Hypotonia, Scoliosis, and Cerebellar Atrophy. American Journal of Human Genetics, 2016, 98, 562-570.	2.6	66
141	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	13.5	66
142	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American Journal of Human Genetics, 2015, 97, 904-913.	2.6	65
143	HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. Science, 2020, 369, 202-207.	6.0	65
144	PacBio-LITS: a large-insert targeted sequencing method for characterization of human disease-associated chromosomal structural variations. BMC Genomics, 2015, 16, 214.	1.2	63

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145	Tissue-specific transcriptome sequencing analysis expands the non-human primate reference transcriptome resource (NHPRTR). Nucleic Acids Research, 2015, 43, D737-D742.	6.5	61
146	Obtaining informed consent for clinical tumor and germline exome sequencing of newly diagnosed childhood cancer patients. Genome Medicine, 2014, 6, 69.	3.6	60
147	Brown marmorated stink bug, Halyomorpha halys (StÃ¥l), genome: putative underpinnings of polyphagy, insecticide resistance potential and biology of a top worldwide pest. BMC Genomics, 2020, 21, 227.	1.2	60
148	Leveraging Human Microbiome Features to Diagnose and Stratify Children with Irritable Bowel Syndrome. Journal of Molecular Diagnostics, 2019, 21, 449-461.	1.2	59
149	Whole-exome sequencing identifies novel homozygous mutation inÂNPAS2 in family with nonobstructive azoospermia. Fertility and Sterility, 2015, 104, 286-291.	0.5	58
150	Identification of Polycystic Kidney Disease 1 Like 1 Gene Variants in Children With Biliary Atresia Splenic Malformation Syndrome. Hepatology, 2019, 70, 899-910.	3.6	58
151	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	1.1	58
152	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. American Journal of Human Genetics, 2014, 94, 784-789.	2.6	57
153	Bi-allelic Mutations in PKD1L1 Are Associated with Laterality Defects in Humans. American Journal of Human Genetics, 2016, 99, 886-893.	2.6	57
154	Comparative genomics of the miniature wasp and pest control agent Trichogramma pretiosum. BMC Biology, 2018, 16, 54.	1.7	57
155	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	2.6	56
156	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	2.6	55
157	Germline Cancer Predisposition Variants inâ€, Pediatric Rhabdomyosarcoma: A Report From the Childrenâ€™s Oncology Group. Journal of the National Cancer Institute, 2021, 113, 875-883.	3.0	55
158	Mutations inVRK1Associated With Complex Motor and Sensory Axonal Neuropathy Plus Microcephaly. JAMA Neurology, 2013, 70, 1491-8.	4.5	54
159	SeqCNV: a novel method for identification of copy number variations in targeted next-generation sequencing data. BMC Bioinformatics, 2017, 18, 147.	1.2	54
160	Genome-enabled insights into the biology of thrips as crop pests. BMC Biology, 2020, 18, 142.	1.7	54
161	Mutations in COL27A1 cause Steel syndrome and suggest a founder mutation effect in the Puerto Rican population. European Journal of Human Genetics, 2015, 23, 342-346.	1.4	53
162	Hybrid de novo genome assembly and centromere characterization of the gray mouse lemur (Microcebus murinus). BMC Biology, 2017, 15, 110.	1.7	53

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163	Exome Sequencing of a Primary Ovarian Insufficiency Cohort Reveals Common Molecular Etiologies for a Spectrum of Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3049-3067.	1.8	53
164	Genomic Signatures of Cooperation and Conflict in the Social Amoeba. Current Biology, 2015, 25, 1661-1665.	1.8	51
165	Parliament2: Accurate structural variant calling at scale. GigaScience, 2020, 9, .	3.3	51
166	Practical Approaches for Whole-Genome Sequence Analysis of Heart- and Blood-Related Traits. American Journal of Human Genetics, 2017, 100, 205-215.	2.6	50
167	Analysis of loss-of-function variants and 20 risk factor phenotypes in 8,554 individuals identifies loci influencing chronic disease. Nature Genetics, 2015, 47, 640-642.	9.4	49
168	Loss of Function Mutations in NNT Are Associated With Left Ventricular Noncompaction. Circulation: Cardiovascular Genetics, 2015, 8, 544-552.	5.1	48
169	Analyses of SLC13A5 -epilepsy patients reveal perturbations of TCA cycle. Molecular Genetics and Metabolism, 2017, 121, 314-319.	0.5	48
170	Best practices for the interpretation and reporting of clinical whole genome sequencing. Npj Genomic Medicine, 2022, 7, 27.	1.7	48
171	Secondary findings and carrier test frequencies in a large multiethnic sample. Genome Medicine, 2015, 7, 54.	3.6	47
172	Rare variants in the notch signaling pathway describe a novel type of autosomal recessive Klippelâ€“Feil syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2795-2799.	0.7	47
173	The genome of the water strider Gerris buenoi reveals expansions of gene repertoires associated with adaptations to life on the water. BMC Genomics, 2018, 19, 832.	1.2	47
174	16S gut community of the Cameron County Hispanic Cohort. Microbiome, 2015, 3, 7.	4.9	46
175	Loss-of-function variants influence the human serum metabolome. Science Advances, 2016, 2, e1600800.	4.7	46
176	Whole-Exome Sequencing Identifies Loci Associated with Blood Cell Traits and Reveals a Role for Alternative GFI1B Splice Variants in Human Hematopoiesis. American Journal of Human Genetics, 2016, 99, 481-488.	2.6	45
177	REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis. American Journal of Human Genetics, 2017, 101, 149-156.	2.6	44
178	Whole-Exome Sequencing Identifies Novel Variants for Tooth Agenesis. Journal of Dental Research, 2018, 97, 49-59.	2.5	44
179	Genetic architecture of laterality defects revealed by whole exome sequencing. European Journal of Human Genetics, 2019, 27, 563-573.	1.4	44
180	Human NK cell deficiency as a result of biallelic mutations in MCM10. Journal of Clinical Investigation, 2020, 130, 5272-5286.	3.9	44

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181	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
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