Richard A Gibbs

List of Publications by Year
in descending order

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388	91,418	2098	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	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	3.7	17
2	Genetic errors of immunity distinguish pediatric nonmalignant lymphoproliferative disorders. Journal of Allergy and Clinical Immunology, 2022, 149, 758-766.	1.5	6
3	Expanding the phenotypic and allelic spectrum of <scp>SMG8</scp>: Clinical observations reveal overlap with <scp>SMG9</scp>â€associated disease trait. American Journal of Medical Genetics, Part A, 2022, 188, 648-657.	0.7	3
4	Quantitative dissection of multilocus pathogenic variation in an Egyptian infant with severe neurodevelopmental disorder resulting from multiple molecular diagnoses. American Journal of Medical Genetics, Part A, 2022, 188, 735-750.	0.7	14
5	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074.	1.0	14
6	Distinct somatic DICER1 hotspot mutations in three metachronous ovarian Sertoli-Leydig cell tumors in a patient with DICER1 syndrome. Cancer Genetics, 2022, 262-263, 53-56.	0.2	2
7	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
8	Rare coding variants in 35 genes associate with circulating lipid levelsâ€”A multi-ancestry analysis of 170,000 exomes. American Journal of Human Genetics, 2022, 109, 81-96.	2.6	24
9	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44
10	Fully resolved assembly of Cryptosporidium parvum. GigaScience, 2022, 11, .	3.3	8
11	Abstract PD9-03: Pam50 intrinsic subtype and risk of recurrence score (ROR) for the prediction of endocrine (ET) sensitivity and pathologic response to chemotherapy in postmenopausal women with clinical stage II/III estrogen receptor positive (ER+) and HER2 negative (HER2-) breast cancer (BC) in the alternate trial (Alliance A011106). Cancer Research, 2022, 82, PD9-03-PD9-03.	0.4	0
12	Whole-genome sequencing as an investigational device for return of hereditary disease risk and pharmacogenomic results as part of the All of Us Research Program. Genome Medicine, 2022, 14, 34.	3.6	27
13	Novel RETREG1 (<scp>FAM134B</scp> founder allele is linked to <scp>HSAN2B</scp> and renal disease in a Turkish family. American Journal of Medical Genetics, Part A, 2022, 188, 2153-2161.	0.7	4
14	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	1.1	8
15	Implementation of preemptive DNA sequenceâ€“based pharmacogenomics testing across a large academic medical center: The Mayo-Baylor RIGHT 10K Study. Genetics in Medicine, 2022, 24, 1062-1072.	1.1	28
16	Multiple Respiratory Syncytial Virus (RSV) Strains Infecting HEp-2 and A549 Cells Reveal Cell Line-Dependent Differences in Resistance to RSV Infection. Journal of Virology, 2022, , e0190421.	1.5	17
17	Best practices for the interpretation and reporting of clinical whole genome sequencing. Npj Genomic Medicine, 2022, 7, 27.	1.7	48
18	Harmonizing variant classification for return of results in the All of Us Research Program. Human Mutation, 2022, 43, 1114-1121.	1.1	7

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19	Association of Pathogenic Variants in Hereditary Cancer Genes With Multiple Diseases. JAMA Oncology, 2022, 8, 835.	3.4	25
20	Biallelic Variants in the Ectonucleotidase <scp>ENTPD1</scp> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Annals of Neurology, 2022, 92, 304-321.	2.8	2
21	Whole-exome sequencing of 14â€‰389 individuals from the ESP and CHARGE consortia identifies novel rare variation associated with hemostatic factors. Human Molecular Genetics, 2022, 31, 3120-3132.	1.4	3
22	Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3593-3600.	0.7	16
23	Germline mutation in POLR2A: a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. Human Genetics and Genomics Advances, 2021, 2, 100014.	1.0	10
24	NMIHBA results from hypomorphic PRUNE1 variants that lack short-chain exopolyphosphatase activity. Human Molecular Genetics, 2021, 29, 3516-3531.	1.4	16
25	Neurodevelopmental disorder in an Egyptian family with a biallelic <scp>ALKBH8</scp> variant. American Journal of Medical Genetics, Part A, 2021, 185, 1288-1293.	0.7	13
26	Perturbations of genes essential for MÃ¼llerian duct and WÃ¶lffian duct development in Mayer-Rokitansky-KÃ¼ster-Hauser syndrome. American Journal of Human Genetics, 2021, 108, 337-345.	2.6	41
27	RCL1 copy number variants are associated with a range of neuropsychiatric phenotypes. Molecular Psychiatry, 2021, 26, 1706-1718.	4.1	10
28	DNA methylation patterns identify subgroups of pancreatic neuroendocrine tumors with clinical association. Communications Biology, 2021, 4, 155.	2.0	26
29	Phenotypic and protein localization heterogeneity associated with AHDC1 pathogenic proteinâ€truncating alleles in Xiaâ€“Gibbs syndrome. Human Mutation, 2021, 42, 577-591.	1.1	14
30	muCNV: genotyping structural variants for population-level sequencing. Bioinformatics, 2021, 37, 2055-2057.	1.8	7
31	Chromosome Xq23 is associated with lower atherogenic lipid concentrations and favorable cardiometabolic indices. Nature Communications, 2021, 12, 2182.	5.8	17
32	A novel homozygous <scp>SLC13A5</scp> wholeâ€gene deletion generated by <scp>Alu/Alu</scp>â€mediated rearrangement in an Iraqi family with epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2021, 185, 1972-1980.	0.7	16
33	Biallelic Pathogenic Variants in TNNT3 Associated With Congenital Myopathy. Neurology: Genetics, 2021, 7, e589.	0.9	6
34	Immune Dysregulation Mimicking Systemic Lupus Erythematosus in a Patient With Lysinuric Protein Intolerance: Case Report and Review of the Literature. Frontiers in Pediatrics, 2021, 9, 673957.	0.9	12
35	Two novel biâ€allelic <scp>KDELR2</scp> missense variants cause osteogenesis imperfecta with neurodevelopmental features. American Journal of Medical Genetics, Part A, 2021, 185, 2241-2249.	0.7	7
36	Genomic considerations for FHIRÂ®; eMERGE implementation lessons. Journal of Biomedical Informatics, 2021, 118, 103795.	2.5	15

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37	Risk of sudden cardiac death in <scp>EXOSC5</scp>â€related disease. American Journal of Medical Genetics, Part A, 2021, 185, 2532-2540.	0.7	6
38	Exome variant discrepancies due to reference-genome differences. American Journal of Human Genetics, 2021, 108, 1239-1250.	2.6	36
39	Neptune: an environment for the delivery of genomic medicine. Genetics in Medicine, 2021, 23, 1838-1846.	1.1	3
40	Sequencing of a central nervous system tumor demonstrates cancer transmission in an organ transplant. Life Science Alliance, 2021, 4, e202000941.	1.3	1
41	Durable Response to Larotrectinib in a Child With Histologic Diagnosis of Recurrent Disseminated Ependymoma Discovered to Harbor an NTRK2 Fusion: The Impact of Integrated Genomic Profiling. JCO Precision Oncology, 2021, 5, 1221-1227.	1.5	5
42	Biallelic loss-of-function variants in the splicing regulator NSRP1 cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. Genetics in Medicine, 2021, 23, 2455-2460.	1.1	9
43	Exome sequencing in children with clinically suspected <scp>maturityâ€onset</scp> diabetes of the young. Pediatric Diabetes, 2021, 22, 960-968.	1.2	6
44	Oligonucleotide capture sequencing of the SARS-CoV-2 genome and subgenomic fragments from COVID-19 individuals. PLoS ONE, 2021, 16, e0244468.	1.1	20
45	Genetic testing in ambulatory cardiology clinics reveals high rate of findings with clinical management implications. Genetics in Medicine, 2021, 23, 2404-2414.	1.1	14
46	COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay. American Journal of Human Genetics, 2021, 108, 1710-1724.	2.6	18
47	Deep clinicopathological phenotyping identifies a previously unrecognized pathogenic EMD splice variant. Annals of Clinical and Translational Neurology, 2021, 8, 2052-2058.	1.7	1
48	High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. American Journal of Human Genetics, 2021, 108, 1981-2005.	2.6	38
49	Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808.	2.6	3
50	AHDC1 missense mutations in Xia-Gibbs syndrome. Human Genetics and Genomics Advances, 2021, 2, 100049.	1.0	5
51	Transmission event of SARS-CoV-2 delta variant reveals multiple vaccine breakthrough infections. BMC Medicine, 2021, 19, 255.	2.3	137
52	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
53	Hidden biases in germline structural variant detection. Genome Biology, 2021, 22, 347.	3.8	19
54	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

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55	Whole exome sequencing in a large pedigree with DCM identifies a novel mutation in RBM20. Acta Cardiologica, 2020, 75, 748-753.	0.3	8
56	Cohort Profile: The Right Drug, Right Dose, Right Time: Using Genomic Data to Individualize Treatment Protocol (RIGHT Protocol). International Journal of Epidemiology, 2020, 49, 23-24k.	0.9	34
57	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	1.1	58
58	Deficiencies in vesicular transport mediated by TRAPPC4 are associated with severe syndromic intellectual disability. Brain, 2020, 143, 112-130.	3.7	33
59	Biallelic in-frame deletion in TRAPPC4 in a family with developmental delay and cerebellar atrophy. Brain, 2020, 143, e83-e83.	3.7	8
60	NF-ÎºB and STAT3 co-operation enhances high glucose induced aggressiveness of cholangiocarcinoma cells. Life Sciences, 2020, 262, 118548.	2.0	9
61	PCM1 is necessary for focal ciliary integrity and is a candidate for severe schizophrenia. Nature Communications, 2020, 11, 5903.	5.8	13
62	The Human Genome Project changed everything. Nature Reviews Genetics, 2020, 21, 575-576.	7.7	84
63	Loss of the Polyketide Synthase StlB Results in Stalk Cell Overproduction in Polysphondylium violaceum. Genome Biology and Evolution, 2020, 12, 674-683.	1.1	8
64	Genome-enabled insights into the biology of thrips as crop pests. BMC Biology, 2020, 18, 142.	1.7	54
65	High-depth African genomes inform human migration and health. Nature, 2020, 586, 741-748.	13.7	197
66	Phenotypic expansion in KIF1A â€related dominant disorders: A description of novel variants and review of published cases. Human Mutation, 2020, 41, 2094-2104.	1.1	8
67	Communityâ€based recruitment and exome sequencing indicates high diagnostic yield in adults with intellectual disability. Molecular Genetics & Genomic Medicine, 2020, 8, e1439.	0.6	6
68	Sequence analysis in Bos taurus reveals pervasiveness of Xâ€“Y arms races in mammalian lineages. Genome Research, 2020, 30, 1716-1726.	2.4	29
69	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	1.1	25
70	Functional biology of the Steel syndrome founder allele and evidence for clan genomics derivation of COL27A1 pathogenic alleles worldwide. European Journal of Human Genetics, 2020, 28, 1243-1264.	1.4	27
71	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	1.1	36
72	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

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73	<scp>Wolffâ€“Parkinsonâ€“White</scp> syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. American Journal of Medical Genetics, Part A, 2020, 182, 1387-1399.	0.7	14
74	HEM1 deficiency disrupts mTORC2 and F-actin control in inherited immunodysregulatory disease. Science, 2020, 369, 202-207.	6.0	65
75	Gene content evolution in the arthropods. Genome Biology, 2020, 21, 15.	3.8	150
76	Genetics of schizophrenia in the South African Xhosa. Science, 2020, 367, 569-573.	6.0	93
77	Combination of whole exome sequencing and animal modeling identifies TMPRSS9 as a candidate gene for autism spectrum disorder. Human Molecular Genetics, 2020, 29, 459-470.	1.4	32
78	Biallelic GRM7 variants cause epilepsy, microcephaly, and cerebral atrophy. Annals of Clinical and Translational Neurology, 2020, 7, 610-627.	1.7	15
79	Cultivating DNA Sequencing Technology After the Human Genome Project. Annual Review of Genomics and Human Genetics, 2020, 21, 117-138.	2.5	18
80	Sawfly Genomes Reveal Evolutionary Acquisitions That Fostered the Mega-Radiation of Parasitoid and Eusocial Hymenoptera. Genome Biology and Evolution, 2020, 12, 1099-1188.	1.1	17
81	Parliament2: Accurate structural variant calling at scale. GigaScience, 2020, 9, .	3.3	51
82	Disease-associated CTNNBL1 mutation impairs somatic hypermutation by decreasing nuclear AID. Journal of Clinical Investigation, 2020, 130, 4411-4422.	3.9	11
83	Human NK cell deficiency as a result of biallelic mutations in MCM10. Journal of Clinical Investigation, 2020, 130, 5272-5286.	3.9	44
84	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. PLoS Biology, 2020, 18, e3000954.	2.6	73
85	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954.		0
86	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954.		0
87	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954.		0
88	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954.		0
89	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954.		0
90	Primate phylogenomics uncovers multiple rapid radiations and ancient interspecific introgression. , 2020, 18, e3000954.		0

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91	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	1.7	163
92	Biallelic CACNA2D2 variants in epileptic encephalopathy and cerebellar atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1395-1406.	1.7	20
93	ARBoR: an identity and security solution for clinical reporting. Journal of the American Medical Informatics Association: JAMIA, 2019, 26, 1370-1374.	2.2	1
94	Pharmacogenomics of statin-related myopathy: Meta-analysis of rare variants from whole-exome sequencing. PLoS ONE, 2019, 14, e0218115.	1.1	18
95	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
96	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	2.6	56
97	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	2.6	30
98	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	2.6	24
99	Evaluation of computational genotyping of structural variation for clinical diagnoses. GigaScience, 2019, 8, .	3.3	36
100	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
101	The comparative genomics and complex population history of Papio baboons. Science Advances, 2019, 5, eaau6947.	4.7	115
102	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
103	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
104	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	13.9	205
105	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	3.6	42
106	Leveraging Human Microbiome Features to Diagnose and Stratify Children with Irritable Bowel Syndrome. Journal of Molecular Diagnostics, 2019, 21, 449-461.	1.2	59
107	Interchromosomal template-switching as a novel molecular mechanism for imprinting perturbations associated with Temple syndrome. Genome Medicine, 2019, 11, 25.	3.6	22
108	Atlas-CNV: a validated approach to call single-exon CNVs in the eMERGESeq gene panel. Genetics in Medicine, 2019, 21, 2135-2144.	1.1	19

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109	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	3.6	23
110	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	13.5	113
111	Rare variants in SLC5A10 are associated with serum 1,5-anhydroglucitol (1,5-AG) in the Atherosclerosis Risk in Communities (ARIC) Study. Scientific Reports, 2019, 9, 5941.	1.6	9
112	Molecular evolutionary trends and feeding ecology diversification in the Hemiptera, anchored by the milkweed bug genome. Genome Biology, 2019, 20, 64.	3.8	114
113	Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. Cell, 2019, 176, 1310-1324.e10.	13.5	73
114	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	2.6	27
115	Prospective virome analyses in young children at increased genetic risk for type 1 diabetes. Nature Medicine, 2019, 25, 1865-1872.	15.2	161
116	Xiaâ€“Gibbs syndrome in adulthood: a case report with insight into the natural history of the condition. Journal of Physical Education and Sports Management, 2019, 5, a003608.	0.5	15
117	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161
118	Genetic architecture of laterality defects revealed by whole exome sequencing. European Journal of Human Genetics, 2019, 27, 563-573.	1.4	44
119	Functionally oriented analysis of cardiometabolic traits in a trans-ethnic sample. Human Molecular Genetics, 2019, 28, 1212-1224.	1.4	12
120	Genomic Characterization of a Pediatric Cohort with Non-Malignant Lymphoproliferative Disorders. Blood, 2019, 134, 83-83.	0.6	0
121	A biallelic ANTXR1 variant expands the anthrax toxin receptor associated phenotype to tooth agenesis. American Journal of Medical Genetics, Part A, 2018, 176, 1015-1022.	0.7	11
122	Sheep genome functional annotation reveals proximal regulatory elements contributed to the evolution of modern breeds. Nature Communications, 2018, 9, 859.	5.8	126
123	Chemistry-First Approach for Nomination of Personalized Treatment in Lung Cancer. Cell, 2018, 173, 864-878.e29.	13.5	102
124	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
125	The Cancer Genome Atlas Comprehensive Molecular Characterization of Renal Cell Carcinoma. Cell Reports, 2018, 23, 313-326.e5.	2.9	523
126	The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. Human Molecular Genetics, 2018, 27, 2064-2075.	1.4	16

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127	The Toxicogenome of Hyalella azteca: A Model for Sediment Ecotoxicology and Evolutionary Toxicology. Environmental Science & Technology, 2018, 52, 6009-6022.	4.6	79
128	Sequence-Based Analysis of Lipid-Related Metabolites in a Multiethnic Study. Genetics, 2018, 209, 607-616.	1.2	8
129	Hemimetabolous genomes reveal molecular basis of termite eusociality. Nature Ecology and Evolution, 2018, 2, 557-566.	3.4	223
130	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
131	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	2.6	88
132	Mutations in PI3K110Î´ cause impaired natural killer cell function partially rescued by rapamycin treatment. Journal of Allergy and Clinical Immunology, 2018, 142, 605-617.e7.	1.5	36
133	Sooty mangabey genome sequence provides insight into AIDS resistance in a natural SIV host. Nature, 2018, 553, 77-81.	13.7	81
134	The phenotypic spectrum of Xiaâ€Gibbs syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1315-1326.	0.7	34
135	Phenotypic expansion illuminates multilocus pathogenic variation. Genetics in Medicine, 2018, 20, 1528-1537.	1.1	104
136	Comprehensive genomic analysis of patients with disorders of cerebral cortical development. European Journal of Human Genetics, 2018, 26, 1121-1131.	1.4	35
137	MLH1-rheMac hereditary nonpolyposis colorectal cancer syndrome in rhesus macaques. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 2806-2811.	3.3	9
138	Lipoprotein-associated phospholipase A2 and risk of incident peripheral arterial disease: Findings from The Atherosclerosis Risk in Communities study (ARIC). Atherosclerosis, 2018, 268, 12-18.	0.4	13
139	Pleiotropic neuropathological and biochemical alterations associated with Myo5a mutation in a rat Model. Brain Research, 2018, 1679, 155-170.	1.1	14
140	Challenges of Francisella classification exemplified by an atypical clinical isolate. Diagnostic Microbiology and Infectious Disease, 2018, 90, 241-247.	0.8	3
141	Whole-Exome Sequencing Identifies Novel Variants for Tooth Agenesis. Journal of Dental Research, 2018, 97, 49-59.	2.5	44
142	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
143	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
144	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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145	Reproductive Longevity Predicts Mutation Rates in Primates. Current Biology, 2018, 28, 3193-3197.e5.	1.8	94
146	Temporal development of the gut microbiome in early childhood from the TEDDY study. Nature, 2018, 562, 583-588.	13.7	1,220
147	Identification of a pathogenic PMP2 variant in a multi-generational family with CMT type 1: Clinical gene panels versus genome-wide approaches to molecular diagnosis. Molecular Genetics and Metabolism, 2018, 125, 302-304.	0.5	13
148	TBIO-20. CLINICAL TUMOR WHOLE EXOME SEQUENCING FOR PEDIATRIC NEURO-ONCOLOGY PATIENTS â€“ RESULTS FROM THE BAYLOR ADVANCING SEQUENCING IN CHILDHOOD CANCER CARE (BASIC3) CLINICAL SEQUENCING STUDY. Neuro-Oncology, 2018, 20, i184-i184.	0.6	0
149	Common Coding Variants in SCN10A Are Associated With the Nav1.8 Late Current and Cardiac Conduction. Circulation Genomic and Precision Medicine, 2018, 11, e001663.	1.6	26
150	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
151	Identification of likely pathogenic and known variants in TSPEAR, LAMB3, BCOR, and WNT10A in four Turkish families with tooth agenesis. Human Genetics, 2018, 137, 689-703.	1.8	24
152	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
153	Comparative genomics of the miniature wasp and pest control agent Trichogramma pretiosum. BMC Biology, 2018, 16, 54.	1.7	57
154	SYT1-associated neurodevelopmental disorder: a case series. Brain, 2018, 141, 2576-2591.	3.7	98
155	Empowering genomic medicine by establishing critical sequencing result data flows: the eMERGE example. Journal of the American Medical Informatics Association: JAMIA, 2018, 25, 1375-1381.	2.2	21
156	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237.	6.5	98
157	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
158	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
159	Kaufman oculo-cerebro-facial syndrome in a child with small and absent terminal phalanges and absent nails. Journal of Human Genetics, 2017, 62, 465-471.	1.1	14
160	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	13.5	66
161	SeqCNV: a novel method for identification of copy number variations in targeted next-generation sequencing data. BMC Bioinformatics, 2017, 18, 147.	1.2	54
162	Whole-genome landscape of pancreatic neuroendocrine tumours. Nature, 2017, 543, 65-71.	13.7	716

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163	Evolutionary History of Chemosensory-Related Gene Families across the Arthropoda. Molecular Biology and Evolution, 2017, 34, 1838-1862.	3.5	157
164	Exome sequencing reveals novel genetic loci influencing obesityâ€related traits in Hispanic children. Obesity, 2017, 25, 1270-1276.	1.5	10
165	Dual molecular diagnosis contributes to atypical Praderâ€“Willi phenotype in monozygotic twins. American Journal of Medical Genetics, Part A, 2017, 173, 2451-2455.	0.7	26
166	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a TUBB3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a000984.	0.5	18
167	Extremely low-coverage whole genome sequencing in South Asians captures population genomics information. BMC Genomics, 2017, 18, 396.	1.2	26
168	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
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