Evan E Eichler

List of Publications by Year
in descending order

574 papers	168,293 citations	114 161 h-index	60 377 g-index
694 all docs	694 docs citations	694 times ranked	150796 citing authors

#	Article	IF	Citations
1	A 25-year odyssey of genomic technology advances and structural variant discovery. Cell, 2024, 187, 1024-1037.	27.8	12
2	Utility of long-read sequencing for All of Us. Nature Communications, 2024, 15, .	13.2	6
3	Utility of long-read sequencing for All of Us. Nature Communications, 2024, 15, .	13.2	0
4	Personal journeys to and in human genetics and dysmorphology. American Journal of Medical Genetics, Part A, 2024, 194, .	1.5	0
5	Genomic data in the All of Us Research Program. Nature, 2024, 627, 340-346.	36.2	62
6	Structurally divergent and recurrently mutated regions of primate genomes. Cell, 2024, 187, 1547-1562.e13.	27.8	10
7	The variation and evolution of complete human centromeres. Nature, 2024, 629, 136-145.	36.2	14
8	Protein target similarity is positive predictor of in vitro antipathogenic activity: a drug repurposing strategy for Plasmodium falciparum. Journal of Cheminformatics, 2024, 16, .	6.4	0
9	The complete sequence and comparative analysis of ape sex chromosomes. Nature, 2024, 630, 401-411.	36.2	8
10	Structural and genetic diversity in the secreted mucins MUC5AC and MUC5B. American Journal of Human Genetics, 2024, , .	6.1	0
11	Graphasing: phasing diploid genome assembly graphs with single-cell strand sequencing. Genome Biology, 2024, 25, .	9.2	0
12	On the non-intrusive extraction of residentsâ€™ privacy- and security-sensitive information from energy smart meters. Neural Computing and Applications, 2023, 35, 119-132.	5.7	6
13	Stereo-EEG Evaluation and Surgical Treatment in Patients With Drug-Resistant Focal Epilepsy Associated With Nodular Heterotopia. Journal of Clinical Neurophysiology, 2023, 40, 17-26.	1.9	3
14	The effect of schizophrenia risk factors on mismatch responses in a rat model. Psychophysiology, 2023, 60, .	2.6	4
15	A predictive ensemble classifier for the gene expression diagnosis of ASD at ages 1 to 4 years. Molecular Psychiatry, 2023, 28, 822-833.	8.2	4
16	Characterization of the immunoglobulin lambda chain locus from diverse populations reveals extensive genetic variation. Genes and Immunity, 2023, 24, 21-31.	4.3	15
17	A novel DSPP frameshift mutation causing dentin dysplasia type 2 and disease management strategies. Oral Diseases, 2023, 29, 2394-2400.	3.2	2
18	Novel Preoxidation-Assisted Mechanism to Preciously Form and Disperse Bi₂O₃ Nanodots in Carbon Nanofibers for Ultralong-Life and High-Rate Sodium Storage. ACS Applied Materials & Interfaces, 2023, 15, 1891-1902.	8.3	4

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19	The stability of the b-family of peakon equations. Nonlinearity, 2023, 36, 1192-1217.	1.5	4
20	The Dynamic Structure and Rapid Evolution of Human Centromeric Satellite DNA. Genes, 2023, 14, 92.	2.4	5
21	Inversion polymorphism in a complete human genome assembly. Genome Biology, 2023, 24, .	9.2	16
22	A draft human pangenome reference. Nature, 2023, 617, 312-324.	36.2	318
23	Gaps and complex structurally variant loci in phased genome assemblies. Genome Research, 2023, 33, 496-510.	5.6	18
24	PSMC3 proteasome subunit variants are associated with neurodevelopmental delay and type I interferon production. Science Translational Medicine, 2023, 15, .	13.4	15
25	Applications of long-read sequencing to Mendelian genetics. Genome Medicine, 2023, 15, .	8.5	25
26	Increased mutation and gene conversion within human segmental duplications. Nature, 2023, 617, 325-334.	36.2	40
27	Sampling a wide swathe of primate genetic diversity. Cell Genomics, 2023, 3, 100358.	7.1	0
28	Assembly of 43 human Y chromosomes reveals extensive complexity and variation. Nature, 2023, 621, 355-364.	36.2	27
29	The complete sequence of a human Y chromosome. Nature, 2023, 621, 344-354.	36.2	124
30	REFRACTORY NON-HEPATIC HYPERAMMONEMIA PRECIPITATED BY ENTEROVESICAL FISTULA. Chest, 2023, 164, A1956-A1957.	0.9	0
31	Envisioning a new era: Complete genetic information from routine, telomere-to-telomere genomes. American Journal of Human Genetics, 2023, 110, 1832-1840.	6.1	7
32	LINE-1 retrotransposons drive human neuronal transcriptome complexity and functional diversification. Science Advances, 2023, 9, .	10.9	11
33	ELOA3 : A primate-specific RNA polymerase II elongation factor encoded by a tandem repeat gene cluster. Science Advances, 2023, 9, .	10.9	0
34	Whole-genome long-read sequencing downsampling and its effect on variant-calling precision and recall. Genome Research, 2023, 33, 2029-2040.	5.6	3
35	Advances in the discovery and analyses of human tandem repeats. Emerging Topics in Life Sciences, 2023, 7, 361-381.	2.6	7
36	Jin, Jiyan, Azadi and the Historical Erasure of Kurds â€“ ERRATUM. International Journal of Middle East Studies, 2023, 55, 829-829.	0.1	0

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37	Mako: A Graph-Based Pattern Growth Approach to Detect Complex Structural Variants. Genomics, Proteomics and Bioinformatics, 2022, 20, 205-218.	7.5	7
38	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.	11.0	44
39	A family study implicates GBE1 in the etiology of autism spectrum disorder. Human Mutation, 2022, 43, 16-29.	2.8	2
40	Gain-of-function variants in GABRD reveal a novel pathway for neurodevelopmental disorders and epilepsy. Brain, 2022, 145, 1299-1309.	8.0	36
41	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	2.8	7
42	StainedGlass: interactive visualization of massive tandem repeat structures with identity heatmaps. Bioinformatics, 2022, 38, 2049-2051.	4.2	60
43	C. Thomas Caskey (1938â€“2022). Genome Research, 2022, 32, vii-viii.	5.6	1
44	Genes To Mental Health (G2MH): A Framework to Map the Combined Effects of Rare and Common Variants on Dimensions of Cognition and Psychopathology. American Journal of Psychiatry, 2022, 179, 189-203.	8.7	37
45	Epigenetic patterns in a complete human genome. Science, 2022, 376, eabj5089.	20.9	149
46	Complete genomic and epigenetic maps of human centromeres. Science, 2022, 376, eabl4178.	20.9	265
47	From telomere to telomere: The transcriptional and epigenetic state of human repeat elements. Science, 2022, 376, eabk3112.	20.9	180
48	Familial long-read sequencing increases yield of de novo mutations. American Journal of Human Genetics, 2022, 109, 631-646.	6.1	41
49	The complete sequence of a human genome. Science, 2022, 376, 44-53.	20.9	1,565
50	Segmental duplications and their variation in a complete human genome. Science, 2022, 376, eabj6965.	20.9	170
51	Rare variants and the oligogenic architecture of autism. Trends in Genetics, 2022, 38, 895-903.	6.9	16
52	Pangenome-based genome inference allows efficient and accurate genotyping across a wide spectrum of variant classes. Nature Genetics, 2022, 54, 518-525.	20.4	113
53	The Human Pangenome Project: a global resource to map genomic diversity. Nature, 2022, 604, 437-446.	36.2	237
54	Novel biallelic variants affecting the OTU domain of the gene OTUD6B associate with severe intellectual disability syndrome and molecular dynamics simulations. European Journal of Medical Genetics, 2022, 65, 104497.	1.3	2

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55	Recurrent inversion polymorphisms in humans associate with genetic instability and genomic disorders. Cell, 2022, 185, 1986-2005.e26.	27.8	83
56	Unique Geothermal Chemistry Shapes Microbial Communities on Mt. Erebus, Antarctica. Frontiers in Microbiology, 2022, 13, 836943.	3.6	6
57	Targeted long-read sequencing identifies missing pathogenic variants in unsolved Werner syndrome cases. Journal of Medical Genetics, 2022, 59, 1087-1094.	3.6	16
58	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	9
59	What Are Lake Beaches Made of? An Assessment of Plastic Beach Litter on the Shores of Como Bay (Italy). Applied Sciences (Switzerland), 2022, 12, 5388.	2.6	9
60	De novo variants in genes regulating stress granule assembly associate with neurodevelopmental disorders. Science Advances, 2022, 8, .	10.9	17
61	GIGYF1 disruption associates with autism and impaired IGF-1R signaling. Journal of Clinical Investigation, 2022, 132, .	8.2	13
62	Semi-automated assembly of high-quality diploid human reference genomes. Nature, 2022, 611, 519-531.	36.2	98
63	Estimating the Prevalence of De Novo Monogenic Neurodevelopmental Disorders from Large Cohort Studies. Biomedicines, 2022, 10, 2865.	3.3	11
64	Integrated gene analyses of de novo variants from 46,612 trios with autism and developmental disorders. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, .	7.6	25
65	Nitrification inhibition by polyphenols from invasive Fallopia japonica under copper stress. Journal of Plant Nutrition and Soil Science, 2022, 185, 923-934.	2.4	5
66	ANALISIS GAYA MENGAJAR GURU KELAS TINGGI PADA PEMBELAJARAN MENULIS NARASI DI KECAMATAN WADO. Jurnal Administrasi Pendidikan, 2022, 19, 127-138.	0.1	0
67	Brief Report: Associations Between Self-injurious Behaviors and Abdominal Pain Among Individuals with ASD-Associated Disruptive Mutations. Journal of Autism and Developmental Disorders, 2021, 51, 3365-3373.	3.1	5
68	Fully phased human genome assembly without parental data using single-cell strand sequencing and long reads. Nature Biotechnology, 2021, 39, 302-308.	20.8	140
69	Human disease genes website series: An international, open and dynamic library for upâ€toâ€date clinical information. American Journal of Medical Genetics, Part A, 2021, 185, 1039-1046.	1.5	20
70	Synthesis, Structures, and Electrochemical Properties of Bisâ€•and Tetrakis(diphenylphosphino)tetrathiafulvalenes Extended with an Anthraquinoid Spacer. European Journal of Organic Chemistry, 2021, 2021, 1960-1963.	2.5	3
71	2020 William Allan Award introduction: Mary-Claire King. American Journal of Human Genetics, 2021, 108, 383-385.	6.1	0
72	Relationship between Vitamin Intake and Health-Related Quality of Life in a Japanese Population: A Cross-Sectional Analysis of the Shika Study. Nutrients, 2021, 13, 1023.	4.2	9

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73	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.	6.1	52
74	Cancer Cell Intrinsic and Immunologic Phenotypes Determine Clinical Outcomes in Basal-like Breast Cancer. Clinical Cancer Research, 2021, 27, 3079-3093.	7.2	10
75	Integrating buccal and occlusal dental microwear with isotope analyses for a complete paleodietary reconstruction of Holocene populations from Hungary. Scientific Reports, 2021, 11, 7034.	3.4	7
76	Diverse genetic causes of polymicrogyria with epilepsy. Epilepsia, 2021, 62, 973-983.	4.6	16
77	The structure, function and evolution of a complete human chromosome 8. Nature, 2021, 593, 101-107.	36.2	246
78	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.5	57
79	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. Science, 2021, 372, .	20.9	410
80	Extended haplotype-phasing of long-read de novo genome assemblies using Hi-C. Nature Communications, 2021, 12, 1935.	13.2	75
81	Sleep Problems in Children with ASD and Gene Disrupting Mutations. Journal of Genetic Psychology, 2021, 182, 317-334.	1.4	7
82	Expectations and blind spots for structural variation detection from long-read assemblies and short-read genome sequencing technologies. American Journal of Human Genetics, 2021, 108, 919-928.	6.1	81
83	Mining the gaps of chromosome 8. Nature, 2021, , .	36.2	3
84	A high-quality bonobo genome refines the analysis of hominid evolution. Nature, 2021, 594, 77-81.	36.2	47
85	Reflections on the genetics-first approach to advancements in molecular genetic and neurobiological research on neurodevelopmental disorders. Journal of Neurodevelopmental Disorders, 2021, 13, 24.	3.2	17
86	The CHD8/CHD7/Kismet family links blood-brain barrier glia and serotonin to ASD-associated sleep defects. Science Advances, 2021, 7, .	10.9	27
87	Metamaterial-Based Reconfigurable Intelligent Surface: 3D Meta-Atoms Controlled by Graphene Structures. IEEE Communications Magazine, 2021, 59, 42-48.	7.4	41
88	Characterizing nucleotide variation and expansion dynamics in human-specific variable number tandem repeats. Genome Research, 2021, 31, 1313-1324.	5.6	16
89	Recent ultra-rare inherited variants implicate new autism candidate risk genes. Nature Genetics, 2021, 53, 1125-1134.	20.4	79
90	Differences in the number of de novo mutations between individuals are due to small family-specific effects and stochasticity. Genome Research, 2021, 31, 1513-1518.	5.6	7

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91	Extreme Ultraviolet Second Harmonic Generation Spectroscopy in a Polar Metal. Nano Letters, 2021, 21, 6095-6101.	9.5	18
92	Prevalence of Multiple-Level Spondylolysis and the Bone Union Rates among Growth-Stage Children with Lower Back Pain. Spine Surgery and Related Research, 2021, 5, 292-297.	0.8	0
93	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	20.4	53
94	Profiling variable-number tandem repeat variation across populations using repeat-pangenome graphs. Nature Communications, 2021, 12, 4250.	13.2	28
95	Evidence for opposing selective forces operating on human-specific duplicated TCAF genes in Neanderthals and humans. Nature Communications, 2021, 12, 5118.	13.2	16
96	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	6.1	122
97	Alpha Satellite Insertion Close to an Ancestral Centromeric Region. Molecular Biology and Evolution, 2021, 38, 5576-5587.	9.2	4
98	DNA methylation regulates the expression of the negative transcriptional regulators ID2 and ID4 during OPC differentiation. Cellular and Molecular Life Sciences, 2021, 78, 6631-6644.	5.5	27
99	Modelling and application of a new method to measure the non-thermal electron current in the edge of magnetically confined plasma. Nuclear Fusion, 2021, 61, 126004.	3.4	1
100	Comments on â€œA set square design metamaterial absorber for X band applicationsâ€• Journal of Electromagnetic Waves and Applications, 2021, 35, 1020-1024.	1.7	3
101	The relative roles of decadal climate variations and changes in the ocean observing system on seasonal prediction skill of tropical Pacific SST. Climate Dynamics, 2021, 56, 3045-3063.	3.8	6
102	Single-cell epigenomics reveals mechanisms of human cortical development. Nature, 2021, 598, 205-213.	36.2	181
103	Quantitative assessment reveals the dominance of duplicated sequences in germline-derived extrachromosomal circular DNA. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.6	23
104	Pengadaan dan Pemasangan Panel Surya sebagai Sumber Energi Listrik Alternatif untuk Alat Otomatis Pencuci Tangan di Pasar Mitra Raya Kota Batam. Jurnal Pengabdian Kepada Masyarakat Politeknik Negeri Batam, 2021, 3, 77-84.	0.1	0
105	CORRELATION OF ASPARTATE AMINOTRANSFERASE TO PLATELET RATIO INDEX (APRI) WITH THE DEGREE OF SEVERITY LIVER ORGAN IN CIRRHOSIS HEPATIC PATIENTS. Mandala of Health, 2021, 14, 16.	0.1	0
106	Clinical Phenotypes of Carriers of Mutations in CHD8 or Its Conserved Target Genes. Biological Psychiatry, 2020, 87, 123-131.	1.3	25
107	Improved assembly and variant detection of a haploid human genome using singleâ€molecule, highâ€fidelity long reads. Annals of Human Genetics, 2020, 84, 125-140.	0.9	106
108	The Chromosome-Based Rubber Tree Genome Provides New Insights into Spurge Genome Evolution and Rubber Biosynthesis. Molecular Plant, 2020, 13, 336-350.	8.4	83

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109	Geriatric nutrition risk index is associated with renal progression, cardiovascular events and all-cause mortality in chronic kidney disease. Journal of Nephrology, 2020, 33, 783-793.	2.1	21
110	A Novel Framework for Characterizing Genomic Haplotype Diversity in the Human Immunoglobulin Heavy Chain Locus. Frontiers in Immunology, 2020, 11, 2136.	4.9	64
111	High Stroma T-Cell Infiltration is Associated with Better Survival in Stage pT1 Bladder Cancer. International Journal of Molecular Sciences, 2020, 21, 8407.	4.2	16
112	Evolution of a Human-Specific Tandem Repeat Associated with ALS. American Journal of Human Genetics, 2020, 107, 445-460.	6.1	43
113	An evolutionary driver of interspersed segmental duplications in primates. Genome Biology, 2020, 21, 202.	9.2	22
114	De novo SMARCA2 variants clustered outside the helicase domain cause a new recognizable syndrome with intellectual disability and blepharophimosis distinct from Nicolaidesâ€“Baraitser syndrome. Genetics in Medicine, 2020, 22, 1838-1850.	2.4	36
115	Single-cell strand sequencing of a macaque genome reveals multiple nested inversions and breakpoint reuse during primate evolution. Genome Research, 2020, 30, 1680-1693.	5.6	20
116	SDHx and Non-Chromaffin Tumors: A Mediastinal Germ Cell Tumor Occurring in a Young Man with Germline SDHB Mutation. Medicina (Lithuania), 2020, 56, 561.	2.0	4
117	Telomere-to-telomere assembly of a complete human X chromosome. Nature, 2020, 585, 79-84.	36.2	590
118	Developmental Predictors of Cognitive and Adaptive Outcomes in Genetic Subtypes of Autism Spectrum Disorder. Autism Research, 2020, 13, 1659-1669.	3.9	18
119	Large Lemurs: Ecological, Demographic and Environmental Risk Factors for Weight Gain in Captivity. Animals, 2020, 10, 1443.	2.3	5
120	HiCanu: accurate assembly of segmental duplications, satellites, and allelic variants from high-fidelity long reads. Genome Research, 2020, 30, 1291-1305.	5.6	513
121	Sequence diversity analyses of an improved rhesus macaque genome enhance its biomedical utility. Science, 2020, 370, .	20.9	118
122	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. American Journal of Human Genetics, 2020, 107, 963-976.	6.1	25
123	Insufficient Evidence for â€œAutism-Specificâ€•Genes. American Journal of Human Genetics, 2020, 106, 587-595.	6.1	118
124	Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. Nature Biotechnology, 2020, 38, 1044-1053.	20.8	390
125	Mobilising community networks for early identification of tuberculosis and treatment initiation in Cambodia: an evaluation of a seed-and-recruit model. ERJ Open Research, 2020, 6, 00368-2019.	2.7	9
126	Recurrent inversion toggling and great ape genome evolution. Nature Genetics, 2020, 52, 849-858.	20.4	46

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127	Long-read human genome sequencing and its applications. Nature Reviews Genetics, 2020, 21, 597-614.	16.7	629
128	Evaluating heterogeneity in <scp>ASD</scp> symptomatology, cognitive ability, and adaptive functioning among 16p11.2 <scp>CNV</scp> carriers. Autism Research, 2020, 13, 1300-1310.	3.9	26
129	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	2.8	12
130	Evolutionary Dynamics of the POTE Gene Family in Human and Nonhuman Primates. Genes, 2020, 11, 213.	2.4	7
131	A slipped-CAG DNA-binding small molecule induces trinucleotide-repeat contractions in vivo. Nature Genetics, 2020, 52, 146-159.	20.4	116
132	Beliefs in vaccine as causes of autism among SPARK cohort caregivers. Vaccine, 2020, 38, 1794-1803.	4.0	14
133	Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. Nature Communications, 2020, 11, 255.	13.2	51
134	Co-occurring medical conditions among individuals with ASD-associated disruptive mutations. Children's Health Care, 2020, 49, 361-384.	0.9	5
135	The Chromosome-Level Reference Genome of Tea Tree Unveils Recent Bursts of Non-autonomous LTR Retrotransposons in Driving Genome Size Evolution. Molecular Plant, 2020, 13, 935-938.	8.4	96
136	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	13.2	126
137	Model-independent test of the parity symmetry of gravity with gravitational waves. European Physical Journal C, 2020, 80, 1.	4.0	26
138	Fuzzy Decision Algorithm for Driver Drowsiness Detection. , 2020, , 458-467.		3
139	Nutritional value and in situ degradability of oak wood roughage and its feeding effects on growth performance and behavior of Hanwoo steers during the early fattening period. Asian-Australasian Journal of Animal Sciences, 2020, 33, 930-940.	2.5	0
140	Role of Internal Halal Committee in Ensuring Business Sustainability: The Case of a Multinational Slaughter House. Journal of Business Management and Accounting, 2020, 10, 57-65.	0.2	0
141	On Materials Which Allow Students to Find Out Mathematical Propositions Using Snapping on GeoGebra. , 2020, 27, 13-17.		0
142	Computational Engineering. Oberwolfach Reports, 2019, 15, 2859-2913.	0.0	0
143	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	3.9	173
144	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	13.2	165

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145	Evolution of Oryza chloroplast genomes promoted adaptation to diverse ecological habitats. Communications Biology, 2019, 2, 278.	4.5	71
146	Oxidative Stress Levels Induced by Mercury Exposure in Amazon Juvenile Populations in Brazil. International Journal of Environmental Research and Public Health, 2019, 16, 2682.	2.7	18
147	Genetic Variation, Comparative Genomics, and the Diagnosis of Disease. New England Journal of Medicine, 2019, 381, 64-74.	30.1	140
148	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	13.2	48
149	Adaptive archaic introgression of copy number variants and the discovery of previously unknown human genes. Science, 2019, 366, .	20.9	65
150	The Human-Specific BOLA2 Duplication Modifies Iron Homeostasis and Anemia Predisposition in Chromosome 16p11.2 Autism Individuals. American Journal of Human Genetics, 2019, 105, 947-958.	6.1	36
151	Differential gene expression in non-transgenic and transgenic â€œM.26â€•apple overexpressing a peach CBF gene during the transition from eco-dormancy to bud break. Horticulture Research, 2019, 6, 86.	6.5	22
152	Long-read assembly of the Chinese rhesus macaque genome and identification of ape-specific structural variants. Nature Communications, 2019, 10, 4233.	13.2	59
153	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.9	37
154	The comparative genomics and complex population history of Papio baboons. Science Advances, 2019, 5, eaau6947.	10.9	120
155	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
156	Habituation Learning Is a Widely Affected Mechanism in Drosophila Models of Intellectual Disability and Autism Spectrum Disorders. Biological Psychiatry, 2019, 86, 294-305.	1.3	43
157	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	13.2	681
158	Genomic inversions and GOLGA core duplicons underlie disease instability at the 15q25 locus. PLoS Genetics, 2019, 15, e1008075.	3.4	20
159	Molecular Genetic Anatomy and Risk Profile of Hirschsprungâ€™s Disease. New England Journal of Medicine, 2019, 380, 1421-1432.	30.1	134
160	Establishing Cerebral Organoids as Models of Human-Specific Brain Evolution. Cell, 2019, 176, 743-756.e17.	27.8	456
161	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.	6.1	37
162	Minimization of Secrecy Outage Probability with Single-Antenna Uncoordinated Cooperative Jamming. , 2019, , .		1

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163	Human-specific tandem repeat expansion and differential gene expression during primate evolution. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 23243-23253.	7.6	86
164	Sex-Based Analysis of De Novo Variants in Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 1274-1285.	6.1	95
165	The Role of De Novo Noncoding Regulatory Mutations in Neurodevelopmental Disorders. Trends in Neurosciences, 2019, 42, 115-127.	8.8	58
166	Neurodevelopmental disease genes implicated by de novo mutation and copy number variation morbidity. Nature Genetics, 2019, 51, 106-116.	20.4	243
167	Long-read sequence and assembly of segmental duplications. Nature Methods, 2019, 16, 88-94.	19.6	152
168	Characterizing the Major Structural Variant Alleles of the Human Genome. Cell, 2019, 176, 663-675.e19.	27.8	382
169	A report on the impact of remote monitoring in patients with Sâ€ICD: Insights from a prospective registry. PACE - Pacing and Clinical Electrophysiology, 2019, 42, 349-355.	1.2	4
170	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	92
171	Genome sequencing identifies multiple deleterious variants in autism patients with more severe phenotypes. Genetics in Medicine, 2019, 21, 1611-1620.	2.4	91
172	The relationship between interpersonal responsibility and interpersonal trust: A longitudinal study. Current Psychology, 2019, 38, 1182-1189.	2.9	6
173	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	114
174	The novel lncRNA lnc-NR2F1 is pro-neurogenic and mutated in human neurodevelopmental disorders. ELife, 2019, 8, .	5.9	62
175	Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. Cell, 2018, 172, 897-909.e21.	27.8	178
176	Baseline and changes in serum uric acid independently predict 11-year incidence of metabolic syndrome among community-dwelling women. Journal of Endocrinological Investigation, 2018, 41, 959-968.	3.4	16
177	Recurrent structural variation, clustered sites of selection, and disease risk for the complement factor H (CFH) gene family. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E4433-E4442.	7.6	46
178	Longitudinal report of child with de novo 16p11.2 triplication. Clinical Case Reports (discontinued), 2018, 6, 147-154.	0.5	5
179	The sea lamprey germline genome provides insights into programmed genome rearrangement and vertebrate evolution. Nature Genetics, 2018, 50, 270-277.	20.4	266
180	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.1	67

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181	New error bounds for the linear complementarity problem of QN-matrices. Numerical Algorithms, 2018, 77, 229-242.	1.9	12
182	Comorbid symptoms of inattention, autism, and executive cognition in youth with putative genetic risk. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2018, 59, 268-276.	6.2	8
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