Evan E Eichler

List of Publications by Year
in descending order

429 papers	154,777 citations	168 155 h-index	84 365 g-index
485 all docs	485 docs citations	485 times ranked	118402 citing authors

#	Article	IF	Citations
1	Mako: A Graph-based Pattern Growth Approach to Detect Complex Structural Variants. Genomics, Proteomics and Bioinformatics, 2022, 20, 205-218.	6.9	6
2	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.	11.1	37
3	A family study implicates GBE1 in the etiology of autism spectrum disorder. Human Mutation, 2022, 43, 16-29.	2.5	2
4	Gain-of-function variants in GABRD reveal a novel pathway for neurodevelopmental disorders and epilepsy. Brain, 2022, 145, 1299-1309.	7.6	34
5	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	2.5	5
6	StainedGlass: interactive visualization of massive tandem repeat structures with identity heatmaps. Bioinformatics, 2022, 38, 2049-2051.	4.1	42
7	C. Thomas Caskey (1938â€“2022). Genome Research, 2022, 32, vii-viii.	5.5	1
8	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.	7.2	29
9	Epigenetic patterns in a complete human genome. Science, 2022, 376, eabj5089.	12.6	118
10	Complete genomic and epigenetic maps of human centromeres. Science, 2022, 376, eabl4178.	12.6	204
11	From telomere to telomere: The transcriptional and epigenetic state of human repeat elements. Science, 2022, 376, eabk3112.	12.6	146
12	Familial long-read sequencing increases yield of de novo mutations. American Journal of Human Genetics, 2022, 109, 631-646.	6.2	32
13	The complete sequence of a human genome. Science, 2022, 376, 44-53.	12.6	1,222
14	Segmental duplications and their variation in a complete human genome. Science, 2022, 376, eabj6965.	12.6	130
15	Rare variants and the oligogenic architecture of autism. Trends in Genetics, 2022, 38, 895-903.	6.7	14
16	Pangenome-based genome inference allows efficient and accurate genotyping across a wide spectrum of variant classes. Nature Genetics, 2022, 54, 518-525.	21.4	92
17	The Human Pangenome Project: a global resource to map genomic diversity. Nature, 2022, 604, 437-446.	27.8	192
18	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	1

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19	Recurrent inversion polymorphisms in humans associate with genetic instability and genomic disorders. Cell, 2022, 185, 1986-2005.e26.	28.9	67
20	Targeted long-read sequencing identifies missing pathogenic variants in unsolved Werner syndrome cases. Journal of Medical Genetics, 2022, 59, 1087-1094.	3.2	14
21	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	6
22	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.	2.7	5
23	Fully phased human genome assembly without parental data using single-cell strand sequencing and long reads. Nature Biotechnology, 2021, 39, 302-308.	17.5	127
24	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.	1.2	19
25	2020 William Allan Award introduction: Mary-Claire King. American Journal of Human Genetics, 2021, 108, 383-385.	6.2	0
26	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.	6.2	48
27	Diverse genetic causes of polymicrogyria with epilepsy. Epilepsia, 2021, 62, 973-983.	5.1	12
28	The structure, function and evolution of a complete human chromosome 8. Nature, 2021, 593, 101-107.	27.8	221
29	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
30	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. Science, 2021, 372, .	12.6	358
31	Extended haplotype-phasing of long-read de novo genome assemblies using Hi-C. Nature Communications, 2021, 12, 1935.	12.8	64
32	Sleep Problems in Children with ASD and Gene Disrupting Mutations. Journal of Genetic Psychology, 2021, 182, 317-334.	1.2	3
33	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.	6.2	72
34	Mining the gaps of chromosome 8. Nature, 2021, , .	27.8	3
35	A high-quality bonobo genome refines the analysis of hominid evolution. Nature, 2021, 594, 77-81.	27.8	39
36	Reflections on the genetics-first approach to advancements in molecular genetic and neurobiological research on neurodevelopmental disorders. Journal of Neurodevelopmental Disorders, 2021, 13, 24.	3.1	12

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37	The CHD8/CHD7/Kismet family links blood-brain barrier glia and serotonin to ASD-associated sleep defects. Science Advances, 2021, 7, .	10.3	24
38	Characterizing nucleotide variation and expansion dynamics in human-specific variable number tandem repeats. Genome Research, 2021, 31, 1313-1324.	5.5	15
39	Recent ultra-rare inherited variants implicate new autism candidate risk genes. Nature Genetics, 2021, 53, 1125-1134.	21.4	68
40	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.	5.5	6
41	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
42	Profiling variable-number tandem repeat variation across populations using repeat-pangenome graphs. Nature Communications, 2021, 12, 4250.	12.8	27
43	Evidence for opposing selective forces operating on human-specific duplicated TCAF genes in Neanderthals and humans. Nature Communications, 2021, 12, 5118.	12.8	14
44	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	6.2	105
45	Alpha Satellite Insertion Close to an Ancestral Centromeric Region. Molecular Biology and Evolution, 2021, 38, 5576-5587.	8.9	4
46	Single-cell epigenomics reveals mechanisms of human cortical development. Nature, 2021, 598, 205-213.	27.8	154
47	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, .	7.1	18
48	Clinical Phenotypes of Carriers of Mutations in CHD8 or Its Conserved Target Genes. Biological Psychiatry, 2020, 87, 123-131.	1.3	22
49	Phenotypeâ€toâ€genotype approach reveals headâ€circumferenceâ€associated genes in an autism spectrum disorder cohort. Clinical Genetics, 2020, 97, 338-346.	2.0	29
50	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.	0.8	100
51	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	2.4	24
52	The Chromosome-Based Rubber Tree Genome Provides New Insights into Spurge Genome Evolution and Rubber Biosynthesis. Molecular Plant, 2020, 13, 336-350.	8.3	73
53	A Novel Framework for Characterizing Genomic Haplotype Diversity in the Human Immunoglobulin Heavy Chain Locus. Frontiers in Immunology, 2020, 11, 2136.	4.8	54
54	Evolution of a Human-Specific Tandem Repeat Associated with ALS. American Journal of Human Genetics, 2020, 107, 445-460.	6.2	39

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55	An evolutionary driver of interspersed segmental duplications in primates. Genome Biology, 2020, 21, 202.	8.8	19
56	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	31
57	Single-cell strand sequencing of a macaque genome reveals multiple nested inversions and breakpoint reuse during primate evolution. Genome Research, 2020, 30, 1680-1693.	5.5	16
58	Telomere-to-telomere assembly of a complete human X chromosome. Nature, 2020, 585, 79-84.	27.8	549
59	Developmental Predictors of Cognitive and Adaptive Outcomes in Genetic Subtypes of Autism Spectrum Disorder. Autism Research, 2020, 13, 1659-1669.	3.8	13
60	HiCanu: accurate assembly of segmental duplications, satellites, and allelic variants from high-fidelity long reads. Genome Research, 2020, 30, 1291-1305.	5.5	440
61	Sequence diversity analyses of an improved rhesus macaque genome enhance its biomedical utility. Science, 2020, 370, .	12.6	105
62	NCKAP1 Disruptive Variants Lead to a Neurodevelopmental Disorder with Core Features of Autism. American Journal of Human Genetics, 2020, 107, 963-976.	6.2	18
63	Insufficient Evidence for â€œAutism-Specificâ€•Genes. American Journal of Human Genetics, 2020, 106, 587-595.	6.2	110
64	Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. Nature Biotechnology, 2020, 38, 1044-1053.	17.5	344
65	Recurrent inversion toggling and great ape genome evolution. Nature Genetics, 2020, 52, 849-858.	21.4	40
66	Long-read human genome sequencing and its applications. Nature Reviews Genetics, 2020, 21, 597-614.	16.3	542
67	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.	3.8	23
68	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	2.5	11
69	Evolutionary Dynamics of the POTE Gene Family in Human and Nonhuman Primates. Genes, 2020, 11, 213.	2.4	7
70	A slipped-CAG DNA-binding small molecule induces trinucleotide-repeat contractions in vivo. Nature Genetics, 2020, 52, 146-159.	21.4	110
71	Beliefs in vaccine as causes of autism among SPARK cohort caregivers. Vaccine, 2020, 38, 1794-1803.	3.8	12
72	Rare copy number variants in over 100,000 European ancestry subjects reveal multiple disease associations. Nature Communications, 2020, 11, 255.	12.8	48

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73	Co-occurring medical conditions among individuals with ASD-associated disruptive mutations. Children's Health Care, 2020, 49, 361-384.	0.9	3
74	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.	8.3	80
75	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
76	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	3.8	163
77	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
78	Evolution of Oryza chloroplast genomes promoted adaptation to diverse ecological habitats. Communications Biology, 2019, 2, 278.	4.4	62
79	Genetic Variation, Comparative Genomics, and the Diagnosis of Disease. New England Journal of Medicine, 2019, 381, 64-74.	27.0	127
80	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
81	Adaptive archaic introgression of copy number variants and the discovery of previously unknown human genes. Science, 2019, 366, .	12.6	65
82	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.	6.2	30
83	Long-read assembly of the Chinese rhesus macaque genome and identification of ape-specific structural variants. Nature Communications, 2019, 10, 4233.	12.8	54
84	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
85	The comparative genomics and complex population history of Papio baboons. Science Advances, 2019, 5, eaau6947.	10.3	115
86	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
87	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	39
88	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	12.8	636
89	Genomic inversions and GOLGA core duplicons underlie disease instability at the 15q25 locus. PLoS Genetics, 2019, 15, e1008075.	3.5	17
90	Molecular Genetic Anatomy and Risk Profile of Hirschsprungâ€™s Disease. New England Journal of Medicine, 2019, 380, 1421-1432.	27.0	131

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91	Establishing Cerebral Organoids as Models of Human-Specific Brain Evolution. Cell, 2019, 176, 743-756.e17.	28.9	423
92	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.	6.2	30
93	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.	7.1	82
94	Sex-Based Analysis of De Novo Variants in Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 1274-1285.	6.2	84
95	The Role of De Novo Noncoding Regulatory Mutations in Neurodevelopmental Disorders. Trends in Neurosciences, 2019, 42, 115-127.	8.6	56
96	Neurodevelopmental disease genes implicated by de novo mutation and copy number variation morbidity. Nature Genetics, 2019, 51, 106-116.	21.4	231
97	Long-read sequence and assembly of segmental duplications. Nature Methods, 2019, 16, 88-94.	19.0	139
98	Characterizing the Major Structural Variant Alleles of the Human Genome. Cell, 2019, 176, 663-675.e19.	28.9	364
99	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
100	Genome sequencing identifies multiple deleterious variants in autism patients with more severe phenotypes. Genetics in Medicine, 2019, 21, 1611-1620.	2.4	88
101	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
102	The novel lncRNA lnc-NR2F1 is pro-neurogenic and mutated in human neurodevelopmental disorders. ELife, 2019, 8, .	6.0	59
103	Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. Cell, 2018, 172, 897-909.e21.	28.9	163
104	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.	7.1	43
105	Longitudinal report of child with de novo 16p11.2 triplication. Clinical Case Reports (discontinued), 2018, 6, 147-154.	0.5	5
106	The sea lamprey germline genome provides insights into programmed genome rearrangement and vertebrate evolution. Nature Genetics, 2018, 50, 270-277.	21.4	262
107	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.	6.2	59
108	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.	5.2	8

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109	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	2.8	32
110	Inherited and multiple de novo mutations in autism/developmental delay risk genes suggest a multifactorial model. Molecular Autism, 2018, 9, 64.	4.9	114
111	Transcriptional fates of human-specific segmental duplications in brain. Genome Research, 2018, 28, 1566-1576.	5.5	54
112	Inversion variants in human and primate genomes. Genome Research, 2018, 28, 910-920.	5.5	34
113	Human-Specific NOTCH2NL Genes Affect Notch Signaling and Cortical Neurogenesis. Cell, 2018, 173, 1356-1369.e22.	28.9	366
114	Evolutionary history and adaptation of a human pygmy population of Flores Island, Indonesia. Science, 2018, 361, 511-516.	12.6	56
115	Strand-seq enables reliable separation of long reads by chromosome via expectation maximization. Bioinformatics, 2018, 34, i115-i123.	4.1	24
116	The autism spectrum phenotype in ADNP syndrome. Autism Research, 2018, 11, 1300-1310.	3.8	49
117	High-resolution comparative analysis of great ape genomes. Science, 2018, 360, .	12.6	304
118	Comparative Annotation Toolkit (CAT)â€"simultaneous clade and personal genome annotation. Genome Research, 2018, 28, 1029-1038.	5.5	86
119	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
120	The evolution and population diversity of human-specific segmental duplications. Nature Ecology and Evolution, 2017, 1, 69.	7.8	123
121	Epigenetic origin of evolutionary novel centromeres. Scientific Reports, 2017, 7, 41980.	3.3	30
122	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
123	Evaluation of GRCh38 and de novo haploid genome assemblies demonstrates the enduring quality of the reference assembly. Genome Research, 2017, 27, 849-864.	5.5	728
124	Resolving Multicopy Duplications de novo Using Polyploid Phasing. Lecture Notes in Computer Science, 2017, 10229, 117-133.	1.3	22
125	The Tea Tree Genome Provides Insights into Tea Flavor and Independent Evolution of Caffeine Biosynthesis. Molecular Plant, 2017, 10, 866-877.	8.3	563
126	The caterpillar fungus, Ophiocordyceps sinensis, genome provides insights into highland adaptation of fungal pathogenicity. Scientific Reports, 2017, 7, 1806.	3.3	49

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127	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
128	Centromere Destiny in Dicentric Chromosomes: New Insights from the Evolution of Human Chromosome 2 Ancestral Centromeric Region. Molecular Biology and Evolution, 2017, 34, 1669-1681.	8.9	15
129	Sequencing of sporadic Attentionâ€Deficit Hyperactivity Disorder (ADHD) identifies novel and potentially pathogenic de novo variants and excludes overlap with genes associated with autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 381-389.	1.7	44
130	The birth of a human-specific neural gene by incomplete duplication and gene fusion. Genome Biology, 2017, 18, 49.	8.8	39
131	denovo-db: a compendium of humande novovariants. Nucleic Acids Research, 2017, 45, D804-D811.	14.5	173
132	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
133	A high-coverage Neandertal genome from Vindija Cave in Croatia. Science, 2017, 358, 655-658.	12.6	501
134	Genomic Patterns of De Novo Mutation in Simplex Autism. Cell, 2017, 171, 710-722.e12.	28.9	308
135	Exploring the heterogeneity of neural social indices for genetically distinct etiologies of autism. Journal of Neurodevelopmental Disorders, 2017, 9, 24.	3.1	19
136	Discovery of large genomic inversions using long range information. BMC Genomics, 2017, 18, 65.	2.8	18
137	Discovery and genotyping of structural variation from long-read haploid genome sequence data. Genome Research, 2017, 27, 677-685.	5.5	323
138	A 3-way hybrid approach to generate a new high-quality chimpanzee reference genome (Pan_tro_3.0). GigaScience, 2017, 6, 1-6.	6.4	17
139	Recurrent de novo mutations in neurodevelopmental disorders: properties and clinical implications. Genome Medicine, 2017, 9, 101.	8.2	112
140	Targeted Capture and High-Throughput Sequencing Using Molecular Inversion Probes (MIPs). Methods in Molecular Biology, 2017, 1492, 95-106.	0.9	17
141	Large Deletions at the SHOX Locus in the Pseudoautosomal Region Are Associated with Skeletal Atavism in Shetland Ponies. G3: Genes, Genomes, Genetics, 2016, 6, 2213-2223.	1.8	29
142	Long-read sequence assembly of the gorilla genome. Science, 2016, 352, aae0344.	12.6	368
143	An Incomplete Understanding of Human Genetic Variation. Genetics, 2016, 202, 1251-1254.	2.9	78
144	Interchromosomal core duplicons drive both evolutionary instability and disease susceptibility of the Chromosome 8p23.1 region. Genome Research, 2016, 26, 1453-1467.	5.5	37

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145	Emergence of a Homo sapiens-specific gene family and chromosome 16p11.2 CNV susceptibility. Nature, 2016, 536, 205-209.	27.8	102
146	Human adaptation and evolution by segmental duplication. Current Opinion in Genetics and Development, 2016, 41, 44-52.	3.3	157
147	De novo genic mutations among a Chinese autism spectrum disorder cohort. Nature Communications, 2016, 7, 13316.	12.8	293
148	A high-quality human reference panel reveals the complexity and distribution of genomic structural variants. Nature Communications, 2016, 7, 12989.	12.8	99
149	Long-read sequencing and de novo assembly of a Chinese genome. Nature Communications, 2016, 7, 12065.	12.8	242
150	Brain white matter structure and COMT gene are linked to second-language learning in adults. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 7249-7254.	7.1	66
151	Molecular subtyping and improved treatment of neurodevelopmental disease. Genome Medicine, 2016, 8, 22.	8.2	17
152	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in Drosophila. European Journal of Human Genetics, 2016, 24, 1145-1153.	2.8	34
153	TRIOloss of function is associated with mild intellectual disability and affects dendritic branching and synapse function. Human Molecular Genetics, 2016, 25, 892-902.	2.9	94
154	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
155	Maternal Modifiers and Parent-of-Origin Bias of the Autism-Associated 16p11.2 CNV. American Journal of Human Genetics, 2016, 98, 45-57.	6.2	55
156	Genome Sequencing of Autism-Affected Families Reveals Disruption of Putative Noncoding Regulatory DNA. American Journal of Human Genetics, 2016, 98, 58-74.	6.2	248
157	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	2.8	108
158	De Novo and Rare Variants at Multiple Loci Support the Oligogenic Origins of Atrioventricular Septal Heart Defects. PLoS Genetics, 2016, 12, e1005963.	3.5	92
159	Copy number variant analysis from exome data in 349 patients with epileptic encephalopathy. Annals of Neurology, 2015, 78, 323-328.	5.3	59
160	Recurrent duplications of 17q12 associated with variable phenotypes. American Journal of Medical Genetics, Part A, 2015, 167, 3038-3045.	1.2	22
161	BovineNK-lysin: Copy number variation and functional diversification. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E7223-9.	7.1	54
162	B56Î´-related protein phosphatase 2A dysfunction identified in patients with intellectual disability. Journal of Clinical Investigation, 2015, 125, 3051-3062.	8.2	91

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163	Extreme selective sweeps independently targeted the X chromosomes of the great apes. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 6413-6418.	7.1	75
164	Genome-wide association study and admixture mapping reveal new loci associated with total IgE levels in Latinos. Journal of Allergy and Clinical Immunology, 2015, 135, 1502-1510.	2.9	52
165	ADCY5 -related dyskinesia. Neurology, 2015, 85, 2026-2035.	1.1	163
166	Sequencing of the human IG light chain loci from a hydatidiform mole BAC library reveals locus-specific signatures of genetic diversity. Genes and Immunity, 2015, 16, 24-34.	4.1	43
167	MIPSTR: a method for multiplex genotyping of germline and somatic STR variation across many individuals. Genome Research, 2015, 25, 750-761.	5.5	37
168	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
169	Global diversity, population stratification, and selection of human copy-number variation. Science, 2015, 349, aab3761.	12.6	293
170	Characteristics of de novo structural changes in the human genome. Genome Research, 2015, 25, 792-801.	5.5	115
171	Excess of rare, inherited truncating mutations in autism. Nature Genetics, 2015, 47, 582-588.	21.4	531
172	Epigenetics of Autism-related Impairment. Journal of Developmental and Behavioral Pediatrics, 2015, 36, 61-67.	1.1	83
173	Copy-Number Variation and False Positive Prenatal Aneuploidy Screening Results. New England Journal of Medicine, 2015, 372, 1639-1645.	27.0	118
174	Mountain gorilla genomes reveal the impact of long-term population decline and inbreeding. Science, 2015, 348, 242-245.	12.6	326
175	Genotype-First Analysis of the 16p11.2 Deletion Defines a New Type of â€œAutismâ€• Biological Psychiatry, 2015, 77, 769-771.	1.3	15
176	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	27.8	13,998
177	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	27.8	1,994
178	Genetic variation and the de novo assembly of human genomes. Nature Reviews Genetics, 2015, 16, 627-640.	16.3	310
179	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	2.4	45
180	The discovery of integrated gene networks for autism and related disorders. Genome Research, 2015, 25, 142-154.	5.5	259

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181	Resolving the complexity of the human genome using single-molecule sequencing. Nature, 2015, 517, 608-611.	27.8	714
182	Whole-Genome Sequencing of Individuals from a Founder Population Identifies Candidate Genes for Asthma. PLoS ONE, 2014, 9, e104396.	2.5	42
183	mrsFAST-Ultra: a compact, SNP-aware mapper for high performance sequencing applications. Nucleic Acids Research, 2014, 42, W494-W500.	14.5	54
184	An inherited duplication at the gene p21 Protein-Activated Kinase 7 (PAK7) is a risk factor for psychosis. Human Molecular Genetics, 2014, 23, 3316-3326.	2.9	37
185	Genomic studies in fragile X premutation carriers. Journal of Neurodevelopmental Disorders, 2014, 6, 27.	3.1	24
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