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	Initial sequencing and analysis of the human genome. Nature, 2001, 409, 860-921.	27.8	21,074
2	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	27.8	13,998
3	Finding the missing heritability of complex diseases. Nature, 2009, 461, 747-753.	27.8	7,490
4	Nonhybrid, finished microbial genome assemblies from long-read SMRT sequencing data. Nature Methods, 2013, 10, 563-569.	19.0	4,029
5	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	8.1	3,833
6	A Draft Sequence of the Neandertal Genome. Science, 2010, 328, 710-722.	12.6	3,588
7	Consensus Statement: Chromosomal Microarray Is a First-Tier Clinical Diagnostic Test for Individuals with Developmental Disabilities or Congenital Anomalies. American Journal of Human Genetics, 2010, 86, 749-764.	6.2	2,325
8	The contribution of de novo coding mutations to autism spectrum disorder. Nature, 2014, 515, 216-221.	27.8	2,188
9	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	27.8	1,994
10	Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. Nature, 2012, 485, 246-250.	27.8	1,960
11	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. Nature, 2004, 428, 493-521.	27.8	1,943
12	The complete genome sequence of a Neanderthal from the Altai Mountains. Nature, 2014, 505, 43-49.	27.8	1,830
13	Targeted capture and massively parallel sequencing of 12 human exomes. Nature, 2009, 461, 272-276.	27.8	1,801
14	A High-Coverage Genome Sequence from an Archaic Denisovan Individual. Science, 2012, 338, 222-226.	12.6	1,695
15	Rare Structural Variants Disrupt Multiple Genes in Neurodevelopmental Pathways in Schizophrenia. Science, 2008, 320, 539-543.	12.6	1,654
16	Genetic history of an archaic hominin group from Denisova Cave in Siberia. Nature, 2010, 468, 1053-1060.	27.8	1,537
17	Missing heritability and strategies for finding the underlying causes of complex disease. Nature Reviews Genetics, 2010, 11, 446-450.	16.3	1,511
18	A comparative encyclopedia of DNA elements in the mouse genome. Nature, 2014, 515, 355-364.	27.8	1,444

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19	De novo mutations in epileptic encephalopathies. Nature, 2013, 501, 217-221.	27.8	1,351
20	Evolutionary and Biomedical Insights from the Rhesus Macaque Genome. Science, 2007, 316, 222-234.	12.6	1,283
21	Genome structural variation discovery and genotyping. Nature Reviews Genetics, 2011, 12, 363-376.	16.3	1,240
22	Recent Segmental Duplications in the Human Genome. Science, 2002, 297, 1003-1007.	12.6	1,238
23	The complete sequence of a human genome. Science, 2022, 376, 44-53.	12.6	1,222
24	Ancient human genomes suggest three ancestral populations for present-day Europeans. Nature, 2014, 513, 409-413.	27.8	1,179
25	A copy number variation morbidity map of developmental delay. Nature Genetics, 2011, 43, 838-846.	21.4	1,141
26	Multiplex Targeted Sequencing Identifies Recurrently Mutated Genes in Autism Spectrum Disorders. Science, 2012, 338, 1619-1622.	12.6	1,133
27	Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. Nature Genetics, 2011, 43, 585-589.	21.4	1,080
28	The Genome Sequence of Taurine Cattle: A Window to Ruminant Biology and Evolution. Science, 2009, 324, 522-528.	12.6	1,038
29	Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65.	27.8	991
30	Mapping and sequencing of structural variation from eight human genomes. Nature, 2008, 453, 56-64.	27.8	983
31	Fine-scale structural variation of the human genome. Nature Genetics, 2005, 37, 727-732.	21.4	897
32	Segmental Duplications and Copy-Number Variation in the Human Genome. American Journal of Human Genetics, 2005, 77, 78-88.	6.2	872
33	The genome of a songbird. Nature, 2010, 464, 757-762.	27.8	770
34	Great ape genetic diversity and population history. Nature, 2013, 499, 471-475.	27.8	768
35	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
36	Resolving the complexity of the human genome using single-molecule sequencing. Nature, 2015, 517, 608-611.	27.8	714

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37	Limitations of next-generation genome sequence assembly. Nature Methods, 2011, 8, 61-65.	19.0	685
38	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	27.0	663
39	Insights into hominid evolution from the gorilla genome sequence. Nature, 2012, 483, 169-175.	27.8	663
40	Genome analysis of the platypus reveals unique signatures of evolution. Nature, 2008, 453, 175-183.	27.8	657
41	Personalized copy number and segmental duplication maps using next-generation sequencing. Nature Genetics, 2009, 41, 1061-1067.	21.4	656
42	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	28.9	637
43	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	12.8	636
44	Diversity of Human Copy Number Variation and Multicopy Genes. Science, 2010, 330, 641-646.	12.6	609
45	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	21.4	583
46	The Tea Tree Genome Provides Insights into Tea Flavor and Independent Evolution of Caffeine Biosynthesis. Molecular Plant, 2017, 10, 866-877.	8.3	563
47	Discovery of previously unidentified genomic disorders from the duplication architecture of the human genome. Nature Genetics, 2006, 38, 1038-1042.	21.4	557
48	Copy number variation detection and genotyping from exome sequence data. Genome Research, 2012, 22, 1525-1532.	5.5	550
49	Telomere-to-telomere assembly of a complete human X chromosome. Nature, 2020, 585, 79-84.	27.8	549
50	Long-read human genome sequencing and its applications. Nature Reviews Genetics, 2020, 21, 597-614.	16.3	542
51	Comparative and demographic analysis of orang-utan genomes. Nature, 2011, 469, 529-533.	27.8	541
52	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	21.4	539
53	LINE-1 Retrotransposition Activity in Human Genomes. Cell, 2010, 141, 1159-1170.	28.9	531
54	Excess of rare, inherited truncating mutations in autism. Nature Genetics, 2015, 47, 582-588.	21.4	531

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55	Population Analysis of Large Copy Number Variants and Hotspots of Human Genetic Disease. American Journal of Human Genetics, 2009, 84, 148-161.	6.2	530
56	Phenotypic Heterogeneity of Genomic Disorders and Rare Copy-Number Variants. New England Journal of Medicine, 2012, 367, 1321-1331.	27.0	519
57	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	21.4	511
58	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	21.4	509
59	A high-coverage Neandertal genome from Vindija Cave in Croatia. Science, 2017, 358, 655-658.	12.6	501
60	Primate segmental duplications: crucibles of evolution, diversity and disease. Nature Reviews Genetics, 2006, 7, 552-564.	16.3	498
61	Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. Nature Genetics, 2009, 41, 211-215.	21.4	482
62	A Comprehensive Analysis of Common Copy-Number Variations in the Human Genome. American Journal of Human Genetics, 2007, 80, 91-104.	6.2	471
63	Length of uninterrupted CGG repeats determines instability in the FMR1 gene. Nature Genetics, 1994, 8, 88-94.	21.4	468
64	Modernizing Reference Genome Assemblies. PLoS Biology, 2011, 9, e1001091.	5.6	458
65	A Higher Mutational Burden in Females Supports a â€œFemale Protective Modelâ€•in Neurodevelopmental Disorders. American Journal of Human Genetics, 2014, 94, 415-425.	6.2	457
66	Sequence and structural variation in a human genome uncovered by short-read, massively parallel ligation sequencing using two-base encoding. Genome Research, 2009, 19, 1527-1541.	5.5	448
67	The bonobo genome compared with the chimpanzee and human genomes. Nature, 2012, 486, 527-531.	27.8	445
68	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
69	HiCanu: accurate assembly of segmental duplications, satellites, and allelic variants from high-fidelity long reads. Genome Research, 2020, 30, 1291-1305.	5.5	440
70	Establishing Cerebral Organoids as Models of Human-Specific Brain Evolution. Cell, 2019, 176, 743-756.e17.	28.9	423
71	Segmental Duplications: Organization and Impact Within the Current Human Genome Project Assembly. Genome Research, 2001, 11, 1005-1017.	5.5	423
72	Lineage-Specific Biology Revealed by a Finished Genome Assembly of the Mouse. PLoS Biology, 2009, 7, e1000112.	5.6	419

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73	Genome-Wide Copy Number Variation in Epilepsy: Novel Susceptibility Loci in Idiopathic Generalized and Focal Epilepsies. PLoS Genetics, 2010, 6, e1000962.	3.5	414
74	A de novo convergence of autism genetics and molecular neuroscience. Trends in Neurosciences, 2014, 37, 95-105.	8.6	410
75	Recurrent microdeletions at 15q11.2 and 16p13.11 predispose to idiopathic generalized epilepsies. Brain, 2010, 133, 23-32.	7.6	406
76	Complete Khoisan and Bantu genomes from southern Africa. Nature, 2010, 463, 943-947.	27.8	400
77	An Alu Transposition Model for the Origin and Expansion of Human Segmental Duplications. American Journal of Human Genetics, 2003, 73, 823-834.	6.2	387
78	Segmental duplications and the evolution of the primate genome. Nature Reviews Genetics, 2002, 3, 65-72.	16.3	374
79	Long-read sequence assembly of the gorilla genome. Science, 2016, 352, aae0344.	12.6	368
80	Human-Specific NOTCH2NL Genes Affect Notch Signaling and Cortical Neurogenesis. Cell, 2018, 173, 1356-1369.e22.	28.9	366
81	De Novo Pathogenic SCN8A Mutation Identified by Whole-Genome Sequencing of a Family Quartet Affected by Infantile Epileptic Encephalopathy and SUDEP. American Journal of Human Genetics, 2012, 90, 502-510.	6.2	365
82	Characterizing the Major Structural Variant Alleles of the Human Genome. Cell, 2019, 176, 663-675.e19.	28.9	364
83	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. Science, 2021, 372, .	12.6	358
84	Structural Dynamics of Eukaryotic Chromosome Evolution. Science, 2003, 301, 793-797.	12.6	357
85	A genome-wide comparison of recent chimpanzee and human segmental duplications. Nature, 2005, 437, 88-93.	27.8	353
86	Noninvasive Whole-Genome Sequencing of a Human Fetus. Science Translational Medicine, 2012, 4, 137ra76.	12.4	348
87	Human Copy Number Variation and Complex Genetic Disease. Annual Review of Genetics, 2011, 45, 203-226.	7.6	344
88	Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. Nature Biotechnology, 2020, 38, 1044-1053.	17.5	344
89	Evolution of Human-Specific Neural SRGAP2 Genes by Incomplete Segmental Duplication. Cell, 2012, 149, 912-922.	28.9	341
90	Challenges and standards in integrating surveys of structural variation. Nature Genetics, 2007, 39, S7-S15.	21.4	331

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91	Mountain gorilla genomes reveal the impact of long-term population decline and inbreeding. Science, 2015, 348, 242-245.	12.6	326
92	Discovery and genotyping of structural variation from long-read haploid genome sequence data. Genome Research, 2017, 27, 677-685.	5.5	323
93	Gibbon genome and the fast karyotype evolution of small apes. Nature, 2014, 513, 195-201.	27.8	320
94	Regional Patterns of Gene Expression in Human and Chimpanzee Brains. Genome Research, 2004, 14, 1462-1473.	5.5	311
95	Genetic variation and the de novo assembly of human genomes. Nature Reviews Genetics, 2015, 16, 627-640.	16.3	310
96	Genomic Patterns of De Novo Mutation in Simplex Autism. Cell, 2017, 171, 710-722.e12.	28.9	308
97	High-resolution comparative analysis of great ape genomes. Science, 2018, 360, .	12.6	304
98	The DNA sequence and biology of human chromosome 19. Nature, 2004, 428, 529-535.	27.8	298
99	Recent duplication, domain accretion and the dynamic mutation of the human genome. Trends in Genetics, 2001, 17, 661-669.	6.7	297
100	Relative Burden of Large CNVs on a Range of Neurodevelopmental Phenotypes. PLoS Genetics, 2011, 7, e1002334.	3.5	293
101	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	21.4	293
102	Global diversity, population stratification, and selection of human copy-number variation. Science, 2015, 349, aab3761.	12.6	293
103	De novo genic mutations among a Chinese autism spectrum disorder cohort. Nature Communications, 2016, 7, 13316.	12.8	293
104	Positive selection of a gene family during the emergence of humans and African apes. Nature, 2001, 413, 514-519.	27.8	284
105	Linkage Disequilibrium and Heritability of Copy-Number Polymorphisms within Duplicated Regions of the Human Genome. American Journal of Human Genetics, 2006, 79, 275-290.	6.2	283
106	Analysis of copy number variations among diverse cattle breeds. Genome Research, 2010, 20, 693-703.	5.5	280
107	Refinement and Discovery of New Hotspots of Copy-Number Variation Associated with Autism Spectrum Disorder. American Journal of Human Genetics, 2013, 92, 221-237.	6.2	279
108	Estimates of penetrance for recurrent pathogenic copy-number variations. Genetics in Medicine, 2013, 15, 478-481.	2.4	277

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109	Combinatorial algorithms for structural variation detection in high-throughput sequenced genomes. Genome Research, 2009, 19, 1270-1278.	5.5	266
110	De novo rates and selection of large copy number variation. Genome Research, 2010, 20, 1469-1481.	5.5	264
111	The sea lamprey germline genome provides insights into programmed genome rearrangement and vertebrate evolution. Nature Genetics, 2018, 50, 270-277.	21.4	262
112	Copy number variation of individual cattle genomes using next-generation sequencing. Genome Research, 2012, 22, 778-790.	5.5	259
113	The discovery of integrated gene networks for autism and related disorders. Genome Research, 2015, 25, 142-154.	5.5	259
114	High-Throughput Variation Detection and Genotyping Using Microarrays. Genome Research, 2001, 11, 1913-1925.	5.5	258
115	A Human Genome Structural Variation Sequencing Resource Reveals Insights into Mutational Mechanisms. Cell, 2010, 143, 837-847.	28.9	249
116	mrsFAST: a cache-oblivious algorithm for short-read mapping. Nature Methods, 2010, 7, 576-577.	19.0	248
117	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
118	Human and murine FMR-1: alternative splicing and translational initiation downstream of the CGGâ€“repeat. Nature Genetics, 1993, 4, 244-251.	21.4	247
119	Mouse regulatory DNA landscapes reveal global principles of cis-regulatory evolution. Science, 2014, 346, 1007-1012.	12.6	244
120	Long-read sequencing and de novo assembly of a Chinese genome. Nature Communications, 2016, 7, 12065.	12.8	242
121	Chromosome evolution in eukaryotes: a multi-kingdom perspective. Trends in Genetics, 2005, 21, 673-682.	6.7	238
122	The DNA sequence of human chromosome 7. Nature, 2003, 424, 157-164.	27.8	236
123	Phenotypic variability and genetic susceptibility to genomic disorders. Human Molecular Genetics, 2010, 19, R176-R187.	2.9	234
124	Hotspots for copy number variation in chimpanzees and humans. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 8006-8011.	7.1	231
125	Neurodevelopmental disease genes implicated by de novo mutation and copy number variation morbidity. Nature Genetics, 2019, 51, 106-116.	21.4	231
126	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

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127	Structure of Chromosomal Duplicons and their Role in Mediating Human Genomic Disorders. Genome Research, 2000, 10, 597-610.	5.5	228
128	Shotgun sequence assembly and recent segmental duplications within the human genome. Nature, 2004, 431, 927-930.	27.8	228
129	Complete Haplotype Sequence of the Human Immunoglobulin Heavy-Chain Variable, Diversity, and Joining Genes and Characterization of Allelic and Copy-Number Variation. American Journal of Human Genetics, 2013, 92, 530-546.	6.2	223
130	Recurrent Reciprocal Genomic Rearrangements of 17q12 Are Associated with Renal Disease, Diabetes, and Epilepsy. American Journal of Human Genetics, 2007, 81, 1057-1069.	6.2	222
131	A burst of segmental duplications in the genome of the African great ape ancestor. Nature, 2009, 457, 877-881.	27.8	222
132	Rare copy number variants are an important cause of epileptic encephalopathies. Annals of Neurology, 2011, 70, 974-985.	5.3	222
133	Reconstructing complex regions of genomes using long-read sequencing technology. Genome Research, 2014, 24, 688-696.	5.5	222
134	Mutational and selective effects on copy-number variants in the human genome. Nature Genetics, 2007, 39, S22-S29.	21.4	221
135	Properties and rates of germline mutations in humans. Trends in Genetics, 2013, 29, 575-584.	6.7	221
136	The structure, function and evolution of a complete human chromosome 8. Nature, 2021, 593, 101-107.	27.8	221
137	Complex SNP-related sequence variation in segmental genome duplications. Nature Genetics, 2004, 36, 861-866.	21.4	220
138	Haplotype-resolved genome sequencing of a Gujarati Indian individual. Nature Biotechnology, 2011, 29, 59-63.	17.5	216
139	Copy number variation and evolution in humans and chimpanzees. Genome Research, 2008, 18, 1698-1710.	5.5	215
140	Population Stratification of a Common APOBEC Gene Deletion Polymorphism. PLoS Genetics, 2007, 3, e63.	3.5	214
141	Low copy number of the salivary amylase gene predisposes to obesity. Nature Genetics, 2014, 46, 492-497.	21.4	214
142	Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. Human Molecular Genetics, 2009, 18, 3626-3631.	2.9	211
143	Mouse segmental duplication and copy number variation. Nature Genetics, 2008, 40, 909-914.	21.4	209
144	Complete genomic and epigenetic maps of human centromeres. Science, 2022, 376, eabl4178.	12.6	204

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145	Estimating the human mutation rate using autozygosity in a founder population. Nature Genetics, 2012, 44, 1277-1281.	21.4	202
146	Systematic assessment of copy number variant detection via genome-wide SNP genotyping. Nature Genetics, 2008, 40, 1199-1203.	21.4	198
147	A Genotype-First Approach to Defining the Subtypes of a Complex Disease. Cell, 2014, 156, 872-877.	28.9	195
148	Ancestral reconstruction of segmental duplications reveals punctuated cores of human genome evolution. Nature Genetics, 2007, 39, 1361-1368.	21.4	192
149	The Human Pangenome Project: a global resource to map genomic diversity. Nature, 2022, 604, 437-446.	27.8	192
150	Duplication hotspots, rare genomic disorders, and common disease. Current Opinion in Genetics and Development, 2009, 19, 196-204.	3.3	191
151	Next-generation VariationHunter: combinatorial algorithms for transposon insertion discovery. Bioinformatics, 2010, 26, i350-i357.	4.1	190
152	The structure and evolution of centromeric transition regions within the human genome. Nature, 2004, 430, 857-864.	27.8	179
153	Completing the map of human genetic variation. Nature, 2007, 447, 161-165.	27.8	178
154	Recurrent 200-kb deletions of 16p11.2 that include the SH2B1 gene are associated with developmental delay and obesity. Genetics in Medicine, 2010, 12, 641-647.	2.4	178
155	Evolutionary toggling of the MAPT 17q21.31 inversion region. Nature Genetics, 2008, 40, 1076-1083.	21.4	176
156	Characterization of a recurrent 15q24 microdeletion syndrome. Human Molecular Genetics, 2007, 16, 567-572.	2.9	173
157	denovo-db: a compendium of humande novovariants. Nucleic Acids Research, 2017, 45, D804-D811.	14.5	173
158	Fine structure of the human FMR1 gene. Human Molecular Genetics, 1993, 2, 1147-1153.	2.9	171
159	Human-Specific Duplication and Mosaic Transcripts: The Recent Paralogous Structure of Chromosome 22. American Journal of Human Genetics, 2002, 70, 83-100.	6.2	168
160	An assessment of the sequence gaps: Unfinished business in a finished human genome. Nature Reviews Genetics, 2004, 5, 345-354.	16.3	165
161	Whole-genome shotgun assembly and comparison of human genome assemblies. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 1916-1921.	7.1	164
162	ADCY5 -related dyskinesia. Neurology, 2015, 85, 2026-2035.	1.1	163

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163	Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. Cell, 2018, 172, 897-909.e21.	28.9	163
164	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	3.8	163
165	High-resolution human genome structure by single-molecule analysis. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 10848-10853.	7.1	161
166	Evolution and diversity of copy number variation in the great ape lineage. Genome Research, 2013, 23, 1373-1382.	5.5	161
167	Human adaptation and evolution by segmental duplication. Current Opinion in Genetics and Development, 2016, 41, 44-52.	3.3	157
168	The sequence and analysis of duplication-rich human chromosome 16. Nature, 2004, 432, 988-994.	27.8	156
169	Single-cell epigenomics reveals mechanisms of human cortical development. Nature, 2021, 598, 205-213.	27.8	154
170	A genome-wide survey of structural variation between human and chimpanzee. Genome Research, 2005, 15, 1344-1356.	5.5	153
171	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
172	Programmed loss of millions of base pairs from a vertebrate genome. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 11212-11217.	7.1	151
173	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
174	Large-Scale Variation Among Human and Great Ape Genomes Determined by Array Comparative Genomic Hybridization. Genome Research, 2003, 13, 347-357.	5.5	149
175	Human uniqueness: genome interactions with environment, behaviour and culture. Nature Reviews Genetics, 2008, 9, 749-763.	16.3	149
176	Prioritization of neurodevelopmental disease genes by discovery of new mutations. Nature Neuroscience, 2014, 17, 764-772.	14.8	148
177	Speech delays and behavioral problems are the predominant features in individuals with developmental delays and 16p11.2 microdeletions and microduplications. Journal of Neurodevelopmental Disorders, 2010, 2, 26-38.	3.1	147
178	Human Hydroxysteroid Sulfotransferase SULT2B1: Two Enzymes Encoded by a Single Chromosome 19 Gene. Genomics, 1998, 53, 284-295.	2.9	146
179	From telomere to telomere: The transcriptional and epigenetic state of human repeat elements. Science, 2022, 376, eabk3112.	12.6	146
180	The genomic architecture of segmental duplications and associated copy number variants in dogs. Genome Research, 2009, 19, 491-499.	5.5	144

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181	Masquerading Repeats: Paralogous Pitfalls of the Human Genome. Genome Research, 1998, 8, 758-762.	5.5	140
182	Genome Duplications and Other Features in 12 Mb of DNA Sequence from Human Chromosome 16p and 16q. Genomics, 1999, 60, 295-308.	2.9	140
183	Interruptions in the Triplet Repeats of SCA1 and FRAXA Reduce the Propensity and Complexity of Slipped Strand DNA (S-DNA) Formationâ€. Biochemistry, 1998, 37, 2701-2708.	2.5	139
184	Long-read sequence and assembly of segmental duplications. Nature Methods, 2019, 16, 88-94.	19.0	139
185	Characterization of missing human genome sequences and copy-number polymorphic insertions. Nature Methods, 2010, 7, 365-371.	19.0	138
186	The origins and impact of primate segmental duplications. Trends in Genetics, 2009, 25, 443-454.	6.7	137
187	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.	2.9	136
188	Evolutionary Formation of New Centromeres in Macaque. Science, 2007, 316, 243-246.	12.6	136
189	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
190	Recurrent Sites for New Centromere Seeding. Genome Research, 2004, 14, 1696-1703.	5.5	135
191	Exonic Deletions in AUTS2 Cause a Syndromic Form of Intellectual Disability and Suggest a Critical Role for the C Terminus. American Journal of Human Genetics, 2013, 92, 210-220.	6.2	135
192	Single haplotype assembly of the human genome from a hydatidiform mole. Genome Research, 2014, 24, 2066-2076.	5.5	133
193	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
194	Molecular Genetic Anatomy and Risk Profile of Hirschsprungâ€™s Disease. New England Journal of Medicine, 2019, 380, 1421-1432.	27.0	131
195	Segmental duplications and their variation in a complete human genome. Science, 2022, 376, eabj6965.	12.6	130
196	Structural diversity and African origin of the 17q21.31 inversion polymorphism. Nature Genetics, 2012, 44, 872-880.	21.4	129
197	Analysis of Primate Genomic Variation Reveals a Repeat-Driven Expansion of the Human Genome. Genome Research, 2003, 13, 358-368.	5.5	127
198	Genetic Variation, Comparative Genomics, and the Diagnosis of Disease. New England Journal of Medicine, 2019, 381, 64-74.	27.0	127

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199	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
200	The evolution and population diversity of human-specific segmental duplications. Nature Ecology and Evolution, 2017, 1, 69.	7.8	123
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