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	Initial sequencing and analysis of the human genome. Nature, 2001, 409, 860-921.	36.2	21,405
2	A global reference for human genetic variation. Nature, 2015, 526, 68-74.	36.2	14,863
3	Finding the missing heritability of complex diseases. Nature, 2009, 461, 747-753.	36.2	7,627
4	Nonhybrid, finished microbial genome assemblies from long-read SMRT sequencing data. Nature Methods, 2013, 10, 563-569.	19.6	4,144
5	Genome sequence of the palaeopolyploid soybean. Nature, 2010, 463, 178-183.	36.2	3,961
6	A Hexanucleotide Repeat Expansion in C9ORF72 Is the Cause of Chromosome 9p21-Linked ALS-FTD. Neuron, 2011, 72, 257-268.	8.0	3,944
7	A Draft Sequence of the Neandertal Genome. Science, 2010, 328, 710-722.	20.9	3,707
8	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.	6.1	2,384
9	The contribution of de novo coding mutations to autism spectrum disorder. Nature, 2014, 515, 216-221.	36.2	2,281
10	An integrated map of structural variation in 2,504 human genomes. Nature, 2015, 526, 75-81.	36.2	2,084
11	Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. Nature, 2012, 485, 246-250.	36.2	1,995
12	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. Nature, 2004, 428, 493-521.	36.2	1,960
13	The complete genome sequence of a Neanderthal from the Altai Mountains. Nature, 2014, 505, 43-49.	36.2	1,931
14	Targeted capture and massively parallel sequencing of 12 human exomes. Nature, 2009, 461, 272-276.	36.2	1,826
15	A High-Coverage Genome Sequence from an Archaic Denisovan Individual. Science, 2012, 338, 222-226.	20.9	1,750
16	Genetic history of an archaic hominin group from Denisova Cave in Siberia. Nature, 2010, 468, 1053-1060.	36.2	1,610
17	The complete sequence of a human genome. Science, 2022, 376, 44-53.	20.9	1,565
18	A comparative encyclopedia of DNA elements in the mouse genome. Nature, 2014, 515, 355-364.	36.2	1,546

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19	Missing heritability and strategies for finding the underlying causes of complex disease. Nature Reviews Genetics, 2010, 11, 446-450.	16.7	1,537
20	De novo mutations in epileptic encephalopathies. Nature, 2013, 501, 217-221.	36.2	1,396
21	Genome structural variation discovery and genotyping. Nature Reviews Genetics, 2011, 12, 363-376.	16.7	1,290
22	Recent Segmental Duplications in the Human Genome. Science, 2002, 297, 1003-1007.	20.9	1,254
23	Ancient human genomes suggest three ancestral populations for present-day Europeans. Nature, 2014, 513, 409-413.	36.2	1,223
24	A copy number variation morbidity map of developmental delay. Nature Genetics, 2011, 43, 838-846.	20.4	1,166
25	Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. Nature Genetics, 2011, 43, 585-589.	20.4	1,093
26	Mapping copy number variation by population-scale genome sequencing. Nature, 2011, 470, 59-65.	36.2	1,012
27	Mapping and sequencing of structural variation from eight human genomes. Nature, 2008, 453, 56-64.	36.2	987
28	Fine-scale structural variation of the human genome. Nature Genetics, 2005, 37, 727-732.	20.4	901
29	Segmental Duplications and Copy-Number Variation in the Human Genome. American Journal of Human Genetics, 2005, 77, 78-88.	6.1	879
30	Evaluation of GRCh38 and de novo haploid genome assemblies demonstrates the enduring quality of the reference assembly. Genome Research, 2017, 27, 849-864.	5.6	802
31	Great ape genetic diversity and population history. Nature, 2013, 499, 471-475.	36.2	787
32	The genome of a songbird. Nature, 2010, 464, 757-762.	36.2	786
33	Resolving the complexity of the human genome using single-molecule sequencing. Nature, 2015, 517, 608-611.	36.2	728
34	Limitations of next-generation genome sequence assembly. Nature Methods, 2011, 8, 61-65.	19.6	691
35	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	13.2	681
36	Insights into hominid evolution from the gorilla genome sequence. Nature, 2012, 483, 169-175.	36.2	672

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37	Recurrent Rearrangements of Chromosome 1q21.1 and Variable Pediatric Phenotypes. New England Journal of Medicine, 2008, 359, 1685-1699.	30.1	668
38	Personalized copy number and segmental duplication maps using next-generation sequencing. Nature Genetics, 2009, 41, 1061-1067.	20.4	667
39	Genome analysis of the platypus reveals unique signatures of evolution. Nature, 2008, 453, 175-183.	36.2	662
40	Disruptive CHD8 Mutations Define a Subtype of Autism Early in Development. Cell, 2014, 158, 263-276.	27.8	655
41	Long-read human genome sequencing and its applications. Nature Reviews Genetics, 2020, 21, 597-614.	16.7	629
42	Diversity of Human Copy Number Variation and Multicopy Genes. Science, 2010, 330, 641-646.	20.9	616
43	Refining analyses of copy number variation identifies specific genes associated with developmental delay. Nature Genetics, 2014, 46, 1063-1071.	20.4	604
44	The Tea Tree Genome Provides Insights into Tea Flavor and Independent Evolution of Caffeine Biosynthesis. Molecular Plant, 2017, 10, 866-877.	8.4	601
45	Telomere-to-telomere assembly of a complete human X chromosome. Nature, 2020, 585, 79-84.	36.2	590
46	Copy number variation detection and genotyping from exome sequence data. Genome Research, 2012, 22, 1525-1532.	5.6	571
47	Discovery of previously unidentified genomic disorders from the duplication architecture of the human genome. Nature Genetics, 2006, 38, 1038-1042.	20.4	558
48	Comparative and demographic analysis of orang-utan genomes. Nature, 2011, 469, 529-533.	36.2	546
49	Excess of rare, inherited truncating mutations in autism. Nature Genetics, 2015, 47, 582-588.	20.4	543
50	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	20.4	542
51	LINE-1 Retrotransposition Activity in Human Genomes. Cell, 2010, 141, 1159-1170.	27.8	542
52	Population Analysis of Large Copy Number Variants and Hotspots of Human Genetic Disease. American Journal of Human Genetics, 2009, 84, 148-161.	6.1	534
53	Phenotypic Heterogeneity of Genomic Disorders and Rare Copy-Number Variants. New England Journal of Medicine, 2012, 367, 1321-1331.	30.1	526
54	15q13.3 microdeletions increase risk of idiopathic generalized epilepsy. Nature Genetics, 2009, 41, 160-162.	20.4	516

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55	HiCanu: accurate assembly of segmental duplications, satellites, and allelic variants from high-fidelity long reads. Genome Research, 2020, 30, 1291-1305.	5.6	513
56	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	20.4	511
57	Primate segmental duplications: crucibles of evolution, diversity and disease. Nature Reviews Genetics, 2006, 7, 552-564.	16.7	507
58	Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. Nature Genetics, 2009, 41, 211-215.	20.4	489
59	Modernizing Reference Genome Assemblies. PLoS Biology, 2011, 9, e1001091.	5.4	482
60	A Higher Mutational Burden in Females Supports a â€œFemale Protective Modelâ€•in Neurodevelopmental Disorders. American Journal of Human Genetics, 2014, 94, 415-425.	6.1	476
61	A Comprehensive Analysis of Common Copy-Number Variations in the Human Genome. American Journal of Human Genetics, 2007, 80, 91-104.	6.1	474
62	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	20.4	473
63	Length of uninterrupted CGG repeats determines instability in the FMR1 gene. Nature Genetics, 1994, 8, 88-94.	20.4	470
64	The bonobo genome compared with the chimpanzee and human genomes. Nature, 2012, 486, 527-531.	36.2	462
65	Establishing Cerebral Organoids as Models of Human-Specific Brain Evolution. Cell, 2019, 176, 743-756.e17.	27.8	456
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.	5.6	455
67	Segmental Duplications: Organization and Impact Within the Current Human Genome Project Assembly. Genome Research, 2001, 11, 1005-1017.	5.6	451
68	Lineage-Specific Biology Revealed by a Finished Genome Assembly of the Mouse. PLoS Biology, 2009, 7, e1000112.	5.4	429
69	Genome-Wide Copy Number Variation in Epilepsy: Novel Susceptibility Loci in Idiopathic Generalized and Focal Epilepsies. PLoS Genetics, 2010, 6, e1000962.	3.4	418
70	A de novo convergence of autism genetics and molecular neuroscience. Trends in Neurosciences, 2014, 37, 95-105.	8.8	418
71	Recurrent microdeletions at 15q11.2 and 16p13.11 predispose to idiopathic generalized epilepsies. Brain, 2010, 133, 23-32.	8.0	410
72	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. Science, 2021, 372, .	20.9	410

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73	Complete Khoisan and Bantu genomes from southern Africa. Nature, 2010, 463, 943-947.	36.2	403
74	Human-Specific NOTCH2NL Genes Affect Notch Signaling and Cortical Neurogenesis. Cell, 2018, 173, 1356-1369.e22.	27.8	399
75	Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. Nature Biotechnology, 2020, 38, 1044-1053.	20.8	390
76	An Alu Transposition Model for the Origin and Expansion of Human Segmental Duplications. American Journal of Human Genetics, 2003, 73, 823-834.	6.1	389
77	Characterizing the Major Structural Variant Alleles of the Human Genome. Cell, 2019, 176, 663-675.e19.	27.8	382
78	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.	6.1	381
79	Segmental duplications and the evolution of the primate genome. Nature Reviews Genetics, 2002, 3, 65-72.	16.7	377
80	Long-read sequence assembly of the gorilla genome. Science, 2016, 352, aae0344.	20.9	374
81	A genome-wide comparison of recent chimpanzee and human segmental duplications. Nature, 2005, 437, 88-93.	36.2	360
82	Identification and characterization of a novel peptide ligand of epidermal growth factor receptor for targeted delivery of therapeutics. FASEB Journal, 2005, 19, 1978-1985.	0.5	355
83	Human Copy Number Variation and Complex Genetic Disease. Annual Review of Genetics, 2011, 45, 203-226.	7.8	352
84	Evolution of Human-Specific Neural SRGAP2 Genes by Incomplete Segmental Duplication. Cell, 2012, 149, 912-922.	27.8	351
85	Noninvasive Whole-Genome Sequencing of a Human Fetus. Science Translational Medicine, 2012, 4, 137ra76.	13.4	350
86	Challenges and standards in integrating surveys of structural variation. Nature Genetics, 2007, 39, S7-S15.	20.4	336
87	Discovery and genotyping of structural variation from long-read haploid genome sequence data. Genome Research, 2017, 27, 677-685.	5.6	334
88	Gibbon genome and the fast karyotype evolution of small apes. Nature, 2014, 513, 195-201.	36.2	326
89	High-resolution comparative analysis of great ape genomes. Science, 2018, 360, .	20.9	318
90	A draft human pangenome reference. Nature, 2023, 617, 312-324.	36.2	318

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91	Genetic variation and the de novo assembly of human genomes. Nature Reviews Genetics, 2015, 16, 627-640.	16.7	317
92	De novo genic mutations among a Chinese autism spectrum disorder cohort. Nature Communications, 2016, 7, 13316.	13.2	313
93	Regional Patterns of Gene Expression in Human and Chimpanzee Brains. Genome Research, 2004, 14, 1462-1473.	5.6	312
94	Genomic Patterns of De Novo Mutation in Simplex Autism. Cell, 2017, 171, 710-722.e12.	27.8	312
95	Human-Specific NOTCH2NL Genes Expand Cortical Neurogenesis through Delta/Notch Regulation. Cell, 2018, 173, 1370-1384.e16.	27.8	311
96	The DNA sequence and biology of human chromosome 19. Nature, 2004, 428, 529-535.	36.2	306
97	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	20.4	303
98	Global diversity, population stratification, and selection of human copy-number variation. Science, 2015, 349, aab3761.	20.9	301
99	Relative Burden of Large CNVs on a Range of Neurodevelopmental Phenotypes. PLoS Genetics, 2011, 7, e1002334.	3.4	299
100	Recent duplication, domain accretion and the dynamic mutation of the human genome. Trends in Genetics, 2001, 17, 661-669.	6.9	297
101	Refinement and Discovery of New Hotspots of Copy-Number Variation Associated with Autism Spectrum Disorder. American Journal of Human Genetics, 2013, 92, 221-237.	6.1	287
102	Estimates of penetrance for recurrent pathogenic copy-number variations. Genetics in Medicine, 2013, 15, 478-481.	2.4	286
103	Positive selection of a gene family during the emergence of humans and African apes. Nature, 2001, 413, 514-519.	36.2	285
104	Linkage Disequilibrium and Heritability of Copy-Number Polymorphisms within Duplicated Regions of the Human Genome. American Journal of Human Genetics, 2006, 79, 275-290.	6.1	285
105	Analysis of copy number variations among diverse cattle breeds. Genome Research, 2010, 20, 693-703.	5.6	284
106	De novo rates and selection of large copy number variation. Genome Research, 2010, 20, 1469-1481.	5.6	272
107	Combinatorial algorithms for structural variation detection in high-throughput sequenced genomes. Genome Research, 2009, 19, 1270-1278.	5.6	267
108	The sea lamprey germline genome provides insights into programmed genome rearrangement and vertebrate evolution. Nature Genetics, 2018, 50, 270-277.	20.4	266

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109	Complete genomic and epigenetic maps of human centromeres. Science, 2022, 376, eabl4178.	20.9	265
110	The discovery of integrated gene networks for autism and related disorders. Genome Research, 2015, 25, 142-154.	5.6	264
111	Copy number variation of individual cattle genomes using next-generation sequencing. Genome Research, 2012, 22, 778-790.	5.6	263
112	High-Throughput Variation Detection and Genotyping Using Microarrays. Genome Research, 2001, 11, 1913-1925.	5.6	258
113	A Human Genome Structural Variation Sequencing Resource Reveals Insights into Mutational Mechanisms. Cell, 2010, 143, 837-847.	27.8	257
114	Inappropriate Prescribing. Drugs and Aging, 2012, 29, 437-452.	3.0	253
115	Human and murine FMR-1: alternative splicing and translational initiation downstream of the CGGâ€“repeat. Nature Genetics, 1993, 4, 244-251.	20.4	249
116	mrsFAST: a cache-oblivious algorithm for short-read mapping. Nature Methods, 2010, 7, 576-577.	19.6	249
117	Long-read sequencing and de novo assembly of a Chinese genome. Nature Communications, 2016, 7, 12065.	13.2	249
118	The structure, function and evolution of a complete human chromosome 8. Nature, 2021, 593, 101-107.	36.2	246
119	Chromosome evolution in eukaryotes: a multi-kingdom perspective. Trends in Genetics, 2005, 21, 673-682.	6.9	243
120	Neurodevelopmental disease genes implicated by de novo mutation and copy number variation morbidity. Nature Genetics, 2019, 51, 106-116.	20.4	243
121	Phenotypic variability and genetic susceptibility to genomic disorders. Human Molecular Genetics, 2010, 19, R176-R187.	3.0	242
122	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.	6.1	242
123	The DNA sequence of human chromosome 7. Nature, 2003, 424, 157-164.	36.2	240
124	The Human Pangenome Project: a global resource to map genomic diversity. Nature, 2022, 604, 437-446.	36.2	237
125	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.	7.6	231
126	Shotgun sequence assembly and recent segmental duplications within the human genome. Nature, 2004, 431, 927-930.	36.2	230

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127	Properties and rates of germline mutations in humans. Trends in Genetics, 2013, 29, 575-584.	6.9	229
128	Structure of Chromosomal Duplicons and their Role in Mediating Human Genomic Disorders. Genome Research, 2000, 10, 597-610.	5.6	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.	6.1	228
130	Reconstructing complex regions of genomes using long-read sequencing technology. Genome Research, 2014, 24, 688-696.	5.6	228
131	Entropically driven binding in a self-assembling molecular capsule. Nature, 1996, 382, 239-241.	36.2	227
132	Rare copy number variants are an important cause of epileptic encephalopathies. Annals of Neurology, 2011, 70, 974-985.	5.8	227
133	Recurrent Reciprocal Genomic Rearrangements of 17q12 Are Associated with Renal Disease, Diabetes, and Epilepsy. American Journal of Human Genetics, 2007, 81, 1057-1069.	6.1	226
134	Persistent Leatherback Turtle Migrations Present Opportunities for Conservation. PLoS Biology, 2008, 6, e171.	5.4	226
135	A burst of segmental duplications in the genome of the African great ape ancestor. Nature, 2009, 457, 877-881.	36.2	223
136	Mutational and selective effects on copy-number variants in the human genome. Nature Genetics, 2007, 39, S22-S29.	20.4	222
137	Complex SNP-related sequence variation in segmental genome duplications. Nature Genetics, 2004, 36, 861-866.	20.4	221
138	Population Stratification of a Common APOBEC Gene Deletion Polymorphism. PLoS Genetics, 2007, 3, e63.	3.4	219
139	Low copy number of the salivary amylase gene predisposes to obesity. Nature Genetics, 2014, 46, 492-497.	20.4	219
140	Copy number variation and evolution in humans and chimpanzees. Genome Research, 2008, 18, 1698-1710.	5.6	218
141	Haplotype-resolved genome sequencing of a Gujarati Indian individual. Nature Biotechnology, 2011, 29, 59-63.	20.8	217
142	Familial and sporadic 15q13.3 microdeletions in idiopathic generalized epilepsy: precedent for disorders with complex inheritance. Human Molecular Genetics, 2009, 18, 3626-3631.	3.0	211
143	Mouse segmental duplication and copy number variation. Nature Genetics, 2008, 40, 909-914.	20.4	209
144	Estimating the human mutation rate using autozygosity in a founder population. Nature Genetics, 2012, 44, 1277-1281.	20.4	204

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145	A Genotype-First Approach to Defining the Subtypes of a Complex Disease. Cell, 2014, 156, 872-877.	27.8	203
146	Systematic assessment of copy number variant detection via genome-wide SNP genotyping. Nature Genetics, 2008, 40, 1199-1203.	20.4	199
147	Ancestral reconstruction of segmental duplications reveals punctuated cores of human genome evolution. Nature Genetics, 2007, 39, 1361-1368.	20.4	193
148	Next-generation VariationHunter: combinatorial algorithms for transposon insertion discovery. Bioinformatics, 2010, 26, i350-i357.	4.2	192
149	Duplication hotspots, rare genomic disorders, and common disease. Current Opinion in Genetics and Development, 2009, 19, 196-204.	3.4	191
150	Calcium influx is sufficient to induce muscular dystrophy through a TRPC-dependent mechanism. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 19023-19028.	7.6	189
151	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	184
152	The structure and evolution of centromeric transition regions within the human genome. Nature, 2004, 430, 857-864.	36.2	182
153	Single-cell epigenomics reveals mechanisms of human cortical development. Nature, 2021, 598, 205-213.	36.2	181
154	Completing the map of human genetic variation. Nature, 2007, 447, 161-165.	36.2	180
155	From telomere to telomere: The transcriptional and epigenetic state of human repeat elements. Science, 2022, 376, eabk3112.	20.9	180
156	denovo-db: a compendium of humande novovariants. Nucleic Acids Research, 2017, 45, D804-D811.	14.0	179
157	Dissecting the Causal Mechanism of X-Linked Dystonia-Parkinsonism by Integrating Genome and Transcriptome Assembly. Cell, 2018, 172, 897-909.e21.	27.8	178
158	Characterization of a recurrent 15q24 microdeletion syndrome. Human Molecular Genetics, 2007, 16, 567-572.	3.0	174
159	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	3.9	173
160	Fine structure of the human FMR1 gene. Human Molecular Genetics, 1993, 2, 1147-1153.	3.0	171
161	Segmental duplications and their variation in a complete human genome. Science, 2022, 376, eabj6965.	20.9	170
162	Human-Specific Duplication and Mosaic Transcripts: The Recent Paralogous Structure of Chromosome 22. American Journal of Human Genetics, 2002, 70, 83-100.	6.1	168

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163	An assessment of the sequence gaps: Unfinished business in a finished human genome. Nature Reviews Genetics, 2004, 5, 345-354.	16.7	168
164	ADCY5 -related dyskinesia. Neurology, 2015, 85, 2026-2035.	1.1	168
165	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.	7.6	166
166	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	13.2	165
167	Evolution and diversity of copy number variation in the great ape lineage. Genome Research, 2013, 23, 1373-1382.	5.6	164
168	Human adaptation and evolution by segmental duplication. Current Opinion in Genetics and Development, 2016, 41, 44-52.	3.4	163
169	The sequence and analysis of duplication-rich human chromosome 16. Nature, 2004, 432, 988-994.	36.2	162
170	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.	7.6	162
171	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.5	159
172	SQSTM1Mutations in French Patients With Frontotemporal Dementia or Frontotemporal Dementia With Amyotrophic Lateral Sclerosis. JAMA Neurology, 2013, 70, 1403-10.	9.3	157
173	Highly tunable efficient second-harmonic generation in a lithium niobate nanophotonic waveguide. Optica, 2018, 5, 1006.	9.3	157
174	A genome-wide survey of structural variation between human and chimpanzee. Genome Research, 2005, 15, 1344-1356.	5.6	153
175	Human uniqueness: genome interactions with environment, behaviour and culture. Nature Reviews Genetics, 2008, 9, 749-763.	16.7	152
176	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.	7.6	152
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.	3.2	152
178	Long-read sequence and assembly of segmental duplications. Nature Methods, 2019, 16, 88-94.	19.6	152
179	Prioritization of neurodevelopmental disease genes by discovery of new mutations. Nature Neuroscience, 2014, 17, 764-772.	14.5	150
180	Human Hydroxysteroid Sulfotransferase SULT2B1: Two Enzymes Encoded by a Single Chromosome 19 Gene. Genomics, 1998, 53, 284-295.	2.9	149

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181	Large-Scale Variation Among Human and Great Ape Genomes Determined by Array Comparative Genomic Hybridization. Genome Research, 2003, 13, 347-357.	5.6	149
182	Epigenetic patterns in a complete human genome. Science, 2022, 376, eabj5089.	20.9	149
183	The genomic architecture of segmental duplications and associated copy number variants in dogs. Genome Research, 2009, 19, 491-499.	5.6	146
184	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.1	143
185	Genome Duplications and Other Features in 12 Mb of DNA Sequence from Human Chromosome 16p and 16q. Genomics, 1999, 60, 295-308.	2.9	142
186	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.	2.6	140
187	Masquerading Repeats: Paralogous Pitfalls of the Human Genome. Genome Research, 1998, 8, 758-762.	5.6	140
188	The origins and impact of primate segmental duplications. Trends in Genetics, 2009, 25, 443-454.	6.9	140
189	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.	6.1	140
190	Genetic Variation, Comparative Genomics, and the Diagnosis of Disease. New England Journal of Medicine, 2019, 381, 64-74.	30.1	140
191	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
192	Recurrent Sites for New Centromere Seeding. Genome Research, 2004, 14, 1696-1703.	5.6	139
193	Characterization of missing human genome sequences and copy-number polymorphic insertions. Nature Methods, 2010, 7, 365-371.	19.6	139
194	Single haplotype assembly of the human genome from a hydatidiform mole. Genome Research, 2014, 24, 2066-2076.	5.6	139
195	Evolutionary Formation of New Centromeres in Macaque. Science, 2007, 316, 243-246.	20.9	138
196	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.	3.0	136
197	Molecular Genetic Anatomy and Risk Profile of Hirschsprungâ€™s Disease. New England Journal of Medicine, 2019, 380, 1421-1432.	30.1	134
198	Structural diversity and African origin of the 17q21.31 inversion polymorphism. Nature Genetics, 2012, 44, 872-880.	20.4	133

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199	The evolution and population diversity of human-specific segmental duplications. Nature Ecology and Evolution, 2017, 1, 69.	8.0	130
200	Analysis of Primate Genomic Variation Reveals a Repeat-Driven Expansion of the Human Genome. Genome Research, 2003, 13, 358-368.	5.6	127
201	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	13.2	126
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