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List of Publications by Year in descending order

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394421 526287 5,510 31 19 27 citations g-index h-index papers 43 43 43 8322 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	The complete sequence of a human genome. Science, 2022, 376, 44-53.	12.6	1,222
2	Accurate circular consensus long-read sequencing improves variant detection and assembly of a human genome. Nature Biotechnology, 2019, 37, 1155-1162.	17.5	1,010
3	M-CAP eliminates a majority of variants of uncertain significance in clinical exomes at high sensitivity. Nature Genetics, 2016, 48, 1581-1586.	21.4	654
4	Multi-platform discovery of haplotype-resolved structural variation in human genomes. Nature Communications, 2019, 10, 1784.	12.8	636
5	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. Science, 2021, 372, .	12.6	358
6	Systematic reanalysis of clinical exome data yields additional diagnoses: implications for providers. Genetics in Medicine, 2017, 19, 209-214.	2.4	261
7	A robust benchmark for detection of germline large deletions and insertions. Nature Biotechnology, 2020, 38, 1347-1355.	17.5	233
8	Microbiota modulate transcription in the intestinal epithelium without remodeling the accessible chromatin landscape. Genome Research, 2014, 24, 1504-1516.	5 . 5	119
9	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
10	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
11	Curated variation benchmarks for challenging medically relevant autosomal genes. Nature Biotechnology, 2022, 40, 672-680.	17.5	90
12	Benchmarking challenging small variants with linked and long reads. Cell Genomics, 2022, 2, 100128.	6.5	77
13	AMELIE speeds Mendelian diagnosis by matching patient phenotype and genotype to primary literature. Science Translational Medicine, 2020, 12, .	12.4	60
14	Genomic answers for children: Dynamic analyses of >1000 pediatric rare disease genomes. Genetics in Medicine, 2022, 24, 1336-1348.	2.4	37
15	The Enhancer Landscape during Early Neocortical Development Reveals Patterns of Dense Regulation and Co-option. PLoS Genetics, 2013, 9, e1003728.	3.5	33
16	Phrank measures phenotype sets similarity to greatly improve Mendelian diagnostic disease prioritization. Genetics in Medicine, 2019, 21, 464-470.	2.4	33
17	PRISM offers a comprehensive genomic approach to transcription factor function prediction. Genome Research, 2013, 23, 889-904.	5.5	32
18	Long-read trio sequencing of individuals with unsolved intellectual disability. European Journal of Human Genetics, 2021, 29, 637-648.	2.8	27

#	Article	IF	Citations
19	Father-to-offspring transmission of extremely long NOTCH2NLC repeat expansions with contractions: genetic and epigenetic profiling with long-read sequencing. Clinical Epigenetics, 2021, 13, 204.	4.1	22
20	Biallelic lossâ€ofâ€function <i>WNT5A</i> mutations in an infant with severe and atypical manifestations of Robinow syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1030-1036.	1.2	15
21	Independent erosion of conserved transcription factor binding sites points to shared hindlimb, vision and external testes loss in different mammals. Nucleic Acids Research, 2018, 46, 9299-9308.	14.5	15
22	An MTF1 binding site disrupted by a homozygous variant in the promoter of ATP7B likely causes Wilson Disease. European Journal of Human Genetics, 2018, 26, 1810-1818.	2.8	15
23	High-coverage, long-read sequencing of Han Chinese trio reference samples. Scientific Data, 2019, 6, 91.	5.3	13
24	Automated Discovery of Tissue-Targeting Enhancers and Transcription Factors from Binding Motif and Gene Function Data. PLoS Computational Biology, 2014, 10, e1003449.	3.2	11
25	A multi-platform reference for somatic structural variation detection. Cell Genomics, 2022, 2, 100139.	6.5	10
26	Germline mosaicism of a missense variant in <scp><i>KCNC2</i></scp> in a multiplex family with autism and epilepsy characterized by longâ€read sequencing. American Journal of Medical Genetics, Part A, 2022, 188, 2071-2081.	1.2	7
27	A crowdsourced set of curated structural variants for the human genome. PLoS Computational Biology, 2020, 16, e1007933.	3.2	6
28	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		0
29	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		0
30	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		0
31	A crowdsourced set of curated structural variants for the human genome. , 2020, 16, e1007933.		O