Multi-platform discovery of haplotype-resolved structu

Nature Communications

10, 1784

DOI: 10.1038/s41467-018-08148-z

Citation Report

#	Article	IF	Citations
1	Versatile Quality Control Methods for Nanopore Sequencing. Evolutionary Bioinformatics, 2019, 15, 117693431986306.	1.2	1
2	Comparison of mitochondrial DNA variants detection using short- and long-read sequencing. Journal of Human Genetics, 2019, 64, 1107-1116.	2.3	8
3	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
4	Goodbye reference, hello genome graphs. Nature Biotechnology, 2019, 37, 866-868.	17.5	19
5	A clinically validated whole genome pipeline for structural variant detection and analysis. BMC Genomics, 2019, 20, 545.	2.8	15
6	Long-read sequencing in human genetics. Medizinische Genetik, 2019, 31, 198-204.	0.2	12
7	Fully-sensitive seed finding in sequence graphs using a hybrid index. Bioinformatics, 2019, 35, i81-i89.	4.1	12
8	Large-scale mammalian genome rearrangements coincide with chromatin interactions. Bioinformatics, 2019, 35, i117-i126.	4.1	4
9	Genetic Variation, Comparative Genomics, and the Diagnosis of Disease. New England Journal of Medicine, 2019, 381, 64-74.	27.0	127
10	Joint inference and alignment of genome structures enables characterization of compartment-independent reorganization across cell types. Epigenetics and Chromatin, 2019, 12, 61.	3.9	4
11	Longshot enables accurate variant calling in diploid genomes from single-molecule long read sequencing. Nature Communications, 2019, 10, 4660.	12.8	156
12	Hecaton: reliably detecting copy number variation in plant genomes using short read sequencing data. BMC Genomics, 2019, 20, 818.	2.8	4
13	The Evolution of DNA Sequencing in Pharmacogenomics. Advances in Molecular Pathology, 2019, 2, 119-131.	0.4	1
14	breakpointR: an R/Bioconductor package to localize strand state changes in Strand-seq data. Bioinformatics, 2020, 36, 1260-1261.	4.1	32
15	The population genetics of structural variants in grapevine domestication. Nature Plants, 2019, 5, 965-979.	9.3	229
16	Clonal crops show structural variation role in domestication. Nature Plants, 2019, 5, 915-916.	9.3	4
17	Evolutionary and functional impact of common polymorphic inversions in the human genome. Nature Communications, 2019, 10, 4222.	12.8	34
18	Mapping Genome Variants Sheds Light on Genetic and Phenotypic Differentiation in Chinese. Genomics, Proteomics and Bioinformatics, 2019, 17, 226-228.	6.9	1

#	Article	IF	Citations
19	Pedigree-based estimation of human mobile element retrotransposition rates. Genome Research, 2019, 29, 1567-1577.	5.5	75
20	VISOR: a versatile haplotype-aware structural variant simulator for short- and long-read sequencing. Bioinformatics, 2020, 36, 1267-1269.	4.1	29
21	SVIM: structural variant identification using mapped long reads. Bioinformatics, 2019, 35, 2907-2915.	4.1	173
22	One reference genome is not enough. Genome Biology, 2019, 20, 104.	8.8	58
23	High-coverage, long-read sequencing of Han Chinese trio reference samples. Scientific Data, 2019, 6, 91.	5.3	13
24	Haplotype-aware diplotyping from noisy long reads. Genome Biology, 2019, 20, 116.	8.8	43
25	Comprehensive evaluation of structural variation detection algorithms for whole genome sequencing. Genome Biology, 2019, 20, 117.	8.8	311
26	Structural variants identified by Oxford Nanopore PromethION sequencing of the human genome. Genome Research, 2019, 29, 1178-1187.	5.5	143
27	Overlap graph-based generation of haplotigs for diploids and polyploids. Bioinformatics, 2019, 35, 4281-4289.	4.1	16
28	Long-Read Sequencing Emerging in Medical Genetics. Frontiers in Genetics, 2019, 10, 426.	2.3	290
29	Genomic Analysis in the Age of Human Genome Sequencing. Cell, 2019, 177, 70-84.	28.9	205
30	Bit-parallel sequence-to-graph alignment. Bioinformatics, 2019, 35, 3599-3607.	4.1	50
31	Discovery of tandem and interspersed segmental duplications using high-throughput sequencing. Bioinformatics, 2019, 35, 3923-3930.	4.1	29
32	A 12-kb structural variation in progressive myoclonic epilepsy was newly identified by long-read whole-genome sequencing. Journal of Human Genetics, 2019, 64, 359-368.	2.3	48
33	HFM: Hierarchical Feature Moment Extraction for Multi-Omic Data Visualization. , 2019, , .		1
34	Capture of complete ciliate chromosomes in single sequencing reads reveals widespread chromosome isoforms. BMC Genomics, 2019, 20, 1037.	2.8	12
35	Paragraph: a graph-based structural variant genotyper for short-read sequence data. Genome Biology, 2019, 20, 291.	8.8	104
36	Evaluating nanopore sequencing data processing pipelines for structural variation identification. Genome Biology, 2019, 20, 237.	8.8	34

#	Article	IF	Citations
37	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
38	SyRI: finding genomic rearrangements and local sequence differences from whole-genome assemblies. Genome Biology, 2019, 20, 277.	8.8	310
39	Structural variant calling: the long and the short of it. Genome Biology, 2019, 20, 246.	8.8	409
40	Detecting, Categorizing, and Correcting Coverage Anomalies of RNA-Seq Quantification. Cell Systems, 2019, 9, 589-599.e7.	6.2	5
41	Assembly of chromosome-scale contigs by efficiently resolving repetitive sequences with long reads. Nature Communications, 2019, 10, 5360.	12.8	62
42	GraphTyper2 enables population-scale genotyping of structural variation using pangenome graphs. Nature Communications, 2019, 10, 5402.	12.8	96
43	ALS Genetics, Mechanisms, and Therapeutics: Where Are We Now?. Frontiers in Neuroscience, 2019, 13, 1310.	2.8	487
44	High throughput barcoding method for genome-scale phasing. Scientific Reports, 2019, 9, 18116.	3.3	13
45	Characterizing the Major Structural Variant Alleles of the Human Genome. Cell, 2019, 176, 663-675.e19.	28.9	364
46	MRLR: unraveling high-resolution meiotic recombination by linked reads. Bioinformatics, 2020, 36, 10-16.	4.1	4
47	Prediction and management of CAD risk based on genetic stratification. Trends in Cardiovascular Medicine, 2020, 30, 328-334.	4.9	14
48	An Evolutionary Perspective on the Impact of Genomic Copy Number Variation on Human Health. Journal of Molecular Evolution, 2020, 88, 104-119.	1.8	27
49	The International Genome Sample Resource (IGSR) collection of open human genomic variation resources. Nucleic Acids Research, 2020, 48, D941-D947.	14.5	221
50	MsPAC: a tool for haplotype-phased structural variant detection. Bioinformatics, 2020, 36, 922-924.	4.1	23
51	A Return to the Origin of the EMGS: Rejuvenating the Quest for Human Germ Cell Mutagens and Determining the Risk to Future Generations. Environmental and Molecular Mutagenesis, 2020, 61, 42-54.	2.2	13
52	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
53	Structural variant identification and characterization. Chromosome Research, 2020, 28, 31-47.	2.2	13
54	Challenges in identifying large germline structural variants for clinical use by long read sequencing. Computational and Structural Biotechnology Journal, 2020, 18, 83-92.	4.1	18

#	Article	IF	Citations
55	Single-cell analysis of structural variations and complex rearrangements with tri-channel processing. Nature Biotechnology, 2020, 38, 343-354.	17.5	59
56	Identification and characterization of occult human-specific LINE-1 insertions using long-read sequencing technology. Nucleic Acids Research, 2020, 48, 1146-1163.	14.5	68
57	Structural variation in the sequencing era. Nature Reviews Genetics, 2020, 21, 171-189.	16.3	337
58	Long walk to genomics: History and current approaches to genome sequencing and assembly. Computational and Structural Biotechnology Journal, 2020, 18, 9-19.	4.1	171
59	Detecting chromatin interactions between and along sister chromatids with SisterC. Nature Methods, 2020, 17, 1002-1009.	19.0	31
60	A Novel Framework for Characterizing Genomic Haplotype Diversity in the Human Immunoglobulin Heavy Chain Locus. Frontiers in Immunology, 2020, 11, 2136.	4.8	54
61	TRiCoLOR: tandem repeat profiling using whole-genome long-read sequencing data. GigaScience, 2020, 9, .	6.4	15
62	Genomic Diagnosis for Pediatric Disorders: Revolution and Evolution. Frontiers in Pediatrics, 2020, 8, 373.	1.9	30
63	Evolutionary Genomics of Structural Variation in Asian Rice (Oryza sativa) Domestication. Molecular Biology and Evolution, 2020, 37, 3507-3524.	8.9	58
64	Towards a better understanding of the low recall of insertion variants with short-read based variant callers. BMC Genomics, 2020, 21, 762.	2.8	12
65	SVFX: a machine learning framework to quantify the pathogenicity of structural variants. Genome Biology, 2020, 21, 274.	8.8	24
66	Extreme enrichment of VNTR-associated polymorphicity in human subtelomeres: genes with most VNTRs are predominantly expressed in the brain. Translational Psychiatry, 2020, 10, 369.	4.8	15
67	First submicroscopic inversion of the OPA1 gene identified in dominant optic atrophy â€“ a case report. BMC Medical Genetics, 2020, 21, 236.	2.1	17
68	Amplification-free long-read sequencing reveals unforeseen CRISPR-Cas9 off-target activity. Genome Biology, 2020, 21, 290.	8.8	35
69	Disease-modifying effects of an SCAF4 structural variant in a predominantly SOD1 ALS cohort. Neurology: Genetics, 2020, 6, e470.	1.9	9
70	Exome Sequencing and Clinical Diagnosis. JAMA - Journal of the American Medical Association, 2020, 324, 627.	7.4	9
71	Long-read-based human genomic structural variation detection with cuteSV. Genome Biology, 2020, 21, 189.	8.8	164
72	Bovine breed-specific augmented reference graphs facilitate accurate sequence read mapping and unbiased variant discovery. Genome Biology, 2020, 21, 184.	8.8	33

#	Article	IF	Citations
73	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
74	Best practices for variant calling in clinical sequencing. Genome Medicine, 2020, 12, 91.	8.2	178
75	Towards a reference genome that captures global genetic diversity. Nature Communications, 2020, 11, 5482.	12.8	34
76	CRISPR-Cas9 enrichment and long read sequencing for fine mapping in plants. Plant Methods, 2020, 16, 121.	4.3	31
77	Decoding of Oxygen Network Distortion in a Layered High-Rate Anode by In Situ Investigation of a Single Microelectrode. ACS Nano, 2020, 14, 11753-11764.	14.6	10
78	Comprehensive analysis of structural variants in breast cancer genomes using single-molecule sequencing. Genome Research, 2020, 30, 1258-1273.	5.5	72
79	Haplotype threading: accurate polyploid phasing from long reads. Genome Biology, 2020, 21, 252.	8.8	50
80	A diploid assembly-based benchmark for variants in the major histocompatibility complex. Nature Communications, 2020, 11, 4794.	12.8	56
81	GraphAligner: rapid and versatile sequence-to-graph alignment. Genome Biology, 2020, 21, 253.	8.8	90
82	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	2.5	12
83	Merqury: reference-free quality, completeness, and phasing assessment for genome assemblies. Genome Biology, 2020, 21, 245.	8.8	975
84	Shiny-SoSV: A web-based performance calculator for somatic structural variant detection. PLoS ONE, 2020, 15, e0238108.	2.5	0
85	HiCanu: accurate assembly of segmental duplications, satellites, and allelic variants from high-fidelity long reads. Genome Research, 2020, 30, 1291-1305.	5.5	440
86	A Practical Guide for Structural Variation Detection in the Human Genome. Current Protocols in Human Genetics, 2020, 107, e103.	3.5	12
87	Evaluation of Single-Molecule Sequencing Technologies for Structural Variant Detection in Two Swedish Human Genomes. Genes, 2020, 11, 1444.	2.4	6
88	Prioritizing long range interactions in noncoding regions using GWAS and deletions perturbed TADs. Computational and Structural Biotechnology Journal, 2020, 18, 2945-2952.	4.1	2
89	Clinical Interpretation and Management of Genetic Variants. JACC Basic To Translational Science, 2020, 5, 1029-1042.	4.1	23
90	Association of a structural variant within the SQSTM1 gene with amyotrophic lateral sclerosis. Neurology: Genetics, 2020, 6, e406.	1.9	9

#	Article	IF	Citations
91	Polymorphic Inversions Underlie the Shared Genetic Susceptibility of Obesity-Related Diseases. American Journal of Human Genetics, 2020, 106, 846-858.	6.2	11
92	Pangenome Graphs. Annual Review of Genomics and Human Genetics, 2020, 21, 139-162.	6.2	148
93	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	27.8	614
94	Population Structure, Stratification, and Introgression of Human Structural Variation. Cell, 2020, 182, 189-199.e15.	28.9	79
95	A robust benchmark for detection of germline large deletions and insertions. Nature Biotechnology, 2020, 38, 1347-1355.	17.5	233
96	Recurrent inversion toggling and great ape genome evolution. Nature Genetics, 2020, 52, 849-858.	21.4	40
97	Ultralow-input single-tube linked-read library method enables short-read second-generation sequencing systems to routinely generate highly accurate and economical long-range sequencing information. Genome Research, 2020, 30, 898-909.	5.5	68
98	Discovery and quality analysis of a comprehensive set of structural variants and short tandem repeats. Nature Communications, 2020, 11, 2928.	12.8	22
99	Mapping and characterization of structural variation in 17,795 human genomes. Nature, 2020, 583, 83-89.	27.8	194
100	Long-read human genome sequencing and its applications. Nature Reviews Genetics, 2020, 21, 597-614.	16.3	542
101	Editorial: Overcoming current limitations of genetic testing in cardiovascular medicine. Current Opinion in Cardiology, 2020, 35, 187-190.	1.8	0
102	Phen2Gene: rapid phenotype-driven gene prioritization for rare diseases. NAR Genomics and Bioinformatics, 2020, 2, lqaa032.	3.2	45
103	A crowdsourced set of curated structural variants for the human genome. PLoS Computational Biology, 2020, 16, e1007933.	3.2	6
104	Identification of Structural Variation in Chimpanzees Using Optical Mapping and Nanopore Sequencing. Genes, 2020, 11, 276.	2.4	14
105	Evaluating Structural Variation Detection Tools for Long-Read Sequencing Datasets in Saccharomyces cerevisiae. Frontiers in Genetics, 2020, 11, 159.	2.3	13
106	VALOR2: characterization of large-scale structural variants using linked-reads. Genome Biology, 2020, 21, 72.	8.8	15
107	NanoVar: accurate characterization of patientsâ€™ genomic structural variants using low-depth nanopore sequencing. Genome Biology, 2020, 21, 56.	8.8	73
108	Fully Phased Sequence of a Diploid Human Genome Determined de Novo from the DNA of a Single Individual. G3: Genes, Genomes, Genetics, 2020, 10, 2911-2925.	1.8	8

#	Article	IF	Citations
109	Discovery and population genomics of structural variation in a songbird genus. Nature Communications, 2020, 11, 3403.	12.8	83
110	Long-read sequencing to understand genome biology and cell function. International Journal of Biochemistry and Cell Biology, 2020, 126, 105799.	2.8	26
111	Contribution of unfixed transposable element insertions to human regulatory variation. Philosophical Transactions of the Royal Society B: Biological Sciences, 2020, 375, 20190331.	4.0	32
112	Structural Variants May Be a Source of Missing Heritability in sALS. Frontiers in Neuroscience, 2020, 14, 47.	2.8	43
113	Structural variation and its potential impact on genome instability: Novel discoveries in the EGFR landscape by long-read sequencing. PLoS ONE, 2020, 15, e0226340.	2.5	21
114	The effects of common structural variants on 3D chromatin structure. BMC Genomics, 2020, 21, 95.	2.8	23
115	Copy number variation in human genomes from three major ethno-linguistic groups in Africa. BMC Genomics, 2020, 21, 289.	2.8	7
116	A Roadmap for Understanding the Evolutionary Significance of Structural Genomic Variation. Trends in Ecology and Evolution, 2020, 35, 561-572.	8.7	190
117	Inverted duplicate DNA sequences increase translocation rates through sequencing nanopores resulting in reduced base calling accuracy. Nucleic Acids Research, 2020, 48, 4940-4945.	14.5	26
118	De novo diploid genome assembly for genome-wide structural variant detection. NAR Genomics and Bioinformatics, 2020, 2, lqz018.	3.2	9
119	First interchromosomal insertion in a patient with cerebral and spinal cavernous malformations. Scientific Reports, 2020, 10, 6306.	3.3	4
120	Critical length in long-read resequencing. NAR Genomics and Bioinformatics, 2020, 2, lqz027.	3.2	4
121	Mobile genomics: tools and techniques for tackling transposons. Philosophical Transactions of the Royal Society B: Biological Sciences, 2020, 375, 20190345.	4.0	39
122	Loss-of-function tolerance of enhancers in the human genome. PLoS Genetics, 2020, 16, e1008663.	3.5	12
123	Pathogenic 12-kb copy-neutral inversion in syndromic intellectual disability identified by high-fidelity long-read sequencing. Genomics, 2021, 113, 1044-1053.	2.9	11
124	Efficient dynamic variation graphs. Bioinformatics, 2021, 36, 5139-5144.	4.1	18
125	Interpreting the impact of noncoding structural variation in neurodevelopmental disorders. Genetics in Medicine, 2021, 23, 34-46.	2.4	32
126	Genomic sequencing analysis reveals copy number variations and their associations with economically important traits in beef cattle. Genomics, 2021, 113, 812-820.	2.9	10

#	Article	IF	Citations
127	Implications of germline copy-number variations in psychiatric disorders: review of large-scale genetic studies. Journal of Human Genetics, 2021, 66, 25-37.	2.3	22
128	Long-read trio sequencing of individuals with unsolved intellectual disability. European Journal of Human Genetics, 2021, 29, 637-648.	2.8	27
129	Chromosome-scale, haplotype-resolved assembly of human genomes. Nature Biotechnology, 2021, 39, 309-312.	17.5	109
130	Customized optical mapping by CRISPRâ€"Cas9 mediated DNA labeling with multiple sgRNAs. Nucleic Acids Research, 2021, 49, e8-e8.	14.5	15
131	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
132	SVIM-asm: structural variant detection from haploid and diploid genome assemblies. Bioinformatics, 2021, 36, 5519-5521.	4.1	47
133	Breakpoint mapping of a t(9;22;12) chronic myeloid leukaemia patient with e14a3 BCRâ€ABL1 transcript using Nanopore sequencing. Journal of Gene Medicine, 2021, 23, e3276.	2.8	6
134	Nebula: ultra-efficient mapping-free structural variant genotyper. Nucleic Acids Research, 2021, 49, e47-e47.	14.5	14
137	SeeCiTe: a method to assess CNV calls from SNP arrays using trio data. Bioinformatics, 2021, 37, 1876-1883.	4.1	4
138	Genome structure variation analyses of peach reveal population dynamics and a 1.67â€‰Mb causal inversion for fruit shape. Genome Biology, 2021, 22, 13.	8.8	50
139	Pharmacogenomics: Genetic Polymorphisms. , 2021, , 1-10.		1
140	Genomic disorders in the genomics era. , 2021, , 35-59.		1
141	Methods to Study Genomic DNA Sequence Variation. , 2021, , 59-92.		0
142	Comparative genome analysis using sample-specific string detection in accurate long reads. Bioinformatics Advances, 2021, 1, .	2.4	5
143	Improving diagnostics of rare genetic diseases with NGS approaches. Journal of Community Genetics, 2021, 12, 247-256.	1.2	25
145	Identification of Somatic Structural Variants in Solid Tumors by Optical Genome Mapping. Journal of Personalized Medicine, 2021, 11, 142.	2.5	20
146	ClinSV: clinical grade structural and copy number variant detection from whole genome sequencing data. Genome Medicine, 2021, 13, 32.	8.2	36
147	A Reference Genome Assembly of American Bison,Bison bison bison. Journal of Heredity, 2021, 112, 174-183.	2.4	14

#	Article	IF	Citations
149	Evolution of genome structure in theDrosophila simulansspecies complex. Genome Research, 2021, 31, 380-396.	5.5	55
150	Haplotype-resolved de novo assembly using phased assembly graphs with hifiasm. Nature Methods, 2021, 18, 170-175.	19.0	1,675
153	Aquila enables reference-assisted diploid personal genome assembly and comprehensive variant detection based on linked reads. Nature Communications, 2021, 12, 1077.	12.8	11
154	AnÂinternationalÂvirtualÂhackathon toÂbuildÂtools for theÂanalysis ofÂstructuralÂvariants withinÂspeciesÂranging fromÂcoronaviruses toÂvertebrates. F1000Research, 2021, 10, 246.	1.6	3
155	When the genome bluffs: a tandem duplication event during generation of a novel Agmo knockout mouse model fools routine genotyping. Cell and Bioscience, 2021, 11, 54.	4.8	12
157	Structural variant detection in cancer genomes: computational challenges and perspectives for precision oncology. Npj Precision Oncology, 2021, 5, 15.	5.4	30
158	Robust Benchmark Structural Variant Calls of An Asian Using State-of-the-art Long-read Sequencing Technologies. Genomics, Proteomics and Bioinformatics, 2022, 20, 192-204.	6.9	6
159	Comprehensive identification of somatic nucleotide variants in human brain tissue. Genome Biology, 2021, 22, 92.	8.8	26
160	Long-read assembly of a Great Dane genome highlights the contribution of GC-rich sequence and mobile elements to canine genomes. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	25
161	Short and long-read genome sequencing methodologies for somatic variant detection; genomic analysis of a patient with diffuse large B-cell lymphoma. Scientific Reports, 2021, 11, 6408.	3.3	14
163	Comparison of Structural and Short Variants Detected by Linked-Read and Whole-Exome Sequencing in Multiple Myeloma. Cancers, 2021, 13, 1212.	3.7	5
165	Fine Mapping of Leaf Trichome Density Revealed a 747-kb Region on Chromosome 1 in Cold-Hardy Hybrid Wine Grape Populations. Frontiers in Plant Science, 2021, 12, 587640.	3.6	12
166	ASHLEYS: automated quality control for single-cell Strand-seq data. Bioinformatics, 2021, 37, 3356-3357.	4.1	9
167	GAMIBHEAR: whole-genome haplotype reconstruction from Genome Architecture Mapping data. Bioinformatics, 2021, 37, 3128-3135.	4.1	1
168	A Population-Specific Major Allele Reference Genome From The United Arab Emirates Population. Frontiers in Genetics, 2021, 12, 660428.	2.3	9
169	The structure, function and evolution of a complete human chromosome 8. Nature, 2021, 593, 101-107.	27.8	221
170	Personalized genome structure via single gamete sequencing. Genome Biology, 2021, 22, 112.	8.8	10
171	Next-generation sequencing for constitutional variants in the clinical laboratory, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1399-1415.	2.4	64

#	Article	IF	Citations
173	CaBagE: A Cas9-based Background Elimination strategy for targeted, long-read DNA sequencing. PLoS ONE, 2021, 16, e0241253.	2.5	17
175	Single-molecule optical genome mapping in nanochannels: multidisciplinarity at the nanoscale. Essays in Biochemistry, 2021, 65, 51-66.	4.7	25
176	Long-Read Sequencing to Unravel Complex Structural Variants of CEP78 Leading to Cone-Rod Dystrophy and Hearing Loss. Frontiers in Cell and Developmental Biology, 2021, 9, 664317.	3.7	11
177	Haplotype-resolved diverse human genomes and integrated analysis of structural variation. Science, 2021, 372, .	12.6	358
178	Extended haplotype-phasing of long-read de novo genome assemblies using Hi-C. Nature Communications, 2021, 12, 1935.	12.8	64
179	Virus-derived variation in diverse human genomes. PLoS Genetics, 2021, 17, e1009324.	3.5	0
180	Detection and inference of interspersed duplicated insertions from paired-end reads. , 2021, 111, 102959.		0
181	Computational methods for chromosome-scale haplotype reconstruction. Genome Biology, 2021, 22, 101.	8.8	49
183	De novo structural mutation rates and gamete-of-origin biases revealed through genome sequencing of 2,396 families. American Journal of Human Genetics, 2021, 108, 597-607.	6.2	57
184	Optimizing Nanopore sequencing-based detection of structural variants enables individualized circulating tumor DNA-based disease monitoring in cancer patients. Genome Medicine, 2021, 13, 86.	8.2	14
185	Long-read sequencing of 3,622 Icelanders provides insight into the role of structural variants in human diseases and other traits. Nature Genetics, 2021, 53, 779-786.	21.4	156
186	Opportunities and challenges for highâ€quality biodiversity tissue archives in the age of longâ€read sequencing. Molecular Ecology, 2021, 30, 5935-5948.	3.9	21
187	An approach to rapid characterization of DMD copy number variants for prenatal risk assessment. American Journal of Medical Genetics, Part A, 2021, 185, 2541-2545.	1.2	3
188	snpXplorer: a web application to explore human SNP-associations and annotate SNP-sets. Nucleic Acids Research, 2021, 49, W603-W612.	14.5	14
189	Samplot: a platform for structural variant visual validation and automated filtering. Genome Biology, 2021, 22, 161.	8.8	52
190	Comparison of structural variants detected by optical mapping with long-read next-generation sequencing. Bioinformatics, 2021, 37, 3398-3404.	4.1	5
191	Characterization of structural variation in Tibetans reveals new evidence of high-altitude adaptation and introgression. Genome Biology, 2021, 22, 159.	8.8	34
192	Chromosome-Level Assembly of the Atlantic Silverside Genome Reveals Extreme Levels of Sequence Diversity and Structural Genetic Variation. Genome Biology and Evolution, 2021, 13, .	2.5	20

#	Article	IF	Citations
193	Characterization of intermediate-sized insertions using whole-genome sequencing data and analysis of their functional impact on gene expression. Human Genetics, 2021, 140, 1201-1216.	3.8	3
195	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
198	Intronic Breakpoint Signatures Enhance Detection and Characterization of Clinically Relevant Germline Structural Variants. Journal of Molecular Diagnostics, 2021, 23, 612-629.	2.8	4
199	Pervasive cis effects of variation in copy number of large tandem repeats on local DNA methylation and gene expression. American Journal of Human Genetics, 2021, 108, 809-824.	6.2	30
200	Optical genome mapping, a promising alternative to gold standard cytogenetic approaches in a series of acute lymphoblastic leukemias. Genes Chromosomes and Cancer, 2021, 60, 657-667.	2.8	47
202	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	1.2	11
203	Towards population-scale long-read sequencing. Nature Reviews Genetics, 2021, 22, 572-587.	16.3	163
204	Hi-C as a molecular rangefinder to examine genomic rearrangements. Seminars in Cell and Developmental Biology, 2022, 121, 161-170.	5.0	7
205	FaNDOM: Fast nested distance-based seeding of optical maps. Patterns, 2021, 2, 100248.	5.9	11
206	Explaining the missing heritability of psychiatric disorders. World Psychiatry, 2021, 20, 294-295.	10.4	18
207	Lynch syndrome and Muir-Torre phenotype associated with a recurrent variant in the 3â€™UTR of the MSH6 gene. Cancer Genetics, 2021, 254-255, 1-10.	0.4	4
208	CIRCNV: Detection of CNVs Based on a Circular Profile of Read Depth from Sequencing Data. Biology, 2021, 10, 584.	2.8	1
209	Towards improved genetic diagnosis of human differences of sex development. Nature Reviews Genetics, 2021, 22, 588-602.	16.3	35
210	An integrated approach for copy number variation discovery in parentâ€“offspring trios. Briefings in Bioinformatics, 2021, 22, .	6.5	0
211	Cas9 targeted enrichment of mobile elements using nanopore sequencing. Nature Communications, 2021, 12, 3586.	12.8	33
212	Comprehensive identification of transposable element insertions using multiple sequencing technologies. Nature Communications, 2021, 12, 3836.	12.8	44
213	lra: A long read aligner for sequences and contigs. PLoS Computational Biology, 2021, 17, e1009078.	3.2	59
217	Mako: A Graph-based Pattern Growth Approach to Detect Complex Structural Variants. Genomics, Proteomics and Bioinformatics, 2022, 20, 205-218.	6.9	6

#	Article	IF	Citations
218	Discovery of genomic variation across a generation. Human Molecular Genetics, 2021, 30, R174-R186.	2.9	9
219	InvertypeR: Bayesian inversion genotyping with Strand-seq data. BMC Genomics, 2021, 22, 582.	2.8	3
220	Haplotype-resolved germline and somatic alterations in renal medullary carcinomas. Genome Medicine, 2021, 13, 114.	8.2	5
221	Profiling variable-number tandem repeat variation across populations using repeat-pangenome graphs. Nature Communications, 2021, 12, 4250.	12.8	27
222	Multicolor Whole-Genome Mapping in Nanochannels for Genetic Analysis. Analytical Chemistry, 2021, 93, 9808-9816.	6.5	6
224	Optical genome mapping enables constitutional chromosomal aberration detection. American Journal of Human Genetics, 2021, 108, 1409-1422.	6.2	108
225	Chromoanagenesis Event Underlies a de novo Pericentric and Multiple Paracentric Inversions in a Single Chromosome Causing Coffinâ€"Siris Syndrome. Frontiers in Genetics, 2021, 12, 708348.	2.3	5
226	SpecHap: a diploid phasing algorithm based on spectral graph theory. Nucleic Acids Research, 2021, 49, e114.	14.5	6
227	Clinical utility and costâ€effectiveness analysis of chromosome testing concomitant with chromosomal microarray of patients with constitutional disorders in a U.S. academic medical center. Journal of Genetic Counseling, 2021, , .	1.6	1
228	PanSVR: Pan-Genome Augmented Short Read Realignment for Sensitive Detection of Structural Variations. Frontiers in Genetics, 2021, 12, 731515.	2.3	2
229	Evidence for opposing selective forces operating on human-specific duplicated TCAF genes in Neanderthals and humans. Nature Communications, 2021, 12, 5118.	12.8	14
230	Evaluating the performance of a clinical genome sequencing program for diagnosis of rare genetic disease, seen through the lens of craniosynostosis. Genetics in Medicine, 2021, 23, 2360-2368.	2.4	13
231	From DNA human sequence to the chromatin higher order organisation and its biological meaning: Using biomolecular interaction networks to understand the influence of structural variation on spatial genome organisation and its functional effect. Seminars in Cell and Developmental Biology, 2022, 121, 171-185.	5.0	17
232	Genetics of hypogonadotropic Hypogonadismâ€"Human and mouse genes, inheritance, oligogenicity, and genetic counseling. Molecular and Cellular Endocrinology, 2021, 534, 111334.	3.2	13
233	Next-generation cytogenetics: Comprehensive assessment of 52 hematological malignancy genomes by optical genome mapping. American Journal of Human Genetics, 2021, 108, 1423-1435.	6.2	85
234	The Need for a Human Pangenome Reference Sequence. Annual Review of Genomics and Human Genetics, 2021, 22, 81-102.	6.2	71
235	Exploring the missing heritability in subjects with hearing loss, enlarged vestibular aqueducts, and a single or no pathogenic SLC26A4 variant. Human Genetics, 2022, 141, 465-484.	3.8	3
236	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	6.2	105

#	Article	IF	Citations
237	Genome sequencing data analysis for rare disease gene discovery. Briefings in Bioinformatics, 2022, 23, .	6.5	6
239	Precise Characterization of Bombyx mori Fibroin Heavy Chain Gene Using Cpf1-Based Enrichment and Oxford Nanopore Technologies. Insects, 2021, 12, 832.	2.2	3
240	Multiplatform discovery and regulatory function analysis of structural variations in non-small cell lung carcinoma. Cell Reports, 2021, 36, 109660.	6.4	3
241	Application of full-genome analysis to diagnose rare monogenic disorders. Npj Genomic Medicine, 2021, 6, 77.	3.8	22
242	Local adaptation and archaic introgression shape global diversity at human structural variant loci. ELife, 2021, 10, .	6.0	33
243	AnÂinternationalÂvirtualÂhackathon toÂbuildÂtools for theÂanalysis ofÂstructuralÂvariants withinÂspeciesÂranging fromÂcoronaviruses toÂvertebrates. F1000Research, 2021, 10, 246.	1.6	2
244	Long-read technologies identify a hidden inverted duplication in a family with choroideremia. Human Genetics and Genomics Advances, 2021, 2, 100046.	1.7	4
245	Characterization of full-length LINE-1 insertions in 154 genomes. Genomics, 2021, 113, 3804-3810.	2.9	2
246	Prevalence and Phenotypic Impact of Robertsonian Translocations. Molecular Syndromology, 2021, 12, 1-11.	0.8	18
247	Next-generation cytogenomics: High-resolution structural variation detection by optical genome mapping. , 2021, , 123-146.		1
248	nanotatoR: a tool for enhanced annotation of genomic structural variants. BMC Genomics, 2021, 22, 10.	2.8	6
249	Thousands of human sequences provide deep insight into single genomes. Nature, 2020, 581, 385-386.	27.8	1
250	Parliament2: Accurate structural variant calling at scale. GigaScience, 2020, 9, .	6.4	51
251	3D-GNOME 2.0: a three-dimensional genome modeling engine for predicting structural variation-driven alterations of chromatin spatial structure in the human genome. Nucleic Acids Research, 2020, 48, W170-W176.	14.5	19
289	Determining the impact of uncharacterized inversions in the human genome by droplet digital PCR. Genome Research, 2020, 30, 724-735.	5.5	18
290	Gamete binning: chromosome-level and haplotype-resolved genome assembly enabled by high-throughput single-cell sequencing of gamete genomes. Genome Biology, 2020, 21, 306.	8.8	44
291	Genotyping structural variants in pangenome graphs using the vg toolkit. Genome Biology, 2020, 21, 35.	8.8	150
292	Integrative analysis of structural variations using short-reads and linked-reads yields highly specific and sensitive predictions. PLoS Computational Biology, 2020, 16, e1008397.	3.2	6

#	Article	IF	Citations
293	A new domestic cat genome assembly based on long sequence reads empowers feline genomic medicine and identifies a novel gene for dwarfism. PLoS Genetics, 2020, 16, e1008926.	3.5	79
294	Characterization of Structural Variations in the Context of 3D Chromatin Structure. Molecules and Cells, 2019, 42, 512-522.	2.6	12
296	A Journey through Genetic Architecture and Predisposition of Coronary Artery Disease. Current Genomics, 2020, 21, 382-398.	1.6	3
297	Enhancing breakpoint resolution with deep segmentation model: A general refinement method for read-depth based structural variant callers. PLoS Computational Biology, 2021, 17, e1009186.	3.2	0
313	phasebook: haplotype-aware de novo assembly of diploid genomes from long reads. Genome Biology, 2021, 22, 299.	8.8	21
314	Haplotype-aware variant calling with PEPPER-Margin-DeepVariant enables high accuracy in nanopore long-reads. Nature Methods, 2021, 18, 1322-1332.	19.0	139
316	TensorSV: structural variation inference using tensors and variable topology neural networks. , 2020, , .		1
319	Resolving Breakpoints of Chromosomal Rearrangements at the Nucleotide Level Using Sanger Sequencing. Current Protocols in Human Genetics, 2020, 108, e107.	3.5	1
320	Computing the Rearrangement Distance of Natural Genomes. Lecture Notes in Computer Science, 2020, , 3-18.	1.3	5
322	Analysis of Cell and Nucleus Genome byÂNext-Generation Sequencing. , 2020, , 35-65.		0
323	Copy Number Variations of SCN5A in Brugada Syndrome. Japanese Journal of Electrocardiology, 2020, 40, 5-15.	0.0	0
324	Harnessing the power of multiâ€omics data for predicting climate change response. Journal of Animal Ecology, 2022, 91, 1064-1072.	2.8	16
325	Nanopore sequencing technology, bioinformatics and applications. Nature Biotechnology, 2021, 39, 1348-1365.	17.5	521
326	Combining callers improves the detection of copy number variants from whole-genome sequencing. European Journal of Human Genetics, 2022, 30, 178-186.	2.8	18
331	DNA methylation-calling tools for Oxford Nanopore sequencing: a survey and human epigenome-wide evaluation. Genome Biology, 2021, 22, 295.	8.8	6
332	Documentation of units of inheritance and their contribution to phenotype. , 2022, , 3-16.		0
333	DNA methylation-calling tools for Oxford Nanopore sequencing: a survey and human epigenome-wide evaluation. Genome Biology, 2021, 22, 295.	8.8	87
334	Evaluation of copy number variants for genetic hearing loss: a review of current approaches and recent findings. Human Genetics, 2022, 141, 387-400.	3.8	10

#	Article	IF	Citations
335	Evaluation of Germline Structural Variant Calling Methods for Nanopore Sequencing Data. Frontiers in Genetics, 2021, 12, 761791.	2.3	12
336	Binderâ€Free Ï‰â€Li₃V₂O₅ Catalytic Network with Multiâ€Polarization Centers Assists Lithiumâ€"Sulfur Batteries for Enhanced Kinetics Behavior. Advanced Functional Materials, 2022, 32, 2110665.	14.9	16
337	Accurate long-read de novo assembly evaluation with Inspector. Genome Biology, 2021, 22, 312.	8.8	46
338	Comprehensive characterization of copy number variation (CNV) called from array, long- and short-read data. BMC Genomics, 2021, 22, 826.	2.8	7
339	Structural variants in the Chinese population and their impact on phenotypes, diseases and population adaptation. Nature Communications, 2021, 12, 6501.	12.8	33
340	Long-read sequencing settings for efficient structural variation detection based on comprehensive evaluation. BMC Bioinformatics, 2021, 22, 552.	2.6	14
341	SVInterpreter: A Comprehensive Topologically Associated Domain-Based Clinical Outcome Prediction Tool for Balanced and Unbalanced Structural Variants. Frontiers in Genetics, 2021, 12, 757170.	2.3	5
342	CNV-P: a machine-learning framework for predicting high confident copy number variations. PeerJ, 2021, 9, e12564.	2.0	2
343	Emerging Insights Into Chronic Renal Disease Pathogenesis in Hypertension From Human and Animal Genomic Studies. Hypertension, 2021, 78, 1689-1700.	2.7	3
344	BreakNet: detecting deletions using long reads and a deep learning approach. BMC Bioinformatics, 2021, 22, 577.	2.6	5
345	IMDSVs: An integrated method based on machine learning and deep learning of calling structural variations from long-read data. , 2021, , .		0
346	Xq27.1 palindrome mediated interchromosomal insertion likely causes familial congenital bilateral laryngeal abductor paralysis (Plott syndrome). Journal of Human Genetics, 2022, 67, 405-410.	2.3	3
348	Born in the mitochondrion and raised in the nucleus: Evolution of a novel tandem repeat family in Medicago polymorpha (Fabaceae). Plant Journal, 2022, , .	5.7	5
349	Advanced techniques for gene heterogeneity research: Singleâ€cell sequencing and onâ€chip gene analysis systems. View, 2022, 3, .	5.3	9
350	Application and Challenge of 3rd Generation Sequencing for Clinical Bacterial Studies. International Journal of Molecular Sciences, 2022, 23, 1395.	4.1	24
351	LongPhase: an ultra-fast chromosome-scale phasing algorithm for small and large variants. Bioinformatics, 2022, 38, 1816-1822.	4.1	20
352	Optical genome mapping identifies rare structural variations as predisposition factors associated with severe COVID-19. IScience, 2022, 25, 103760.	4.1	15
353	Characterizing mobile element insertions in 5675 genomes. Nucleic Acids Research, 2022, 50, 2493-2508.	14.5	16

#	Article	IF	Citations
354	GCAT|Panel, a comprehensive structural variant haplotype map of the Iberian population from high-coverage whole-genome sequencing. Nucleic Acids Research, 2022, 50, 2464-2479.	14.5	6
355	High-speed rail model reveals the gene tandem amplification mediated by short repeated sequence in eukaryote. Scientific Reports, 2022, 12, 2289.	3.3	0
356	Pangenomics enables genotyping of known structural variants in 5202 diverse genomes. Science, 2021, 374, abg8871.	12.6	132
357	An Alu insertion map of the Indian population: identification and analysis in 1021 genomes of the IndiGen project. NAR Genomics and Bioinformatics, 2022, 4, lqac009.	3.2	1
358	Accelerating minimap2 for long-read sequencing applications on modern CPUs. Nature Computational Science, 2022, 2, 78-83.	8.0	19
359	Comparison of structural variants in the whole genome sequences of two Medicago truncatula ecotypes: Jemalong A17 and R108. BMC Plant Biology, 2022, 22, 77.	3.6	8
360	Combined use of Oxford Nanopore and Illumina sequencing yields insights into soybean structural variation biology. BMC Biology, 2022, 20, 53.	3.8	10
362	SNPs, short tandem repeats, and structural variants are responsible for differential gene expression across C57BL/6 and C57BL/10 substrains. Cell Genomics, 2022, 2, 100102.	6.5	9
364	Spinâ€"Phonon Coupling in Ferromagnetic Monolayer Chromium Tribromide. Advanced Materials, 2022, 34, e2108506.	21.0	8
366	A hidden layer of structural variation in transposable elements reveals potential genetic modifiers in human disease-risk loci. Genome Research, 2022, 32, 656-670.	5.5	13
367	Whole-genome sequencing of 1,171 elderly admixed individuals from Brazil. Nature Communications, 2022, 13, 1004.	12.8	35
368	Towards accurate and reliable resolution of structural variants for clinical diagnosis. Genome Biology, 2022, 23, 68.	8.8	34
369	Haplotype-resolved Chinese male genome assembly based on high-fidelity sequencing. Fundamental Research, 2022, 2, 946-953.	3.3	11
370	New Insights for Biosensing: Lessons from Microbial Defense Systems. Chemical Reviews, 2022, 122, 8126-8180.	47.7	15
371	Integrating whole-genome sequencing with multi-omic data reveals the impact of structural variants on gene regulation in the human brain. Nature Neuroscience, 2022, 25, 504-514.	14.8	27
372	European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the state of genetic testing for cardiac diseases. Europace, 2022, 24, 1307-1367.	1.7	108
374	Lessons learned: next-generation sequencing applied to undiagnosed genetic diseases. Journal of Clinical Investigation, 2022, 132, .	8.2	11
375	European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the State of Genetic Testing for Cardiac Diseases. Heart Rhythm, 2022, 19, e1-e60.	0.7	78

#	Article	IF	Citations
376	Familial long-read sequencing increases yield of de novo mutations. American Journal of Human Genetics, 2022, 109, 631-646.	6.2	32
377	A complete reference genome improves analysis of human genetic variation. Science, 2022, 376, eabl3533.	12.6	144
378	Segmental duplications and their variation in a complete human genome. Science, 2022, 376, eabj6965.	12.6	130
379	Long-read mapping to repetitive reference sequences using Winnowmap2. Nature Methods, 2022, 19, 705-710.	19.0	80
380	Searching thousands of genomes to classify somatic and novel structural variants using STIX. Nature Methods, 2022, 19, 445-448.	19.0	8
381	European Heart Rhythm Association (<scp>EHRA</scp>)/Heart Rhythm Society (<scp>HRS</scp>)/Asia Pacific Heart Rhythm Society (<scp>APHRS</scp>)/Latin American Heart Rhythm Society (<scp>LAHRS</scp>) Expert Consensus Statement on the state of genetic testing for cardiac diseases. Journal of Arrhythmia, 2022, 38, 491-553.	1.2	24
382	A benchmark of structural variation detection by long reads through a realistic simulated model. Genome Biology, 2021, 22, 342.	8.8	21
385	Hidden biases in germline structural variant detection. Genome Biology, 2021, 22, 347.	8.8	19
386	AluÂelement in the RNA binding motif protein, X-linked 2 (RBMX2) gene found to be linked to bipolar disorder. PLoS ONE, 2021, 16, e0261170.	2.5	2
388	Limitations of lymphoblastoid cell lines for establishing genetic reference datasets in the immunoglobulin loci. PLoS ONE, 2021, 16, e0261374.	2.5	4
389	Computational analysis of cancer genome sequencing data. Nature Reviews Genetics, 2022, 23, 298-314.	16.3	38
390	trfermikit: a tool to discover VNTR-associated deletions. Bioinformatics, 2022, 38, 1231-1234.	4.1	0
393	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
394	The Human Pangenome Project: a global resource to map genomic diversity. Nature, 2022, 604, 437-446.	27.8	192
395	Multiplex structural variant detection by whole-genome mapping and nanopore sequencing. Scientific Reports, 2022, 12, 6512.	3.3	3
396	Population-scale long-read sequencing uncovers transposable elements associated with gene expression variation and adaptive signatures in Drosophila. Nature Communications, 2022, 13, 1948.	12.8	53
397	Overview of structural variation calling: Simulation, identification, and visualization. Computers in Biology and Medicine, 2022, 145, 105534.	7.0	4
403	Bridging the gap: Short structural variants in the genetics of anorexia nervosa. International Journal of Eating Disorders, 2022, 55, 747-753.	4.0	1

#	Article	IF	Citations
404	Oxford Nanopore and Bionano Genomics technologies evaluation for plant structural variation detection. BMC Genomics, 2022, 23, 317.	2.8	4
405	Comprehensive evaluation of structural variant genotyping methods based on long-read sequencing data. BMC Genomics, 2022, 23, 324.	2.8	6
406	MAMnet: detecting and genotyping deletions and insertions based on long reads and a deep learning approach. Briefings in Bioinformatics, 2022, 23, .	6.5	4
407	SvAnna: efficient and accurate pathogenicity prediction of coding and regulatory structural variants in long-read genome sequencing. Genome Medicine, 2022, 14, 44.	8.2	7
409	Recurrent inversion polymorphisms in humans associate with genetic instability and genomic disorders. Cell, 2022, 185, 1986-2005.e26.	28.9	67
410	TT-Mars: structural variants assessment based on haplotype-resolved assemblies. Genome Biology, 2022, 23, 110.	8.8	9
411	Initial Analysis of Structural Variation Detections in Cattle Using Long-Read Sequencing Methods. Genes, 2022, 13, 828.	2.4	4
412	LT1, an ONT long-read-based assembly scaffolded with Hi-C data and polished with short reads. GigaByte, 0, 2022, 1-16.	0.0	0
413	A Draft Reference Genome Assembly of the Critically Endangered Black Abalone,Haliotis cracherodii. Journal of Heredity, 2022, 113, 665-672.	2.4	4
414	Dynamic Interplay between Structural Variations and 3D Genome Organization in Pancreatic Cancer. Advanced Science, 2022, 9, e2200818.	11.2	10
417	Population-scale genotyping of structural variation in the era of long-read sequencing. Computational and Structural Biotechnology Journal, 2022, 20, 2639-2647.	4.1	7
418	Flip-flop genomics: Charting inversions in the human population. Cell, 2022, 185, 1811-1813.	28.9	0
421	Exome-wide analysis of copy number variation shows association of the human leukocyte antigen region with asthma in UK Biobank. BMC Medical Genomics, 2022, 15, .	1.5	6
422	The missing heritability in type 1 diabetes. Diabetes, Obesity and Metabolism, 2022, 24, 1901-1911.	4.4	4
423	Integrating Genetic Structural Variations and Whole-Genome Sequencing Into Clinical Neurology. Neurology: Genetics, 2022, 8, e200005.	1.9	4
426	Chemoenzymatic labeling of DNA methylation patterns for single-molecule epigenetic mapping. Nucleic Acids Research, 2022, 50, e92-e92.	14.5	16
427	A phenome-wide association study identifies effects of copy-number variation of VNTRs and multicopy genes on multiple human traits. American Journal of Human Genetics, 2022, 109, 1065-1076.	6.2	12
428	Pharmacogenomics: Genetic Polymorphisms. , 2022, , 890-899.		0

#	Article	IF	Citations
429	Physlr: Next-Generation Physical Maps. Dna, 2022, 2, 116-130.	1.3	5
432	Reanalysis of exome negative patients with rare disease: a pragmatic workflow for diagnostic applications. Genome Medicine, 2022, 14, .	8.2	17
434	Polishing copy number variant calls on exome sequencing data via deep learning. Genome Research, 2022, 32, 1170-1182.	5.5	5
435	A multi-platform reference for somatic structural variation detection. Cell Genomics, 2022, 2, 100139.	6.5	10
436	A Map of 3â€² DNA Transduction Variants Mediated by Non-LTR Retroelements on 3202 Human Genomes. Biology, 2022, 11, 1032.	2.8	3
437	DeepLoop robustly maps chromatin interactions from sparse allele-resolved or single-cell Hi-C data at kilobase resolution. Nature Genetics, 2022, 54, 1013-1025.	21.4	19
440	Complex genomic rearrangements: an underestimated cause of rare diseases. Trends in Genetics, 2022, 38, 1134-1146.	6.7	19
444	Whole-genome long-read TAPS deciphers DNA methylation patterns at base resolution using PacBio SMRT sequencing technology. Nucleic Acids Research, 2022, 50, e104-e104.	14.5	10
446	Efficient detection and assembly of non-reference DNA sequences with synthetic long reads. Nucleic Acids Research, 2022, 50, e108-e108.	14.5	4
448	Photoâ€Enhanced Magnesiumâ€Ion Capacitors Using Photoactive Electrodes. Small, 2022, 18, .	10.0	10
449	Structural variants in the barley gene pool: precision and sensitivity to detect them using short-read sequencing and their association with gene expression and phenotypic variation. Theoretical and Applied Genetics, 2022, 135, 3511-3529.	3.6	5
450	Large Fragment InDels Reshape Genome Structure of Porcine Alveolar Macrophage 3D4/21 Cells. Genes, 2022, 13, 1515.	2.4	1
451	Rare coding variation provides insight into the genetic architecture and phenotypic context of autism. Nature Genetics, 2022, 54, 1320-1331.	21.4	155
452	Neuromuscular disorders: finding the missing genetic diagnoses. Trends in Genetics, 2022, 38, 956-971.	6.7	4
453	Challenges in quantifying genome erosion for conservation. Frontiers in Genetics, 0, 13, .	2.3	6
454	SVision: a deep learning approach to resolve complex structural variants. Nature Methods, 2022, 19, 1230-1233.	19.0	24
455	Evolutionary genomics of structural variation in the tea plant, <i>Camellia sinensis</i>. , 2022, 1, 1-11.		3
457	A survey of current methods to detect and genotype inversions. Human Mutation, 2022, 43, 1576-1589.	2.5	11

#	Article	IF	Citations
461	Genome sequence assembly algorithms and misassembly identification methods. Molecular Biology Reports, 2022, 49, 11133-11148.	2.3	2
462	A high-resolution map of small-scale inversions in the gibbon genome. Genome Research, 0, , gr.276960.122.	5.5	0
463	High-coverage whole-genome sequencing of the expanded 1000 Genomes Project cohort including 602 trios. Cell, 2022, 185, 3426-3440.e19.	28.9	285
464	Construction of a trio-based structural variation panel utilizing activated T lymphocytes and long-read sequencing technology. Communications Biology, 2022, 5, .	4.4	4
466	Structural Variants Identified Using Non-Mendelian Inheritance Patterns Advance the Mechanistic Understanding of Autism Spectrum Disorder. Human Genetics and Genomics Advances, 2022, , 100150.	1.7	3
468	Clinical Validation and Diagnostic Utility of Optical Genome Mapping for Enhanced Cytogenomic Analysis of Hematological Neoplasms. Journal of Molecular Diagnostics, 2022, 24, 1279-1291.	2.8	24
469	Human Retrotransposons and Effective Computational Detection Methods for Next-Generation Sequencing Data. Life, 2022, 12, 1583.	2.4	3
470	INSERT-seq enables high-resolution mapping of genomically integrated DNA using Nanopore sequencing. Genome Biology, 2022, 23, .	8.8	7
471	Characteristics and potential functional effects of long insertions in Asian butternuts. BMC Genomics, 2022, 23, .	2.8	0
472	Improvement of large copy number variant detection by whole genome nanopore sequencing. Journal of Advanced Research, 2022, , .	9.5	0
473	PGG.SV: a whole-genome-sequencing-based structural variant resource and data analysis platform. Nucleic Acids Research, 2023, 51, D1109-D1116.	14.5	3
474	Comprehensive analysis of structural variants in chickens using PacBio sequencing. Frontiers in Genetics, 0, 13, .	2.3	4
475	Assembly-free discovery of human novel sequences using long reads. DNA Research, 2022, 29, .	3.4	2
476	Integration of Hi-C with short and long-read genome sequencing reveals the structure of germline rearranged genomes. Nature Communications, 2022, 13, .	12.8	9
477	ConsensuSVâ€"from the whole-genome sequencing data to the complete variant list. Bioinformatics, 2022, 38, 5440-5442.	4.1	3
478	ParseCNV2: efficient sequencing tool for copy number variation genome-wide association studies. European Journal of Human Genetics, 2023, 31, 304-312.	2.8	4
479	Applications of Long-Read Sequencing Technology in Clinical Genomics. Advances in Molecular Pathology, 2022, 5, 85-108.	0.4	0
480	Identification of a novel large multigene deletion and a frameshift indel inPDE6Bas the underlying cause of early onset recessive rod-cone degeneration. Journal of Physical Education and Sports Management, 0, , mcs.a006247.	1.2	0

#	Article	IF	Citations
481	Retrotransposon insertions associated with risk of neurologic and psychiatric diseases. EMBO Reports, 2023, 24, .	4.5	3
482	Duet: SNP-assisted structural variant calling and phasing using Oxford nanopore sequencing. BMC Bioinformatics, 2022, 23, .	2.6	3
484	Low-Cost Genome-Scale Phasing with Barcode-Linked Sequencing. Methods in Molecular Biology, 2023, , 85-99.	0.9	0
485	Transposable element-mediated rearrangements are prevalent in human genomes. Nature Communications, 2022, 13, .	12.8	17
487	Recent advances and current challenges in population genomics of structural variation in animals and plants. Frontiers in Genetics, 0, 13, .	2.3	3
488	Parent-of-origin detection and chromosome-scale haplotyping using long-read DNA methylation sequencing and Strand-seq. Cell Genomics, 2023, 3, 100233.	6.5	8
489	Points to consider in the detection of germline structural variants using next-generation sequencing: A statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2023, 25, 100316.	2.4	7
490	Optical genome mapping and revisiting short-read genome sequencing data reveal previously overlooked structural variants disrupting retinal diseaseâ˜associated genes. Genetics in Medicine, 2023, 25, 100345.	2.4	9
492	Recent advances in cancer fusion transcript detection. Briefings in Bioinformatics, 2023, 24, .	6.5	13
493	Comparison of structural variants detected by PacBio-CLR and ONT sequencing in pear. BMC Genomics, 2022, 23, .	2.8	2
494	CONGA: Copy number variation genotyping in ancient genomes and low-coverage sequencing data. PLoS Computational Biology, 2022, 18, e1010788.	3.2	2
495	Haplotype-aware pantranscriptome analyses using spliced pangenome graphs. Nature Methods, 2023, 20, 239-247.	19.0	15
496	Advances in sequencing technologies for amyotrophic lateral sclerosis research. Molecular Neurodegeneration, 2023, 18, .	10.8	4
497	Deciphering the exact breakpoints of structural variations using long sequencing reads with DeBreak. Nature Communications, 2023, 14, .	12.8	10
498	Whole genome sequencing for USH2A-associated disease reveals several pathogenic deep-intronic variants that are amenable to splice correction. Human Genetics and Genomics Advances, 2023, 4, 100181.	1.7	4
501	A framework for associating structural variants with cell-specific transcription factors and histone modifications in defect phenotypes. , 2022, , .		1
504	Genetics in prenatal diagnosis. Singapore Medical Journal, 2023, 64, 27.	0.6	2
507	Jasmine and Iris: population-scale structural variant comparison and analysis. Nature Methods, 2023, 20, 408-417.	19.0	31

#	Article	IF	Citations
509	In it for the long run: perspectives on exploiting long-read sequencing in livestock for population scale studies of structural variants. Genetics Selection Evolution, 2023, 55, .	3.0	6
512	Long-read sequencing of diagnosis and post-therapy medulloblastoma reveals complex rearrangement patterns and epigenetic signatures. Cell Genomics, 2023, 3, 100281.	6.5	9
513	Genomeâ€Wide Analysis of Structural Variants in Parkinson Disease. Annals of Neurology, 2023, 93, 1012-1022.	5.3	10
516	Linear: a framework to enable existing software to resolve structural variants in long reads with flexible and efficient alignment-free statistical models. Briefings in Bioinformatics, 2023, 24, .	6.5	0
517	Genomic structural variation: A complex but important driver of human evolution. American Journal of Biological Anthropology, 2023, 181, 118-144.	1.1	3
518	Multisite Assessment of Optical Genome Mapping for Analysis of Structural Variants in Constitutional Postnatal Cases. Journal of Molecular Diagnostics, 2023, 25, 175-188.	2.8	14
519	MaxDEL: Accurate and Efficient Calling of Genomic Deletions from Single Molecular Real-time Sequencing Using Integrated Method. Current Bioinformatics, 2023, 18, .	1.5	0
520	Unified views on variant impact across many diseases. Trends in Genetics, 2023, 39, 442-450.	6.7	3
521	Stepwise use of genomics and transcriptomics technologies increases diagnostic yield in Mendelian disorders. Frontiers in Cell and Developmental Biology, 0, 11, .	3.7	2
522	INSnet: a method for detecting insertions based on deep learning network. BMC Bioinformatics, 2023, 24, .	2.6	1
525	Cue: a deep-learning framework for structural variant discovery and genotyping. Nature Methods, 2023, 20, 559-568.	19.0	8
526	Localized assembly for long reads enables genome-wide analysis of repetitive regions at single-base resolution in human genomes. Human Genomics, 2023, 17, .	2.9	2
528	Artificial intelligence in precision medicine. , 2023, , 531-569.		1
529	A collection of read depth profiles at structural variant breakpoints. Scientific Data, 2023, 10, .	5.3	2
530	Resolution of structural variation in diverse mouse genomes reveals chromatin remodeling due to transposable elements. Cell Genomics, 2023, 3, 100291.	6.5	20
532	Variant calling and benchmarking in an era of complete human genome sequences. Nature Reviews Genetics, 2023, 24, 464-483.	16.3	26
533	Long-Read DNA Sequencing: Recent Advances and Remaining Challenges. Annual Review of Genomics and Human Genetics, 2023, 24, 109-132.	6.2	10
534	Inversion polymorphism in a complete human genome assembly. Genome Biology, 2023, 24, .	8.8	7

#	Article	IF	Citations
535	A draft human pangenome reference. Nature, 2023, 617, 312-324.	27.8	187
536	Gaps and complex structurally variant loci in phased genome assemblies. Genome Research, 2023, 33, 496-510.	5.5	10
538	Pangenomic genotyping with the marker array. Algorithms for Molecular Biology, 2023, 18, .	1.2	3
539	High-performance ionic thermoelectric materials and emerging applications of ionic thermoelectric devices. Materials Today Energy, 2023, 36, 101342.	4.7	6
540	Applications of long-read sequencing to Mendelian genetics. Genome Medicine, 2023, 15, .	8.2	11
543	A survey of algorithms for the detection of genomic structural variants from long-read sequencing data. Nature Methods, 2023, 20, 1143-1158.	19.0	5
544	Disruption of regulatory domains and novel transcripts as diseaseâ€causing mechanisms. BioEssays, 2023, 45, .	2.5	3
547	Detection of trait-associated structural variations using short-read sequencing. Cell Genomics, 2023, 3, 100328.	6.5	0
548	Comparison and benchmark of structural variants detected from long read and long-read assembly. Briefings in Bioinformatics, 2023, 24, .	6.5	1
549	Graph construction method impacts variation representation and analyses in a bovine super-pangenome. Genome Biology, 2023, 24, .	8.8	12
550	Genomics in the long-read sequencing era. Trends in Genetics, 2023, 39, 649-671.	6.7	14
551	SVJedi-graph: improving the genotyping of close and overlapping structural variants with long reads using a variation graph. Bioinformatics, 2023, 39, i270-i278.	4.1	0
552	Targeted adaptive long-read sequencing for discovery of complex phased variants in inherited retinal disease patients. Scientific Reports, 2023, 13, .	3.3	3
554	Effects of spaced k-mers on alignment-free genotyping. Bioinformatics, 2023, 39, i213-i221.	4.1	1
558	Chromosomal inversion polymorphisms shape human brain morphology. Cell Reports, 2023, 42, 112896.	6.4	2
559	vamos: variable-number tandem repeats annotation using efficient motif sets. Genome Biology, 2023, 24, .	8.8	4
560	Linked-read based analysis of the medulloblastoma genome. Frontiers in Oncology, 0, 13, .	2.8	0
561	Beyond the exome: Whatâ€™s next in diagnostic testing for Mendelian conditions. American Journal of Human Genetics, 2023, 110, 1229-1248.	6.2	13

#	Article	IF	Citations
562	Boosting variant-calling performance with multi-platform sequencing data using Clair3-MP. BMC Bioinformatics, 2023, 24, .	2.6	0
563	UniAligner: a parameter-free framework for fast sequence alignment. Nature Methods, 0, , .	19.0	1
564	AML with complex karyotype: extreme genomic complexity revealed by combined long-read sequencing and Hi-C technology. Blood Advances, 2023, 7, 6520-6531.	5.2	3
565	Resolving complex structural variants via nanopore sequencing. Frontiers in Genetics, 0, 14, .	2.3	2
566	Systematic evaluation of genome sequencing for the diagnostic assessment of autism spectrum disorder and fetal structural anomalies. American Journal of Human Genetics, 2023, 110, 1454-1469.	6.2	7
567	A Toolbox for Visualization of Sequencing Coverage Signal. Biophysics (Russian Federation), 2023, 68, 195-198.	0.7	0
568	Whole genomic analysis reveals atypical non-homologous off-target large structural variants induced by CRISPR-Cas9-mediated genome editing. Nature Communications, 2023, 14, .	12.8	5
569	A genomic platform for surveillance and antigen discovery in Plasmodium spp. using long-read amplicon sequencing. Cell Reports Methods, 2023, , 100574.	2.9	0
570	Structural variants and short tandem repeats impact gene expression and splicing in bovine testis tissue. Genetics, 0, , .	2.9	1
571	IGHV allele similarity clustering improves genotype inference from adaptive immune receptor repertoire sequencing data. Nucleic Acids Research, 2023, 51, e86-e86.	14.5	4
572	Long-read whole-genome analysis of human single cells. Nature Communications, 2023, 14, .	12.8	9
573	A biallelic multiple nucleotide length polymorphism explains functional causality at 5p15.33 prostate cancer risk locus. Nature Communications, 2023, 14, .	12.8	3
574	Familial co-segregation and the emerging role of long-read sequencing to re-classify variants of uncertain significance in inherited retinal diseases. Npj Genomic Medicine, 2023, 8, .	3.8	1
575	Balanced chromosomal rearrangements implicate YIPF5 and SPATC1L in non-obstructive oligoasthenozoospermia and oligozoospermia and of a derivative chromosome 22 in recurrent miscarriage. Gene, 2023, 887, 147737.	2.2	0
576	HQAlign: aligning nanopore reads for SV detection using current-level modeling. Bioinformatics, 2023, 39, .	4.1	0
577	Delineation of two multi-invasion-induced rearrangement pathways that differently affect genome stability. Genes and Development, 2023, 37, 621-639.	5.9	2
578	New whole-genome alignment tools are needed for tapping into plant diversity. Trends in Plant Science, 2024, 29, 355-369.	8.8	2
579	kâ€merâ€based GWAS enhances the discovery of causal variants and candidate genes in soybean. Plant Genome, 2023, 16, .	2.8	1

#	Article	IF	Citations
580	Toward Cytogenomics. Journal of Molecular Diagnostics, 2023, 25, 796-805.	2.8	2
582	A Reference Genome Assembly and Initial Polymorphism Data for Aotus nancymaae. Developments in Primatology, 2023, , 251-264.	0.1	2
584	Genome-Wide Copy Number Variation and Structural Variation: A Novel Tool for Improved Livestock Genomic Selection. Livestock Diseases and Management, 2023, , 75-88.	0.5	0
585	Comparative Benchmarking of Optical Genome Mapping and Chromosomal Microarray Reveals High Technological Concordance in CNV Identification and Additional Structural Variant Refinement. Genes, 2023, 14, 1868.	2.4	0
586	Constructing founder sets under allelic and non-allelic homologous recombination. Algorithms for Molecular Biology, 2023, 18, .	1.2	1
587	Copy number variations and their effect on the plasma proteome. Genetics, 0, , .	2.9	0
589	Emerging Opportunities to Study Mobile Element Insertions and Their Source Elements in an Expanding Universe of Sequenced Human Genomes. Genes, 2023, 14, 1923.	2.4	1
591	The landscape of human SVA retrotransposons. Nucleic Acids Research, 2023, 51, 11453-11465.	14.5	2
592	Detection and annotation of transposable element insertions and deletions on the human genome using nanopore sequencing. IScience, 2023, 26, 108214.	4.1	0
593	The Role of Genetics in the Management of Heart Failure Patients. International Journal of Molecular Sciences, 2023, 24, 15221.	4.1	0
595	Envisioning a new era: Complete genetic information from routine, telomere-to-telomere genomes. American Journal of Human Genetics, 2023, 110, 1832-1840.	6.2	1
596	Cytogenetics Is a Science, Not a Technique! Why Optical Genome Mapping Is So Important to Clinical Genetic Laboratories. Cancers, 2023, 15, 5470.	3.7	1
597	Trio-binning of a hinny refines the comparative organization of the horse and donkey X chromosomes and reveals novel species-specific features. Scientific Reports, 2023, 13, .	3.3	0
598	PhenoSV: interpretable phenotype-aware model for the prioritization of genes affected by structural variants. Nature Communications, 2023, 14, .	12.8	0
599	Applications of advanced technologies for detecting genomic structural variation. Mutation Research - Reviews in Mutation Research, 2023, 792, 108475.	5.5	0
601	BLR: a flexible pipeline for haplotype analysis of multiple linked-read technologies. Nucleic Acids Research, 2023, 51, e114-e114.	14.5	0
602	Unravelling inversions: Technological advances, challenges, and potential impact on crop breeding. Plant Biotechnology Journal, 2024, 22, 544-554.	8.3	0
603	Bottomâ€Up Extrusionâ€Based Biofabrication of the Osteoid Niche. Macromolecular Bioscience, 0, , .	4.1	0

#	Article	IF	Citations
604	Quartet DNA reference materials and datasets for comprehensively evaluating germline variant calling performance. Genome Biology, 2023, 24, .	8.8	4
605	A 39â€‰kb structural variant causing Lynch Syndrome detected by optical genome mapping and nanopore sequencing. European Journal of Human Genetics, 0, , .	2.8	0
606	Whole-genome long-read sequencing downsampling and its effect on variant-calling precision and recall. Genome Research, 2023, 33, 2029-2040.	5.5	1
607	Advances in the discovery and analyses of human tandem repeats. Emerging Topics in Life Sciences, 2023, 7, 361-381.	2.6	1
608	Analytic Validation of Optical Genome Mapping in Hematological Malignancies. Biomedicines, 2023, 11, 3263.	3.2	1
609	Evolutionary insights from profiling LINE-1 activity at allelic resolution in a single human genome. EMBO Journal, 2024, 43, 112-131.	7.8	0
611	The landscape of genomic structural variation in Indigenous Australians. Nature, 2023, 624, 602-610.	27.8	1
613	Benchmarking long-read genome sequence alignment tools for human genomics applications. PeerJ, 0, 11, e16515.	2.0	0
614	Genomic Tools in Biological Invasions: Current State and Future Frontiers. Genome Biology and Evolution, 2024, 16, .	2.5	1
615	Longâ€read genome sequencing reveals a novel intronic retroelement insertion in <scp>NR5A1</scp> associated with 46,<scp>XY</scp> differences of sexual development. American Journal of Medical Genetics, Part A, 0, , .	1.2	0
616	ECOLE: Learning to call copy number variants on whole exome sequencing data. Nature Communications, 2024, 15, .	12.8	0
617	Detection of mosaic and population-level structural variants with Sniffles2. Nature Biotechnology, 0, , .	17.5	12
618	Detection of Constitutional Structural Variants by Optical Genome Mapping. Journal of Molecular Diagnostics, 2024, 26, 213-226.	2.8	0
619	Whole genome sequencing enables new genetic diagnosis for inherited retinal diseases by identifying pathogenic variants. Npj Genomic Medicine, 2024, 9, .	3.8	1
620	A graph clustering algorithm for detection and genotyping of structural variants from long reads. GigaScience, 2024, 13, .	6.4	0
621	Small polymorphisms are a source of ancestral bias in structural variant breakpoint placement. Genome Research, 2024, 34, 7-19.	5.5	0
622	Monitoring Genomic Structural Rearrangements Resulting from Gene Editing. Journal of Personalized Medicine, 2024, 14, 110.	2.5	0
624	A 25-year odyssey of genomic technology advances and structural variant discovery. Cell, 2024, 187, 1024-1037.	28.9	0

#	Article	IF	Citations
625	Utility of long-read sequencing for All of Us. Nature Communications, 2024, 15, .	12.8	1
626	DandD: Efficient measurement of sequence growth and similarity. IScience, 2024, 27, 109054.	4.1	0
627	A 3â€²UTR Insertion Is a Candidate Causal Variant at the TMEM106B Locus Associated With Increased Risk for FTLD-TDP. Neurology: Genetics, 2024, 10, .	1.9	0
628	Pangenome-genotyped structural variation improves molecular phenotype mapping in cattle. Genome Research, 2024, 34, 300-309.	5.5	0
629	Sequencing and characterizing short tandem repeats in the human genome. Nature Reviews Genetics, 0, , .	16.3	0
630	Assessing the efficacy of target adaptive sampling long-read sequencing through hereditary cancer patient genomes. Npj Genomic Medicine, 2024, 9, .	3.8	1
631	A comprehensive review of deep learning-based variant calling methods. Briefings in Functional Genomics, 0, , .	2.7	0
632	A long-read sequencing strategy with overlapping linkers on adjacent fragments (OLAF-Seq) for targeted resequencing and enrichment. Scientific Reports, 2024, 14, .	3.3	0
633	Optical Genome Mapping as a Potential Routine Clinical Diagnostic Method. Genes, 2024, 15, 342.	2.4	0
634	Benchmarking long-read aligners and SV callers for structural variation detection in Oxford nanopore sequencing data. Scientific Reports, 2024, 14, .	3.3	0
635	Tradeoffs in alignment and assembly-based methods for structural variant detection with long-read sequencing data. Nature Communications, 2024, 15, .	12.8	0