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List of Publications by Year in Descending Order

120 papers	6,536 citations	33 h-index	80 g-index
149 ext. papers	11,092 ext. citations	16.6 avg, IF	6.03 L-index

#	Paper	IF	Citations
120	Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting.. *New England Journal of Medicine*, **2022**,	59.2	10
119	Centers for Mendelian Genomics: A decade of facilitating gene discovery.. *Genetics in Medicine*, **2022**,	8.1	5
118	Curated variation benchmarks for challenging medically relevant autosomal genes.. *Nature Biotechnology*, **2022**,	44.5	12
117	Ultra-Rapid Nanopore Whole Genome Genetic Diagnosis of Dilated Cardiomyopathy in an Adolescent With Cardiogenic Shock.. *Circulation Genomic and Precision Medicine*, **2022**, CIRCGEN121003591	5.2	1
116	Accurate profiling of forensic autosomal STRs using the Oxford Nanopore Technologies MinION device. *Forensic Science International: Genetics*, **2022**, 56, 102629	4.3	0
115	Assessing reproducibility of inherited variants detected with short-read whole genome sequencing.. *Genome Biology*, **2022**, 23, 2	18.3	3
114	Rescuing low frequency variants within intra-host viral populations directly from Oxford Nanopore sequencing data.. *Nature Communications*, **2022**, 13, 1321	17.4	3
113	Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing.. *Nature Biotechnology*, **2022**,	44.5	4
112	Towards accurate and reliable resolution of structural variants for clinical diagnosis.. *Genome Biology*, **2022**, 23, 68	18.3	1
111	A complete reference genome improves analysis of human genetic variation.. *Science*, **2022**, 376, eabl3533	33.3	12
110	The complete sequence of a human genome.. *Science*, **2022**, 376, 44-53	33.3	107
109	PrecisionFDA Truth Challenge V2: Calling variants from short and long reads in difficult-to-map regions. *Cell Genomics*, **2022**, 2, 100129		4
108	Benchmarking challenging small variants with linked and long reads. *Cell Genomics*, **2022**, 2, 100128		2
107	An international virtual hackathon to build tools for the analysis of structural variants within species ranging from coronaviruses to vertebrates. *F1000Research*, **2021**, 10, 246	3.6	2
106	Shotgun transcriptome, spatial omics, and isothermal profiling of SARS-CoV-2 infection reveals unique host responses, viral diversification, and drug interactions. *Nature Communications*, **2021**, 12, 1660	17.4	60
105	muCNV: Genotyping Structural Variants for Population-level Sequencing. *Bioinformatics*, **2021**,	7.2	1
104	Optimized sample selection for cost-efficient long-read population sequencing. *Genome Research*, **2021**, 31, 910-918	9.7	2

103	Intronic Haplotypes in the GBA Gene Do Not Predict Age at Diagnosis of Parkinson's Disease. *Movement Disorders*, **2021**, 36, 1456-1460	7	2
102	Investigation of product-derived lymphoma following infusion of piggyBac-modified CD19 chimeric antigen receptor T cells. *Blood*, **2021**, 138, 1391-1405	2.2	26
101	Towards population-scale long-read sequencing. *Nature Reviews Genetics*, **2021**, 22, 572-587	30.1	29
100	Chromosome-scale, haplotype-resolved assembly of human genomes. *Nature Biotechnology*, **2021**, 39, 309-312	44.5	44
99	SARS-CoV-2 genomic diversity and the implications for qRT-PCR diagnostics and transmission. *Genome Research*, **2021**, 31, 635-644	9.7	20
98	Oligonucleotide capture sequencing of the SARS-CoV-2 genome and subgenomic fragments from COVID-19 individuals. *PLoS ONE*, **2021**, 16, e0244468	3.7	8
97	Vulcan: Improved long-read mapping and structural variant calling via dual-mode alignment. *GigaScience*, **2021**, 10,	7.6	1
96	High resolution copy number inference in cancer using short-molecule nanopore sequencing. *Nucleic Acids Research*, **2021**, 49, e124	20.1	1
95	Rescuing Low Frequency Variants within Intra-Host Viral Populations directly from Oxford Nanopore sequencing data **2021**,		3
94	Performance assessment of DNA sequencing platforms in the ABRF Next-Generation Sequencing Study. *Nature Biotechnology*, **2021**, 39, 1129-1140	44.5	10
93	PRINCESS: comprehensive detection of haplotype resolved SNVs, SVs, and methylation. *Genome Biology*, **2021**, 22, 268	18.3	0
92	An international virtual hackathon to build tools for the analysis of structural variants within species ranging from coronaviruses to vertebrates. *F1000Research*, **2021**, 10, 246	3.6	1
91	Hidden biases in germline structural variant detection.. *Genome Biology*, **2021**, 22, 347	18.3	3
90	Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. *Nature Biotechnology*, **2020**, 38, 1044-1053	44.5	143
89	A robust benchmark for detection of germline large deletions and insertions. *Nature Biotechnology*, **2020**, 38, 1347-1355	44.5	98
88	Major Impacts of Widespread Structural Variation on Gene Expression and Crop Improvement in Tomato. *Cell*, **2020**, 182, 145-161.e23	56.2	171
87	Discovery and population genomics of structural variation in a songbird genus. *Nature Communications*, **2020**, 11, 3403	17.4	27
86	Targeted nanopore sequencing with Cas9-guided adapter ligation. *Nature Biotechnology*, **2020**, 38, 433-438	44.5	113

85	Parliament2: Accurate structural variant calling at scale. *GigaScience*, **2020**, 9,	7.6	17
84	Shotgun Transcriptome and Isothermal Profiling of SARS-CoV-2 Infection Reveals Unique Host Responses, Viral Diversification, and Drug Interactions **2020**,		51
83	Hidden genomic diversity of SARS-CoV-2: implications for qRT-PCR diagnostics and transmission **2020**,		13
82	Oligonucleotide capture sequencing of the SARS-CoV-2 genome and subgenomic fragments from COVID-19 individuals **2020**,		8
81	Oligonucleotide Capture Sequencing of the SARS-CoV-2 Genome and Subgenomic Fragments from COVID-19 Individuals **2020**,		7
80	Simultaneous profiling of chromatin accessibility and methylation on human cell lines with nanopore sequencing. *Nature Methods*, **2020**, 17, 1191-1199	21.6	40
79	Complex mosaic structural variations in human fetal brains. *Genome Research*, **2020**, 30, 1695-1704	9.7	9
78	Comprehensive analysis of structural variants in breast cancer genomes using single-molecule sequencing. *Genome Research*, **2020**, 30, 1258-1273	9.7	25
77	A diploid assembly-based benchmark for variants in the major histocompatibility complex. *Nature Communications*, **2020**, 11, 4794	17.4	22
76	PhaseME: Automatic rapid assessment of phasing quality and phasing improvement. *GigaScience*, **2020**, 9,	7.6	5
75	Potential applications of nanopore sequencing for forensic analysis. *Forensic Science Review*, **2020**, 32, 23-54	1.5	0
74	Evaluation of computational genotyping of structural variation for clinical diagnoses. *GigaScience*, **2019**, 8,	7.6	18
73	Ancestral Admixture Is the Main Determinant of Global Biodiversity in Fission Yeast. *Molecular Biology and Evolution*, **2019**, 36, 1975-1989	8.3	29
72	Duplication of a domestication locus neutralized a cryptic variant that caused a breeding barrier in tomato. *Nature Plants*, **2019**, 5, 471-479	11.5	35
71	Atlas-CNV: a validated approach to call single-exon CNVs in the eMERGESeq gene panel. *Genetics in Medicine*, **2019**, 21, 2135-2144	8.1	13
70	Accurate circular consensus long-read sequencing improves variant detection and assembly of a human genome. *Nature Biotechnology*, **2019**, 37, 1155-1162	44.5	427
69	A Genocentric Approach to Discovery of Mendelian Disorders. *American Journal of Human Genetics*, **2019**, 105, 974-986	11	18
68	RaGOO: fast and accurate reference-guided scaffolding of draft genomes. *Genome Biology*, **2019**, 20, 224	18.3	173

67	A strategy for building and using a human reference pangenome. *F1000Research*, **2019**, 8, 1751	3.6	8
66	Combined transcriptome and proteome profiling reveals specific molecular brain signatures for sex, maturation and circalunar clock phase. *ELife*, **2019**, 8,	8.9	26
65	Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. *Cell*, **2019**, 176, 1310-1324.e10	56.2	34
64	A multi-task convolutional deep neural network for variant calling in single molecule sequencing. *Nature Communications*, **2019**, 10, 998	17.4	63
63	Paragraph: a graph-based structural variant genotyper for short-read sequence data. *Genome Biology*, **2019**, 20, 291	18.3	55
62	Approaches to Whole Mitochondrial Genome Sequencing on the Oxford Nanopore MinION. *Current Protocols in Human Genetics*, **2019**, 104, e94	3.2	4
61	Structural variant calling: the long and the short of it. *Genome Biology*, **2019**, 20, 246	18.3	141
60	Evaluation of the detection of GBA missense mutations and other variants using the Oxford Nanopore MinION. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e564	2.3	35
59	Genome-wide patterns of transposon proliferation in an evolutionary young hybrid fish. *Molecular Ecology*, **2019**, 28, 1491-1505	5.7	11
58	A strategy for building and using a human reference pangenome. *F1000Research*, **2019**, 8, 1751	3.6	2
57	Accurate detection of complex structural variations using single-molecule sequencing. *Nature Methods*, **2018**, 15, 461-468	21.6	585
56	Piercing the dark matter: bioinformatics of long-range sequencing and mapping. *Nature Reviews Genetics*, **2018**, 19, 329-346	30.1	250
55	Complex rearrangements and oncogene amplifications revealed by long-read DNA and RNA sequencing of a breast cancer cell line. *Genome Research*, **2018**, 28, 1126-1135	9.7	74
54	Transient structural variations have strong effects on quantitative traits and reproductive isolation in fission yeast. *Nature Communications*, **2017**, 8, 14061	17.4	212
53	GenomeScope: fast reference-free genome profiling from short reads. *Bioinformatics*, **2017**, 33, 2202-2204	7.4	540
52	Copy number increases of transposable elements and protein-coding genes in an invasive fish of hybrid origin. *Molecular Ecology*, **2017**, 26, 4712-4724	5.7	17
51	LRSim: A Linked-Reads Simulator Generating Insights for Better Genome Partitioning. *Computational and Structural Biotechnology Journal*, **2017**, 15, 478-484	6.8	20
50	DangerTrack: A scoring system to detect difficult-to-assess regions. *F1000Research*, **2017**, 6, 443	3.6	6

49	Tools for annotation and comparison of structural variation. *F1000Research*, **2017**, 6, 1795	3.6	11
48	The genomic basis of circadian and circalunar timing adaptations in a midge. *Nature*, **2016**, 540, 69-73	50.4	64
47	Phased diploid genome assembly with single-molecule real-time sequencing. *Nature Methods*, **2016**, 13, 1050-1054	21.6	1015
46	Chromosomal-Level Assembly of the Asian Seabass Genome Using Long Sequence Reads and Multi-layered Scaffolding. *PLoS Genetics*, **2016**, 12, e1005954	6	77
45	The pineapple genome and the evolution of CAM photosynthesis. *Nature Genetics*, **2015**, 47, 1435-42	36.3	309
44	Ectodysplasin signalling genes and phenotypic evolution in sculpins (Cottus). *Proceedings of the Royal Society B: Biological Sciences*, **2015**, 282,	4.4	5
43	The Candida albicans Histone Acetyltransferase Hat1 Regulates Stress Resistance and Virulence via Distinct Chromatin Assembly Pathways. *PLoS Pathogens*, **2015**, 11, e1005218	7.6	33
42	Decreased expression of endogenous feline leukemia virus in cat lymphomas: a case control study. *BMC Veterinary Research*, **2015**, 11, 90	2.7	3
41	Teaser: Individualized benchmarking and optimization of read mapping results for NGS data. *Genome Biology*, **2015**, 16, 235	18.3	21
40	ADAR2 induces reproducible changes in sequence and abundance of mature microRNAs in the mouse brain. *Nucleic Acids Research*, **2014**, 42, 12155-68	20.1	38
39	NextGenMap: fast and accurate read mapping in highly polymorphic genomes. *Bioinformatics*, **2013**, 29, 2790-1	7.2	246
38	Benefit-of-doubt (BOD) scoring: a sequencing-based method for SNP candidate assessment from high to medium read number data sets. *Genomics*, **2013**, 101, 204-9	4.3	
37	Updating benchtop sequencing performance comparison. *Nature Biotechnology*, **2013**, 31, 294-6	44.5	255
36	Adenosine deaminases that act on RNA induce reproducible changes in abundance and sequence of embryonic miRNAs. *Genome Research*, **2012**, 22, 1468-76	9.7	71
35	Advanced methylome analysis after bisulfite deep sequencing: an example in Arabidopsis. *PLoS ONE*, **2012**, 7, e41528	3.7	17
34	Methods developed during the first National Center for Biotechnology Information Structural Variation Codeathon at Baylor College of Medicine. *F1000Research*,9, 1141	3.6	
33	The population genomics of structural variation in a songbird genus		2
32	Transient structural variations have strong effects on quantitative traits and reproductive isolation in fission yeast		1

31	Phased Diploid Genome Assembly with Single Molecule Real-Time Sequencing	16
30	GenomeScope: Fast reference-free genome profiling from short reads	5
29	SplitThreader: Exploration and analysis of rearrangements in cancer genomes	8
28	LRSim: a Linked Reads Simulator generating insights for better genome partitioning	1
27	Accurate detection of complex structural variations using single molecule sequencing	33
26	Complex rearrangements and oncogene amplifications revealed by long-read DNA and RNA sequencing of a breast cancer cell line	6
25	Multi-Platform Assessment of DNA Sequencing Performance using Human and Bacterial Reference Genomes in the ABRF Next-Generation Sequencing Study	1
24	Benchmarking challenging small variants with linked and long reads	26
23	SVCollector: Optimized sample selection for cost-efficient long-read population sequencing	2
22	precisionFDA Truth Challenge V2: Calling variants from short- and long-reads in difficult-to-map regions	25
21	Detection of GBA missense mutations and other variants using the Oxford Nanopore MinION	2
20	xAtlas: Scalable small variant calling across heterogeneous next-generation sequencing experiments	15
19	Clairvoyante: a multi-task convolutional deep neural network for variant calling in Single Molecule Sequencing	11
18	SVCollector: Optimized sample selection for validating and long-read resequencing of structural variants	4
17	Parliament2: Fast Structural Variant Calling Using Optimized Combinations of Callers	12
16	Simultaneous profiling of chromatin accessibility and methylation on human cell lines with nanopore sequencing	15
15	Highly-accurate long-read sequencing improves variant detection and assembly of a human genome	29
14	Fast and accurate reference-guided scaffolding of draft genomes	15

13	Targeted Nanopore Sequencing with Cas9 for studies of methylation, structural variants, and mutations	26
12	Paragraph: A graph-based structural variant genotyper for short-read sequence data	8
11	A robust benchmark for germline structural variant detection	34
10	Efficient de novo assembly of eleven human genomes using PromethION sequencing and a novel nanopore toolkit	29
9	Accurate chromosome-scale haplotype-resolved assembly of human genomes	18
8	Comprehensive analysis of structural variants in breast cancer genomes using single molecule sequencing	3
7	The complete sequence of a human genome	58
6	Towards a Comprehensive Variation Benchmark for Challenging Medically-Relevant Autosomal Genes	8
5	A complete reference genome improves analysis of human genetic variation	9
4	Construction of a new chromosome-scale, long-read reference genome assembly of the Syrian hamster, Mesocricetus auratus	1
3	Truvari: Refined Structural Variant Comparison Preserves Allelic Diversity	1
2	Whole genome sequencing identifies common and rare structural variants contributing to hematologic traits in the NHLBI TOPMed program	1
1	The third international hackathon for applying insights into large-scale genomic composition to use cases in a wide range of organisms. *F1000Research*,11, 530	3.6