Fritz J Sedlazeck

List of Publications by Year
in descending order

87	14,707	76326 40	46799 89
papers	citations	h-index	g-index

149	149	149	17478
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Phased diploid genome assembly with single-molecule real-time sequencing. Nature Methods, 2016, 13, 1050-1054.	19.0	1,658
2	The complete sequence of a human genome. Science, 2022, 376, 44-53.	12.6	1,222
3	GenomeScope: fast reference-free genome profiling from short reads. Bioinformatics, 2017, 33, 2202-2204.	4.1	1,183
4	Accurate detection of complex structural variations using single-molecule sequencing. Nature Methods, 2018, 15, 461-468.	19.0	1,175
5	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
6	The pineapple genome and the evolution of CAM photosynthesis. Nature Genetics, 2015, 47, 1435-1442.	21.4	472
7	Transient structural variations have strong effects on quantitative traits and reproductive isolation in fission yeast. Nature Communications, 2017, 8, 14061.	12.8	472
8	RaGOO: fast and accurate reference-guided scaffolding of draft genomes. Genome Biology, 2019, 20, 224.	8.8	469
9	Major Impacts of Widespread Structural Variation on Gene Expression and Crop Improvement in Tomato. Cell, 2020, 182, 145-161.e23.	28.9	464
10	Updating benchtop sequencing performance comparison. Nature Biotechnology, 2013, 31, 294-296.	17.5	423
11	Structural variant calling: the long and the short of it. Genome Biology, 2019, 20, 246.	8.8	409
12	NextGenMap: fast and accurate read mapping in highly polymorphic genomes. Bioinformatics, 2013, 29, 2790-2791.	4.1	408
13	Piercing the dark matter: bioinformatics of long-range sequencing and mapping. Nature Reviews Genetics, 2018, 19, 329-346.	16.3	395
14	Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. Nature Biotechnology, 2020, 38, 1044-1053.	17.5	344
15	Targeted nanopore sequencing with Cas9-guided adapter ligation. Nature Biotechnology, 2020, 38, 433-438.	17.5	286
16	A robust benchmark for detection of germline large deletions and insertions. Nature Biotechnology, 2020, 38, 1347-1355.	17.5	233
17	Towards population-scale long-read sequencing. Nature Reviews Genetics, 2021, 22, 572-587.	16.3	163
18	A complete reference genome improves analysis of human genetic variation. Science, 2022, 376, eabl3533.	12.6	144

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19	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.	5.5	142
20	Simultaneous profiling of chromatin accessibility and methylation on human cell lines with nanopore sequencing. Nature Methods, 2020, 17, 1191-1199.	19.0	133
21	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.	12.8	132
22	Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting. New England Journal of Medicine, 2022, 386, 700-702.	27.0	116
23	Chromosome-scale, haplotype-resolved assembly of human genomes. Nature Biotechnology, 2021, 39, 309-312.	17.5	109
24	Chromosomal-Level Assembly of the Asian Seabass Genome Using Long Sequence Reads and Multi-layered Scaffolding. PLoS Genetics, 2016, 12, e1005954.	3.5	105
25	Paragraph: a graph-based structural variant genotyper for short-read sequence data. Genome Biology, 2019, 20, 291.	8.8	104
26	A multi-task convolutional deep neural network for variant calling in single molecule sequencing. Nature Communications, 2019, 10, 998.	12.8	102
27	The genomic basis of circadian and circalunar timing adaptations in a midge. Nature, 2016, 540, 69-73.	27.8	96
28	Curated variation benchmarks for challenging medically relevant autosomal genes. Nature Biotechnology, 2022, 40, 672-680.	17.5	90
29	Investigation of product-derived lymphoma following infusion of piggyBac-modified CD19 chimeric antigen receptor T cells. Blood, 2021, 138, 1391-1405.	1.4	87
30	Discovery and population genomics of structural variation in a songbird genus. Nature Communications, 2020, 11, 3403.	12.8	83
31	Adenosine deaminases that act on RNA induce reproducible changes in abundance and sequence of embryonic miRNAs. Genome Research, 2012, 22, 1468-1476.	5.5	80
32	Benchmarking challenging small variants with linked and long reads. Cell Genomics, 2022, 2, 100128.	6.5	77
33	Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. Cell, 2019, 176, 1310-1324.e10.	28.9	73
34	Comprehensive analysis of structural variants in breast cancer genomes using single-molecule sequencing. Genome Research, 2020, 30, 1258-1273.	5.5	72
35	PrecisionFDA Truth Challenge V2: Calling variants from short and long reads in difficult-to-map regions. Cell Genomics, 2022, 2, 100129.	6.5	72
36	Performance assessment of DNA sequencing platforms in the ABRF Next-Generation Sequencing Study. Nature Biotechnology, 2021, 39, 1129-1140.	17.5	69

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37	Duplication of a domestication locus neutralized a cryptic variant that caused a breeding barrier in tomato. Nature Plants, 2019, 5, 471-479.	9.3	66
38	Evaluation of the detection of GBA missense mutations and other variants using the Oxford Nanopore MinION. Molecular Genetics & Genomic Medicine, 2019, 7, e564.	1.2	65
39	A diploid assembly-based benchmark for variants in the major histocompatibility complex. Nature Communications, 2020, 11, 4794.	12.8	56
40	Parliament2: Accurate structural variant calling at scale. GigaScience, 2020, 9, .	6.4	51
41	Combined transcriptome and proteome profiling reveals specific molecular brain signatures for sex, maturation and circalunar clock phase. ELife, 2019, 8, .	6.0	51
42	Ancestral Admixture Is the Main Determinant of Global Biodiversity in Fission Yeast. Molecular Biology and Evolution, 2019, 36, 1975-1989.	8.9	50
43	The Candida albicans Histone Acetyltransferase Hat1 Regulates Stress Resistance and Virulence via Distinct Chromatin Assembly Pathways. PLoS Pathogens, 2015, 11, e1005218.	4.7	48
44	Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing. Nature Biotechnology, 2022, 40, 1035-1041.	17.5	45
45	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
46	ADAR2 induces reproducible changes in sequence and abundance of mature microRNAs in the mouse brain. Nucleic Acids Research, 2014, 42, 12155-12168.	14.5	42
47	LRSim: A Linked-Reads Simulator Generating Insights for Better Genome Partitioning. Computational and Structural Biotechnology Journal, 2017, 15, 478-484.	4.1	42
48	SARS-CoV-2 genomic diversity and the implications for qRT-PCR diagnostics and transmission. Genome Research, 2021, 31, 635-644.	5.5	39
49	Evaluation of computational genotyping of structural variation for clinical diagnoses. GigaScience, 2019, 8, .	6.4	36
50	Towards accurate and reliable resolution of structural variants for clinical diagnosis. Genome Biology, 2022, 23, 68.	8.8	34
51	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	6.2	30
52	Copy number increases of transposable elements and proteinâ€coding genes in an invasive fish of hybrid origin. Molecular Ecology, 2017, 26, 4712-4724.	3.9	28
53	PRINCESS: comprehensive detection of haplotype resolved SNVs, SVs, and methylation. Genome Biology, 2021, 22, 268.	8.8	28
54	Tools for annotation and comparison of structural variation. F1000Research, 2017, 6, 1795.	1.6	26

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55	Teaser: Individualized benchmarking and optimization of read mapping results for NGS data. Genome Biology, 2015, 16, 235.	8.8	25
56	TOP-LD: A tool to explore linkage disequilibrium with TOPMed whole-genome sequence data. American Journal of Human Genetics, 2022, 109, 1175-1181.	6.2	25
57	Complex mosaic structural variations in human fetal brains. Genome Research, 2020, 30, 1695-1704.	5.5	21
58	Oligonucleotide capture sequencing of the SARS-CoV-2 genome and subgenomic fragments from COVID-19 individuals. PLoS ONE, 2021, 16, e0244468.	2.5	20
59	Atlas-CNV: a validated approach to call single-exon CNVs in the eMERGESeq gene panel. Genetics in Medicine, 2019, 21, 2135-2144.	2.4	19
60	Advanced Methylome Analysis after Bisulfite Deep Sequencing: An Example in Arabidopsis. PLoS ONE, 2012, 7, e41528.	2.5	19
61	Hidden biases in germline structural variant detection. Genome Biology, 2021, 22, 347.	8.8	19
62	Genomeâ€wide patterns of transposon proliferation in an evolutionary young hybrid fish. Molecular Ecology, 2019, 28, 1491-1505.	3.9	18
63	Assessing reproducibility of inherited variants detected with short-read whole genome sequencing. Genome Biology, 2022, 23, 2.	8.8	18
64	Vulcan: Improved long-read mapping and structural variant calling via dual-mode alignment. GigaScience, 2021, 10, .	6.4	14
65	High resolution copy number inference in cancer using short-molecule nanopore sequencing. Nucleic Acids Research, 2021, 49, e124-e124.	14.5	14
66	A strategy for building and using a human reference pangenome. F1000Research, 2019, 8, 1751.	1.6	14
67	Approaches to Whole Mitochondrial Genome Sequencing on the Oxford Nanopore MinION. Current Protocols in Human Genetics, 2019, 104, e94.	3.5	13
68	Accurate profiling of forensic autosomal STRs using the Oxford Nanopore Technologies MinION device. Forensic Science International: Genetics, 2022, 56, 102629.	3.1	12
69	Rescuing low frequency variants within intra-host viral populations directly from Oxford Nanopore sequencing data. Nature Communications, 2022, 13, 1321.	12.8	11
70	DangerTrack: A scoring system to detect difficult-to-assess regions. F1000Research, 2017, 6, 443.	1.6	10
71	Ectodysplasin signalling genes and phenotypic evolution in sculpins (Cottus). Proceedings of the Royal Society B: Biological Sciences, 2015, 282, 20150746.	2.6	9
72	Decreased expression of endogenous feline leukemia virus in cat lymphomas: a case control study. BMC Veterinary Research, 2015, 11, 90.	1.9	8

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73	PhaseME: Automatic rapid assessment of phasing quality and phasing improvement. GigaScience, 2020, 9, .	6.4	8
74	Fully resolved assembly of Cryptosporidium parvum. GigaScience, 2022, 11, .	6.4	8
75	Searching thousands of genomes to classify somatic and novel structural variants using STIX. Nature Methods, 2022, 19, 445-448.	19.0	8
76	muCNV: genotyping structural variants for population-level sequencing. Bioinformatics, 2021, 37, 2055-2057.	4.1	7
77	Intronic Haplotypes in the <scp>GBA</scp> Gene Do Not Predict Age at Diagnosis of Parkinson's Disease. Movement Disorders, 2021, 36, 1456-1460.	3.9	5
78	A strategy for building and using a human reference pangenome. F1000Research, 2019, 8, 1751.	1.6	5
79	Potential applications of nanopore sequencing for forensic analysis. Forensic Science Review, 2020, 32, 23-54.	0.6	5
80	Optimized sample selection for cost-efficient long-read population sequencing. Genome Research, 2021, 31, 910-918.	5.5	4
81	AnÂinternationalÂvirtualÂhackathon toÂbuildÂtools for theÂanalysis ofÂstructuralÂvariants withinÂspeciesÂranging fromÂcoronaviruses toÂvertebrates. F1000Research, 2021, 10, 246.	1.6	3
82	Ultra-Rapid Nanopore Whole Genome Genetic Diagnosis of Dilated Cardiomyopathy in an Adolescent With Cardiogenic Shock. Circulation Genomic and Precision Medicine, 2022, 15, CIRCGEN121003591.	3.6	3
83	AnÂinternationalÂvirtualÂhackathon toÂbuildÂtools for theÂanalysis ofÂstructuralÂvariants withinÂspeciesÂranging fromÂcoronaviruses toÂvertebrates. F1000Research, 2021, 10, 246.	1.6	2
84	Long-read sequencing for diagnosis in the Undiagnosed Diseases Network. Molecular Genetics and Metabolism, 2021, 132, S253-S254.	1.1	1
85	The third international hackathon for applying insights into large-scale genomic composition to use cases in a wide range of organisms. F1000Research, 0, 11, 530.	1.6	1
86	Benefit-of-doubt (BOD) scoring: A sequencing-based method for SNP candidate assessment from high to medium read number data sets. Genomics, 2013, 101, 204-209.	2.9	0
87	Methods developed during the first National Center for Biotechnology Information Structural Variation Codeathon at Baylor College of Medicine. F1000Research, 0, 9, 1141.	1.6	0