

Fritz J Sedlazeck

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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 | Phased diploid genome assembly with single-molecule real-time sequencing. <i>Nature Methods</i> , 2016 , 13, 1050-1054 | 21.6 | 1015 |
| 119 | Accurate detection of complex structural variations using single-molecule sequencing. <i>Nature Methods</i> , 2018 , 15, 461-468 | 21.6 | 585 |
| 118 | GenomeScope: fast reference-free genome profiling from short reads. <i>Bioinformatics</i> , 2017 , 33, 2202-2204 | 21.6 | 540 |
| 117 | Accurate circular consensus long-read sequencing improves variant detection and assembly of a human genome. <i>Nature Biotechnology</i> , 2019 , 37, 1155-1162 | 44.5 | 427 |
| 116 | The pineapple genome and the evolution of CAM photosynthesis. <i>Nature Genetics</i> , 2015 , 47, 1435-42 | 36.3 | 309 |
| 115 | Updating benchtop sequencing performance comparison. <i>Nature Biotechnology</i> , 2013 , 31, 294-6 | 44.5 | 255 |
| 114 | Piercing the dark matter: bioinformatics of long-range sequencing and mapping. <i>Nature Reviews Genetics</i> , 2018 , 19, 329-346 | 30.1 | 250 |
| 113 | NextGenMap: fast and accurate read mapping in highly polymorphic genomes. <i>Bioinformatics</i> , 2013 , 29, 2790-1 | 7.2 | 246 |
| 112 | Transient structural variations have strong effects on quantitative traits and reproductive isolation in fission yeast. <i>Nature Communications</i> , 2017 , 8, 14061 | 17.4 | 212 |
| 111 | RaGOO: fast and accurate reference-guided scaffolding of draft genomes. <i>Genome Biology</i> , 2019 , 20, 224 | 18.3 | 173 |
| 110 | Major Impacts of Widespread Structural Variation on Gene Expression and Crop Improvement in Tomato. <i>Cell</i> , 2020 , 182, 145-161.e23 | 56.2 | 171 |
| 109 | Nanopore sequencing and the Shasta toolkit enable efficient de novo assembly of eleven human genomes. <i>Nature Biotechnology</i> , 2020 , 38, 1044-1053 | 44.5 | 143 |
| 108 | Structural variant calling: the long and the short of it. <i>Genome Biology</i> , 2019 , 20, 246 | 18.3 | 141 |
| 107 | Targeted nanopore sequencing with Cas9-guided adapter ligation. <i>Nature Biotechnology</i> , 2020 , 38, 433-435 | 44.5 | 113 |
| 106 | The complete sequence of a human genome.. <i>Science</i> , 2022 , 376, 44-53 | 33.3 | 107 |
| 105 | A robust benchmark for detection of germline large deletions and insertions. <i>Nature Biotechnology</i> , 2020 , 38, 1347-1355 | 44.5 | 98 |
| 104 | Chromosomal-Level Assembly of the Asian Seabass Genome Using Long Sequence Reads and Multi-layered Scaffolding. <i>PLoS Genetics</i> , 2016 , 12, e1005954 | 6 | 77 |

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| 103 | Complex rearrangements and oncogene amplifications revealed by long-read DNA and RNA sequencing of a breast cancer cell line. <i>Genome Research</i> , 2018 , 28, 1126-1135 | 9.7 | 74 |
| 102 | Adenosine deaminases that act on RNA induce reproducible changes in abundance and sequence of embryonic miRNAs. <i>Genome Research</i> , 2012 , 22, 1468-76 | 9.7 | 71 |
| 101 | The genomic basis of circadian and circalunar timing adaptations in a midge. <i>Nature</i> , 2016 , 540, 69-73 | 50.4 | 64 |
| 100 | A multi-task convolutional deep neural network for variant calling in single molecule sequencing. <i>Nature Communications</i> , 2019 , 10, 998 | 17.4 | 63 |
| 99 | Shotgun transcriptome, spatial omics, and isothermal profiling of SARS-CoV-2 infection reveals unique host responses, viral diversification, and drug interactions. <i>Nature Communications</i> , 2021 , 12, 1660 | 17.4 | 60 |
| 98 | The complete sequence of a human genome | | 58 |
| 97 | Paragraph: a graph-based structural variant genotyper for short-read sequence data. <i>Genome Biology</i> , 2019 , 20, 291 | 18.3 | 55 |
| 96 | Shotgun Transcriptome and Isothermal Profiling of SARS-CoV-2 Infection Reveals Unique Host Responses, Viral Diversification, and Drug Interactions 2020 , | | 51 |
| 95 | Chromosome-scale, haplotype-resolved assembly of human genomes. <i>Nature Biotechnology</i> , 2021 , 39, 309-312 | 44.5 | 44 |
| 94 | Simultaneous profiling of chromatin accessibility and methylation on human cell lines with nanopore sequencing. <i>Nature Methods</i> , 2020 , 17, 1191-1199 | 21.6 | 40 |
| 93 | ADAR2 induces reproducible changes in sequence and abundance of mature microRNAs in the mouse brain. <i>Nucleic Acids Research</i> , 2014 , 42, 12155-68 | 20.1 | 38 |
| 92 | Duplication of a domestication locus neutralized a cryptic variant that caused a breeding barrier in tomato. <i>Nature Plants</i> , 2019 , 5, 471-479 | 11.5 | 35 |
| 91 | Evaluation of the detection of GBA missense mutations and other variants using the Oxford Nanopore MinION. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e564 | 2.3 | 35 |
| 90 | A robust benchmark for germline structural variant detection | | 34 |
| 89 | Megabase Length Hypermutation Accompanies Human Structural Variation at 17p11.2. <i>Cell</i> , 2019 , 176, 1310-1324.e10 | 56.2 | 34 |
| 88 | The <i>Candida albicans</i> Histone Acetyltransferase Hat1 Regulates Stress Resistance and Virulence via Distinct Chromatin Assembly Pathways. <i>PLoS Pathogens</i> , 2015 , 11, e1005218 | 7.6 | 33 |
| 87 | Accurate detection of complex structural variations using single molecule sequencing | | 33 |
| 86 | Ancestral Admixture Is the Main Determinant of Global Biodiversity in Fission Yeast. <i>Molecular Biology and Evolution</i> , 2019 , 36, 1975-1989 | 8.3 | 29 |

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| 85 | Highly-accurate long-read sequencing improves variant detection and assembly of a human genome | | 29 |
| 84 | Efficient de novo assembly of eleven human genomes using PromethION sequencing and a novel nanopore toolkit | | 29 |
| 83 | Towards population-scale long-read sequencing. <i>Nature Reviews Genetics</i> , 2021 , 22, 572-587 | 30.1 | 29 |
| 82 | Discovery and population genomics of structural variation in a songbird genus. <i>Nature Communications</i> , 2020 , 11, 3403 | 17.4 | 27 |
| 81 | Combined transcriptome and proteome profiling reveals specific molecular brain signatures for sex, maturation and circalunar clock phase. <i>ELife</i> , 2019 , 8, | 8.9 | 26 |
| 80 | Benchmarking challenging small variants with linked and long reads | | 26 |
| 79 | Targeted Nanopore Sequencing with Cas9 for studies of methylation, structural variants, and mutations | | 26 |
| 78 | Investigation of product-derived lymphoma following infusion of piggyBac-modified CD19 chimeric antigen receptor T cells. <i>Blood</i> , 2021 , 138, 1391-1405 | 2.2 | 26 |
| 77 | precisionFDA Truth Challenge V2: Calling variants from short- and long-reads in difficult-to-map regions | | 25 |
| 76 | Comprehensive analysis of structural variants in breast cancer genomes using single-molecule sequencing. <i>Genome Research</i> , 2020 , 30, 1258-1273 | 9.7 | 25 |
| 75 | A diploid assembly-based benchmark for variants in the major histocompatibility complex. <i>Nature Communications</i> , 2020 , 11, 4794 | 17.4 | 22 |
| 74 | Teaser: Individualized benchmarking and optimization of read mapping results for NGS data. <i>Genome Biology</i> , 2015 , 16, 235 | 18.3 | 21 |
| 73 | LRSim: A Linked-Reads Simulator Generating Insights for Better Genome Partitioning. <i>Computational and Structural Biotechnology Journal</i> , 2017 , 15, 478-484 | 6.8 | 20 |
| 72 | SARS-CoV-2 genomic diversity and the implications for qRT-PCR diagnostics and transmission. <i>Genome Research</i> , 2021 , 31, 635-644 | 9.7 | 20 |
| 71 | Evaluation of computational genotyping of structural variation for clinical diagnoses. <i>GigaScience</i> , 2019 , 8, | 7.6 | 18 |
| 70 | A Genocentric Approach to Discovery of Mendelian Disorders. <i>American Journal of Human Genetics</i> , 2019 , 105, 974-986 | 11 | 18 |
| 69 | Accurate chromosome-scale haplotype-resolved assembly of human genomes | | 18 |
| 68 | Copy number increases of transposable elements and protein-coding genes in an invasive fish of hybrid origin. <i>Molecular Ecology</i> , 2017 , 26, 4712-4724 | 5.7 | 17 |

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| 67 | Advanced methylome analysis after bisulfite deep sequencing: an example in Arabidopsis. <i>PLoS ONE</i> , 2012 , 7, e41528 | 3.7 | 17 |
| 66 | Parliament2: Accurate structural variant calling at scale. <i>GigaScience</i> , 2020 , 9, | 7.6 | 17 |
| 65 | Phased Diploid Genome Assembly with Single Molecule Real-Time Sequencing | | 16 |
| 64 | xAtlas: Scalable small variant calling across heterogeneous next-generation sequencing experiments | | 15 |
| 63 | Simultaneous profiling of chromatin accessibility and methylation on human cell lines with nanopore sequencing | | 15 |
| 62 | Fast and accurate reference-guided scaffolding of draft genomes | | 15 |
| 61 | Atlas-CNV: a validated approach to call single-exon CNVs in the eMERGESeq gene panel. <i>Genetics in Medicine</i> , 2019 , 21, 2135-2144 | 8.1 | 13 |
| 60 | Hidden genomic diversity of SARS-CoV-2: implications for qRT-PCR diagnostics and transmission 2020 , | | 13 |
| 59 | Curated variation benchmarks for challenging medically relevant autosomal genes.. <i>Nature Biotechnology</i> , 2022 , | 44.5 | 12 |
| 58 | Parliament2: Fast Structural Variant Calling Using Optimized Combinations of Callers | | 12 |
| 57 | A complete reference genome improves analysis of human genetic variation.. <i>Science</i> , 2022 , 376, eabl35333 | 33.3 | 12 |
| 56 | Tools for annotation and comparison of structural variation. <i>F1000Research</i> , 2017 , 6, 1795 | 3.6 | 11 |
| 55 | Clairvoyante: a multi-task convolutional deep neural network for variant calling in Single Molecule Sequencing | | 11 |
| 54 | Genome-wide patterns of transposon proliferation in an evolutionary young hybrid fish. <i>Molecular Ecology</i> , 2019 , 28, 1491-1505 | 5.7 | 11 |
| 53 | Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting.. <i>New England Journal of Medicine</i> , 2022 , | 59.2 | 10 |
| 52 | Performance assessment of DNA sequencing platforms in the ABRF Next-Generation Sequencing Study. <i>Nature Biotechnology</i> , 2021 , 39, 1129-1140 | 44.5 | 10 |
| 51 | Complex mosaic structural variations in human fetal brains. <i>Genome Research</i> , 2020 , 30, 1695-1704 | 9.7 | 9 |
| 50 | A complete reference genome improves analysis of human genetic variation | | 9 |

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| 49 | A strategy for building and using a human reference pangenome. <i>F1000Research</i> , 2019 , 8, 1751 | 3.6 | 8 |
| 48 | SplitThreader: Exploration and analysis of rearrangements in cancer genomes | | 8 |
| 47 | Oligonucleotide capture sequencing of the SARS-CoV-2 genome and subgenomic fragments from COVID-19 individuals 2020 , | | 8 |
| 46 | Paragraph: A graph-based structural variant genotyper for short-read sequence data | | 8 |
| 45 | Towards a Comprehensive Variation Benchmark for Challenging Medically-Relevant Autosomal Genes | | 8 |
| 44 | Oligonucleotide capture sequencing of the SARS-CoV-2 genome and subgenomic fragments from COVID-19 individuals. <i>PLoS ONE</i> , 2021 , 16, e0244468 | 3.7 | 8 |
| 43 | Oligonucleotide Capture Sequencing of the SARS-CoV-2 Genome and Subgenomic Fragments from COVID-19 Individuals 2020 , | | 7 |
| 42 | DangerTrack: A scoring system to detect difficult-to-assess regions. <i>F1000Research</i> , 2017 , 6, 443 | 3.6 | 6 |
| 41 | Complex rearrangements and oncogene amplifications revealed by long-read DNA and RNA sequencing of a breast cancer cell line | | 6 |
| 40 | Ectodysplasin signalling genes and phenotypic evolution in sculpins (<i>Cottus</i>). <i>Proceedings of the Royal Society B: Biological Sciences</i> , 2015 , 282, | 4.4 | 5 |
| 39 | Centers for Mendelian Genomics: A decade of facilitating gene discovery.. <i>Genetics in Medicine</i> , 2022 , | 8.1 | 5 |
| 38 | GenomeScope: Fast reference-free genome profiling from short reads | | 5 |
| 37 | PhaseME: Automatic rapid assessment of phasing quality and phasing improvement. <i>GigaScience</i> , 2020 , 9, | 7.6 | 5 |
| 36 | SVCollector: Optimized sample selection for validating and long-read resequencing of structural variants | | 4 |
| 35 | Approaches to Whole Mitochondrial Genome Sequencing on the Oxford Nanopore MinION. <i>Current Protocols in Human Genetics</i> , 2019 , 104, e94 | 3.2 | 4 |
| 34 | Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing.. <i>Nature Biotechnology</i> , 2022 , | 44.5 | 4 |
| 33 | PrecisionFDA Truth Challenge V2: Calling variants from short and long reads in difficult-to-map regions. <i>Cell Genomics</i> , 2022 , 2, 100129 | | 4 |
| 32 | Decreased expression of endogenous feline leukemia virus in cat lymphomas: a case control study. <i>BMC Veterinary Research</i> , 2015 , 11, 90 | 2.7 | 3 |

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| 31 | Assessing reproducibility of inherited variants detected with short-read whole genome sequencing.. <i>Genome Biology</i> , 2022 , 23, 2 | 18.3 | 3 |
| 30 | Comprehensive analysis of structural variants in breast cancer genomes using single molecule sequencing | | 3 |
| 29 | Rescuing Low Frequency Variants within Intra-Host Viral Populations directly from Oxford Nanopore sequencing data 2021 , | | 3 |
| 28 | Rescuing low frequency variants within intra-host viral populations directly from Oxford Nanopore sequencing data.. <i>Nature Communications</i> , 2022 , 13, 1321 | 17.4 | 3 |
| 27 | Hidden biases in germline structural variant detection.. <i>Genome Biology</i> , 2021 , 22, 347 | 18.3 | 3 |
| 26 | The population genomics of structural variation in a songbird genus | | 2 |
| 25 | SVCollector: Optimized sample selection for cost-efficient long-read population sequencing | | 2 |
| 24 | Detection of GBA missense mutations and other variants using the Oxford Nanopore MinION | | 2 |
| 23 | An International Virtual Hackathon to Build Tools for the Analysis of Structural Variants within Species Ranging from Coronaviruses to Vertebrates. <i>F1000Research</i> , 2021 , 10, 246 | 3.6 | 2 |
| 22 | Optimized sample selection for cost-efficient long-read population sequencing. <i>Genome Research</i> , 2021 , 31, 910-918 | 9.7 | 2 |
| 21 | Intronic Haplotypes in the GBA Gene Do Not Predict Age at Diagnosis of Parkinson's Disease. <i>Movement Disorders</i> , 2021 , 36, 1456-1460 | 7 | 2 |
| 20 | A strategy for building and using a human reference pangenome. <i>F1000Research</i> , 2019 , 8, 1751 | 3.6 | 2 |
| 19 | Benchmarking challenging small variants with linked and long reads. <i>Cell Genomics</i> , 2022 , 2, 100128 | | 2 |
| 18 | Ultra-Rapid Nanopore Whole Genome Genetic Diagnosis of Dilated Cardiomyopathy in an Adolescent With Cardiogenic Shock.. <i>Circulation Genomic and Precision Medicine</i> , 2022 , CIRCGEN121003591 | 5.3 | 1 |
| 17 | Transient structural variations have strong effects on quantitative traits and reproductive isolation in fission yeast | | 1 |
| 16 | LRSim: a Linked Reads Simulator generating insights for better genome partitioning | | 1 |
| 15 | Multi-Platform Assessment of DNA Sequencing Performance using Human and Bacterial Reference Genomes in the ABRF Next-Generation Sequencing Study | | 1 |
| 14 | muCNV: Genotyping Structural Variants for Population-level Sequencing. <i>Bioinformatics</i> , 2021 , | 7.2 | 1 |

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| 13 | Construction of a new chromosome-scale, long-read reference genome assembly of the Syrian hamster, <i>Mesocricetus auratus</i> | | 1 |
| 12 | Vulcan: Improved long-read mapping and structural variant calling via dual-mode alignment. <i>GigaScience</i> , 2021 , 10, | 7.6 | 1 |
| 11 | High resolution copy number inference in cancer using short-molecule nanopore sequencing. <i>Nucleic Acids Research</i> , 2021 , 49, e124 | 20.1 | 1 |
| 10 | An International Virtual Hackathon to build tools for the analysis of structural variants within species ranging from coronaviruses to vertebrates. <i>F1000Research</i> , 2021 , 10, 246 | 3.6 | 1 |
| 9 | Truvari: Refined Structural Variant Comparison Preserves Allelic Diversity | | 1 |
| 8 | Towards accurate and reliable resolution of structural variants for clinical diagnosis.. <i>Genome Biology</i> , 2022 , 23, 68 | 18.3 | 1 |
| 7 | Whole genome sequencing identifies common and rare structural variants contributing to hematologic traits in the NHLBI TOPMed program | | 1 |
| 6 | Accurate profiling of forensic autosomal STRs using the Oxford Nanopore Technologies MinION device. <i>Forensic Science International: Genetics</i> , 2022 , 56, 102629 | 4.3 | 0 |
| 5 | PRINCESS: comprehensive detection of haplotype resolved SNVs, SVs, and methylation. <i>Genome Biology</i> , 2021 , 22, 268 | 18.3 | 0 |
| 4 | Potential applications of nanopore sequencing for forensic analysis. <i>Forensic Science Review</i> , 2020 , 32, 23-54 | 1.5 | 0 |
| 3 | Benefit-of-doubt (BOD) scoring: a sequencing-based method for SNP candidate assessment from high to medium read number data sets. <i>Genomics</i> , 2013 , 101, 204-9 | 4.3 | |
| 2 | Methods developed during the first National Center for Biotechnology Information Structural Variation Codeathon at Baylor College of Medicine. <i>F1000Research</i> , 9, 1141 | 3.6 | |
| 1 | The third international hackathon for applying insights into large-scale genomic composition to use cases in a wide range of organisms. <i>F1000Research</i> , 11, 530 | 3.6 | |