# Jesper Eisfeldt

List of Publications by Year
in descending order



| 34 | 918 | 623734 | 526287 |
|---|---|---|---|
| papers | citations | 14 h-index | 27 g-index |

| 39 | 39 | 39 | 1639 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | SweGen: a whole-genome data resource of genetic variability in a cross-section of the Swedish population. European Journal of Human Genetics, 2017, 25, 1253-1260. | 2.8 | 148 |
| 2 | Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40. | 8.2 | 116 |
| 3 | Sarek: A portable workflow for whole-genome sequencing analysis of germline and somatic variants. F1000Research, 2020, 9, 63. | 1.6 | 89 |
| 4 | From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. Genome Medicine, 2019, 11, 68. | 8.2 | 88 |
| 5 | TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664. | 1.6 | 76 |
| 6 | TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664. | 1.6 | 51 |
| 7 | Comprehensive structural variation genome map of individuals carrying complex chromosomal rearrangements. PLoS Genetics, 2019, 15, e1007858. | 3.5 | 36 |
| 8 | Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780. | 3.5 | 28 |
| 9 | Discovery of Novel Sequences in 1,000 Swedish Genomes. Molecular Biology and Evolution, 2020, 37, 18-30. | 8.9 | 25 |
| 10 | A Large Inversion Involving GNAS Exon A/B and All Exons Encoding GsÎ± Is Associated With Autosomal Dominant Pseudohypoparathyroidism Type Ib (PHP1B). Journal of Bone and Mineral Research, 2017, 32, 776-783. | 2.8 | 22 |
| 11 | Sarek: A portable workflow for whole-genome sequencing analysis of germline and somatic variants. F1000Research, 2020, 9, 63. | 1.6 | 21 |
| 12 | High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. Journal of Human Genetics, 2021, 66, 995-1008. | 2.3 | 19 |
| 13 | AMYCNE: Confident copy number assessment using whole genome sequencing data. PLoS ONE, 2018, 13, e0189710. | 2.5 | 19 |
| 14 | Chromatin interactions in differentiating keratinocytes reveal novel atopic dermatitisâ€“ and psoriasis-associated genes. Journal of Allergy and Clinical Immunology, 2021, 147, 1742-1752. | 2.9 | 18 |
| 15 | Targeted copy number screening highlights an intragenic deletion of WDR63 as the likely cause of human occipital encephalocele and abnormal CNS development in zebrafish. Human Mutation, 2018, 39, 495-505. | 2.5 | 17 |
| 16 | Wholeâ€genome sequencing reveals complex chromosome rearrangement disrupting <scp>NIPBL</scp> in infant with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1143-1151. | 1.2 | 17 |
| 17 | Alu-Alu mediated intragenic duplications in IFT81 and MATN3 are associated with skeletal dysplasias. Human Mutation, 2018, 39, 1456-1467. | 2.5 | 16 |
| 18 | Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3593-3600. | 1.2 | 16 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Cell-free tumour DNA analysis detects copy number alterations in gastro-oesophageal cancer patients. PLoS ONE, 2021, 16, e0245488. | 2.5 | 13 |
| 20 | Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998. | 2.5 | 12 |
| 21 | Hybrid sequencing resolves two germline ultra-complex chromosomal rearrangements consisting of 137 breakpoint junctions in a single carrier. Human Genetics, 2021, 140, 775-790. | 3.8 | 9 |
| 22 | pyCancerSig: subclassifying human cancer with comprehensive single nucleotide, structural and microsatellite mutational signature deconstruction from whole genome sequencing. BMC Bioinformatics, 2020, 21, 128. | 2.6 | 7 |
| 23 | Flanking complex copy number variants in the same family formed through unequal crossing-over during meiosis. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2018, 812, 1-4. | 1.0 | 6 |
| 24 | Whole genome sequencing unveils genetic heterogeneity in optic nerve hypoplasia. PLoS ONE, 2020, 15, e0228622. | 2.5 | 6 |
| 25 | Rare variants in dynein heavy chain genes in two individuals with situs inversus and developmental dyslexia: a case report. BMC Medical Genetics, 2020, 21, 87. | 2.1 | 5 |
| 26 | Loqusdb: added value of an observations database of local genomic variation. BMC Bioinformatics, 2020, 21, 273. | 2.6 | 5 |
| 27 | 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 |
| 28 | Genomic profile â€“ a possible diagnostic and prognostic marker in upper tract urothelial carcinoma. BJU International, 2022, 130, 92-101. | 2.5 | 5 |
| 29 | A somatic UBA2 variant preceded ETV6-RUNX1 in the concordant BCP-ALL of monozygotic twins. Blood Advances, 2022, 6, 2275-2289. | 5.2 | 5 |
| 30 | Single-cell multimodal analysis in a case with reduced penetrance of Progranulin-Frontotemporal Dementia. Acta Neuropathologica Communications, 2021, 9, 132. | 5.2 | 3 |
| 31 | Multiâ€omics analysis reveals multiple mechanisms causing Praderâ€“Willi like syndrome in a family with a X;15 translocation. Human Mutation, 2022, 43, 1567-1575. | 2.5 | 3 |
| 32 | A database on differentially expressed microRNAs during rodent bladder healing. Scientific Reports, 2021, 11, 21881. | 3.3 | 2 |
| 33 | Mosaic Deletions of Known Genes Explain Skeletal Dysplasias With High and Low Bone Mass. JBMR Plus, 2022, 6, . | 2.7 | 2 |
| 34 | Partial Monosomy 21 Mirrors Gene Expression of Trisomy 21 in a Patient-Derived Neuroepithelial Stem Cell Model. Frontiers in Genetics, 2021, 12, 803683. | 2.3 | 1 |