Jesper Eisfeldt

List of Publications by Year
in descending order

34	918	706676 14	591227 27
papers	citations	h-index	g-index

39	39	39	1760
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genomic profile â€“ a possible diagnostic and prognostic marker in upper tract urothelial carcinoma. BJU International, 2022, 130, 92-101.	1.3	5
2	A somatic UBA2 variant preceded ETV6-RUNX1 in the concordant BCP-ALL of monozygotic twins. Blood Advances, 2022, 6, 2275-2289.	2.5	5
3	Mosaic Deletions of Known Genes Explain Skeletal Dysplasias With High and Low Bone Mass. JBMR Plus, 2022, 6, .	1.3	2
4	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.	1.1	3
5	Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3593-3600.	0.7	16
6	Chromatin interactions in differentiating keratinocytes reveal novel atopic dermatitisâ€“ and psoriasis-associated genes. Journal of Allergy and Clinical Immunology, 2021, 147, 1742-1752.	1.5	18
7	Cell-free tumour DNA analysis detects copy number alterations in gastro-oesophageal cancer patients. PLoS ONE, 2021, 16, e0245488.	1.1	13
8	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.	3.6	116
9	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.	1.1	19
10	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.	1.1	5
11	Single-cell multimodal analysis in a case with reduced penetrance of Progranulin-Frontotemporal Dementia. Acta Neuropathologica Communications, 2021, 9, 132.	2.4	3
12	Hybrid sequencing resolves two germline ultra-complex chromosomal rearrangements consisting of 137 breakpoint junctions in a single carrier. Human Genetics, 2021, 140, 775-790.	1.8	9
13	A database on differentially expressed microRNAs during rodent bladder healing. Scientific Reports, 2021, 11, 21881.	1.6	2
14	Partial Monosomy 21 Mirrors Gene Expression of Trisomy 21 in a Patient-Derived Neuroepithelial Stem Cell Model. Frontiers in Genetics, 2021, 12, 803683.	1.1	1
15	Discovery of Novel Sequences in 1,000 Swedish Genomes. Molecular Biology and Evolution, 2020, 37, 18-30.	3.5	25
16	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	1.1	12
17	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
18	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.	0.7	17

#	Article	IF	Citations
19	Loqusdb: added value of an observations database of local genomic variation. BMC Bioinformatics, 2020, 21, 273.	1.2	5
20	Whole genome sequencing unveils genetic heterogeneity in optic nerve hypoplasia. PLoS ONE, 2020, 15, e0228622.	1.1	6
21	pyCancerSig: subclassifying human cancer with comprehensive single nucleotide, structural and microsatellite mutational signature deconstruction from whole genome sequencing. BMC Bioinformatics, 2020, 21, 128.	1.2	7
22	Sarek: A portable workflow for whole-genome sequencing analysis of germline and somatic variants. F1000Research, 2020, 9, 63.	0.8	21
23	Sarek: A portable workflow for whole-genome sequencing analysis of germline and somatic variants. F1000Research, 2020, 9, 63.	0.8	89
24	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.	3.6	88
25	Comprehensive structural variation genome map of individuals carrying complex chromosomal rearrangements. PLoS Genetics, 2019, 15, e1007858.	1.5	36
26	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.	1.1	17
27	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780.	1.5	28
28	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.	0.4	6
29	Alu-Alu mediated intragenic duplications in IFT81 and MATN3 are associated with skeletal dysplasias. Human Mutation, 2018, 39, 1456-1467.	1.1	16
30	AMYCNE: Confident copy number assessment using whole genome sequencing data. PLoS ONE, 2018, 13, e0189710.	1.1	19
31	A Large Inversion Involving GNAS Exon A/B and All Exons Encoding Gsĵ± Is Associated With Autosomal Dominant Pseudohypoparathyroidism Type Ib (PHP1B). Journal of Bone and Mineral Research, 2017, 32, 776-783.	3.1	22
32	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.	1.4	148
33	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664.	0.8	76
34	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664.	0.8	51