Lars Feuk

List of Publications by Year in Descending Order

88 papers	20,492 citations	40 h-index	92 g-index
92 ext. papers	23,096 ext. citations	11.8 avg, IF	5.89 L-index

#	Paper	IF	Citations
88	CRISPR-Cas9 induces large structural variants at on-target and off-target sites in vivo that segregate across generations.. *Nature Communications*, **2022**, 13, 627	17.4	5
87	Hybrid sequencing resolves two germline ultra-complex chromosomal rearrangements consisting of 137 breakpoint junctions in a single carrier. *Human Genetics*, **2021**, 140, 775-790	6.3	2
86	Characterization of the nuclear and cytosolic transcriptomes in human brain tissue reveals new insights into the subcellular distribution of RNA transcripts. *Scientific Reports*, **2021**, 11, 4076	4.9	9
85	R.ROSETTA: an interpretable machine learning framework. *BMC Bioinformatics*, **2021**, 22, 110	3.6	5
84	DLG4-related synaptopathy: a new rare brain disorder. *Genetics in Medicine*, **2021**, 23, 888-899	8.1	0
83	Interpretable Machine Learning Reveals Dissimilarities Between Subtypes of Autism Spectrum Disorder. *Frontiers in Genetics*, **2021**, 12, 618277	4.5	3
82	Whole genome sequencing of familial isolated oesophagus atresia uncover shared structural variants. *BMC Medical Genomics*, **2020**, 13, 85	3.7	1
81	Transcriptome analysis of fibroblasts from schizophrenia patients reveals differential expression of schizophrenia-related genes. *Scientific Reports*, **2020**, 10, 630	4.9	7
80	Exploring autoantibody signatures in brain tissue from patients with severe mental illness. *Translational Psychiatry*, **2020**, 10, 401	8.6	3
79	Amplification-free long-read sequencing reveals unforeseen CRISPR-Cas9 off-target activity. *Genome Biology*, **2020**, 21, 290	18.3	12
78	Identification and rescue of a tRNA wobble inosine deficiency causing intellectual disability disorder. *Rna*, **2020**, 26, 1654-1666	5.8	2
77	Evaluation of Single-Molecule Sequencing Technologies for Structural Variant Detection in Two Swedish Human Genomes. *Genes*, **2020**, 11,	4.2	3
76	Copy number determination of the gene for the human pancreatic polypeptide receptor NPY4R using read depth analysis and droplet digital PCR. *BMC Biotechnology*, **2019**, 19, 31	3.5	1
75	Analyzing DNA methylation patterns in subjects diagnosed with schizophrenia using machine learning methods. *Journal of Psychiatric Research*, **2019**, 114, 41-47	5.2	7
74	Novel Y-Chromosome Long Non-Coding RNAs Expressed in Human Male CNS During Early Development. *Frontiers in Genetics*, **2019**, 10, 891	4.5	6
73	Linkage and exome analysis implicate multiple genes in non-syndromic intellectual disability in a large Swedish family. *BMC Medical Genomics*, **2019**, 12, 156	3.7	3
72	Whole genome sequencing of consanguineous families reveals novel pathogenic variants in intellectual disability. *Clinical Genetics*, **2019**, 95, 436-439	4	9

71	Exome sequencing reveals NAA15 and PUF60 as candidate genes associated with intellectual disability. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2018**, 177, 10-20	3.5	19
70	Detailed analysis of HTT repeat elements in human blood using targeted amplification-free long-read sequencing. *Human Mutation*, **2018**, 39, 1262-1272	4.7	37
69	Expression profiling and in situ screening of circular RNAs in human tissues. *Scientific Reports*, **2018**, 8, 16953	4.9	15
68	De Novo Assembly of Two Swedish Genomes Reveals Missing Segments from the Human GRCh38 Reference and Improves Variant Calling of Population-Scale Sequencing Data. *Genes*, **2018**, 9,	4.2	28
67	Copy number of pancreatic polypeptide receptor gene NPY4R correlates with body mass index and waist circumference. *PLoS ONE*, **2018**, 13, e0194668	3.7	11
66	Reduced cell surface levels of GPI-linked markers in a new case with PIGG loss of function. *Human Mutation*, **2017**, 38, 1394-1401	4.7	17
65	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	5.3	103
64	A 3-way hybrid approach to generate a new high-quality chimpanzee reference genome (Pan_tro_3.0). *GigaScience*, **2017**, 6, 1-6	7.6	16
63	Spatial sexual dimorphism of X and Y homolog gene expression in the human central nervous system during early male development. *Biology of Sex Differences*, **2016**, 7, 5	9.3	19
62	A Role for the Chromatin-Remodeling Factor BAZ1A in Neurodevelopment. *Human Mutation*, **2016**, 37, 964-75	4.7	22
61	One CNV Discordance in NRXN1 Observed Upon Genome-wide Screening in 38 Pairs of Adult Healthy Monozygotic Twins. *Twin Research and Human Genetics*, **2016**, 19, 97-103	2.2	2
60	Mutations in HECW2 are associated with intellectual disability and epilepsy. *Journal of Medical Genetics*, **2016**, 53, 697-704	5.8	39
59	Transcriptome Profiling Reveals Degree of Variability in Induced Pluripotent Stem Cell Lines: Impact for Human Disease Modeling. *Cellular Reprogramming*, **2015**, 17, 327-37	2.1	19
58	The Database of Genomic Variants: a curated collection of structural variation in the human genome. *Nucleic Acids Research*, **2014**, 42, D986-92	20.1	764
57	Splicing in the human brain. *International Review of Neurobiology*, **2014**, 116, 95-125	4.4	14
56	Deleterious mutation in FDX1L gene is associated with a novel mitochondrial muscle myopathy. *European Journal of Human Genetics*, **2014**, 22, 902-6	5.3	56
55	Abolished InsP3R2 function inhibits sweat secretion in both humans and mice. *Journal of Clinical Investigation*, **2014**, 124, 4773-80	15.9	45
54	Welander distal myopathy caused by an ancient founder mutation in TIA1 associated with perturbed splicing. *Human Mutation*, **2013**, 34, 572-7	4.7	76

53	Efficient cellular fractionation improves RNA sequencing analysis of mature and nascent transcripts from human tissues. *BMC Biotechnology*, **2013**, 13, 99	3.5	33
52	Genome-wide association study of susceptibility loci for cervical cancer. *Journal of the National Cancer Institute*, **2013**, 105, 624-33	9.7	113
51	Mechanisms of formation of structural variation in a fully sequenced human genome. *Human Mutation*, **2013**, 34, 345-54	4.7	25
50	Exome RNA sequencing reveals rare and novel alternative transcripts. *Nucleic Acids Research*, **2013**, 41, e6	20.1	38
49	RNA-binding protein QKI regulates Glial fibrillary acidic protein expression in human astrocytes. *Human Molecular Genetics*, **2013**, 22, 1373-82	5.6	18
48	Intractable epilepsy of infancy due to homozygous mutation in the EFHC1 gene. *Epilepsia*, **2012**, 53, 1436-40	6.4	14
47	Diagnostic interpretation of array data using public databases and internet sources. *Human Mutation*, **2012**, 33, 930-40	4.7	74
46	Infantile cerebellar-retinal degeneration associated with a mutation in mitochondrial aconitase, ACO2. *American Journal of Human Genetics*, **2012**, 90, 518-23	11	72
45	Genetic adaptation of fatty-acid metabolism: a human-specific haplotype increasing the biosynthesis of long-chain omega-3 and omega-6 fatty acids. *American Journal of Human Genetics*, **2012**, 90, 809-20	11	148
44	Total RNA sequencing reveals nascent transcription and widespread co-transcriptional splicing in the human brain. *Nature Structural and Molecular Biology*, **2011**, 18, 1435-40	17.6	217
43	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. *Nature Biotechnology*, **2011**, 29, 512-20	44.5	333
42	Characterization of copy number-stable regions in the human genome. *Human Mutation*, **2011**, 32, 947-55	4.7	17
41	Origins and functional impact of copy number variation in the human genome. *Nature*, **2010**, 464, 704-12	50.4	1467
40	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. *Nature*, **2010**, 464, 713-20	50.4	639
39	Public data archives for genomic structural variation. *Nature Genetics*, **2010**, 42, 813-4	36.3	67
38	Identification of novel exons and transcribed regions by chimpanzee transcriptome sequencing. *Genome Biology*, **2010**, 11, R78	18.3	26
37	Global and unbiased detection of splice junctions from RNA-seq data. *Genome Biology*, **2010**, 11, R34	18.3	65
36	Inversion variants in the human genome: role in disease and genome architecture. *Genome Medicine* , **2010**, 2, 11	14.4	47

35	Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. *American Journal of Human Genetics*, **2010**, 86, 749-64	11	1831
34	Prepublication data sharing. *Nature*, **2009**, 461, 168-70	50.4	197
33	Multiple recurrent genetic events converge on control of histone lysine methylation in medulloblastoma. *Nature Genetics*, **2009**, 41, 465-72	36.3	337
32	ASHG 2008 Annual Meeting: from enormous cohorts to individual genomes. *Genome Medicine*, **2009** , 1, 9	14.4	3
31	Excessive genomic DNA copy number variation in the Li-Fraumeni cancer predisposition syndrome. *Proceedings of the National Academy of Sciences of the United States of America*, **2008**, 105, 11264-9	11.5	155
30	Copy number variation in the autism genome. *Expert Opinion on Medical Diagnostics*, **2008**, 2, 417-28		1
29	Structural variation of chromosomes in autism spectrum disorder. *American Journal of Human Genetics*, **2008**, 82, 477-88	11	1413
28	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. *Nature Genetics*, **2007**, 39, 319-28	36.3	1083
27	Challenges and standards in integrating surveys of structural variation. *Nature Genetics*, **2007**, 39, S7-15	36.3	279
26	The diploid genome sequence of an individual human. *PLoS Biology*, **2007**, 5, e254	9.7	1249
25	Accurate and reliable high-throughput detection of copy number variation in the human genome. *Genome Research*, **2006**, 16, 1566-74	9.7	122
24	Structural variants: changing the landscape of chromosomes and design of disease studies. *Human Molecular Genetics*, **2006**, 15 Spec No 1, R57-66	5.6	190
23	Copy number variation: new insights in genome diversity. *Genome Research*, **2006**, 16, 949-61	9.7	580
22	Absence of a paternally inherited FOXP2 gene in developmental verbal dyspraxia. *American Journal of Human Genetics*, **2006**, 79, 965-72	11	143
21	Frequent appearance of novel protein-coding sequences by frameshift translation. *Genomics*, **2006**, 88, 690-697	4.3	36
20	Genome assembly comparison identifies structural variants in the human genome. *Nature Genetics*, **2006**, 38, 1413-8	36.3	133
19	Structural variation in the human genome. *Nature Reviews Genetics*, **2006**, 7, 85-97	30.1	1416
18	Global variation in copy number in the human genome. *Nature*, **2006**, 444, 444-54	50.4	3306

17	Longitudinal memory performance during normal aging: twin association models of APOE and other Alzheimer candidate genes. *Behavior Genetics*, **2006**, 36, 185-94	3.2	34
16	Towards compendia of negative genetic association studies: an example for Alzheimer disease. *Human Genetics*, **2006**, 119, 29-37	6.3	38
15	Sequence variants of IDE are associated with the extent of beta-amyloid deposition in the Alzheimer disease brain. *Neurobiology of Aging*, **2005**, 26, 795-802	5.6	42
14	Linkage disequilibrium patterns vary substantially among populations. *European Journal of Human Genetics*, **2005**, 13, 677-86	5.3	113
13	Discovery of human inversion polymorphisms by comparative analysis of human and chimpanzee DNA sequence assemblies. *PLoS Genetics*, **2005**, 1, e56	6	130
12	Elevated amyloid beta protein (Abeta42) and late onset Alzheimer disease are associated with single nucleotide polymorphisms in the urokinase-type plasminogen activator gene. *Human Molecular Genetics*, **2005**, 14, 447-60	5.6	58
11	Detection of large-scale variation in the human genome. *Nature Genetics*, **2004**, 36, 949-51	36.3	2371
10	Variants of CYP46A1 may interact with age and APOE to influence CSF Abeta42 levels in Alzheimer disease. *Human Genetics*, **2004**, 114, 581-7	6.3	57
9	Further evidence for role of a promoter variant in the TNFRSF6 gene in Alzheimer disease. *Human Mutation*, **2003**, 21, 53-60	4.7	20
8	Genetic variation in a haplotype block spanning IDE influences Alzheimer disease. *Human Mutation*, **2003**, 22, 363-71	4.7	82
7	Haplotypes extending across ACE are associated with Alzheimer disease. *Human Molecular Genetics*, **2003**, 12, 859-67	5.6	93
6	Lack of replication of association findings in complex disease: an analysis of 15 polymorphisms in prior candidate genes for sporadic Alzheimer disease. *European Journal of Human Genetics*, **2001**, 9, 437-44	5.3	132
5	The TNFRSF6 gene is not implicated in familial early-onset Alzheimer disease. *Human Genetics*, **2001**, 108, 552-3	6.3	6
4	apolipoprotein-E dependent role for the FAS receptor in early onset Alzheimer disease: finding of a positive association for a polymorphism in the TNFRSF6 gene. *Human Genetics*, **2000**, 107, 391-6	6.3	36
3	Amplification-free long read sequencing reveals unforeseen CRISPR-Cas9 off-target activity		5
2	De novo assembly of two Swedish genomes reveals missing segments from the human GRCh38 reference and improves variant calling of population-scale sequencing data		5
1	Long-read whole genome analysis of human single cells		3