

Lars Feuk

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.

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	Global variation in copy number in the human genome. <i>Nature</i> , 2006 , 444, 444-54	50.4	3306
87	Detection of large-scale variation in the human genome. <i>Nature Genetics</i> , 2004 , 36, 949-51	36.3	2371
86	Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. <i>American Journal of Human Genetics</i> , 2010 , 86, 749-64	11	1831
85	Origins and functional impact of copy number variation in the human genome. <i>Nature</i> , 2010 , 464, 704-12	50.4	1467
84	Structural variation in the human genome. <i>Nature Reviews Genetics</i> , 2006 , 7, 85-97	30.1	1416
83	Structural variation of chromosomes in autism spectrum disorder. <i>American Journal of Human Genetics</i> , 2008 , 82, 477-88	11	1413
82	The diploid genome sequence of an individual human. <i>PLoS Biology</i> , 2007 , 5, e254	9.7	1249
81	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. <i>Nature Genetics</i> , 2007 , 39, 319-28	36.3	1083
80	The Database of Genomic Variants: a curated collection of structural variation in the human genome. <i>Nucleic Acids Research</i> , 2014 , 42, D986-92	20.1	764
79	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. <i>Nature</i> , 2010 , 464, 713-20	50.4	639
78	Copy number variation: new insights in genome diversity. <i>Genome Research</i> , 2006 , 16, 949-61	9.7	580
77	Multiple recurrent genetic events converge on control of histone lysine methylation in medulloblastoma. <i>Nature Genetics</i> , 2009 , 41, 465-72	36.3	337
76	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. <i>Nature Biotechnology</i> , 2011 , 29, 512-20	44.5	333
75	Challenges and standards in integrating surveys of structural variation. <i>Nature Genetics</i> , 2007 , 39, S7-15	36.3	279
74	Total RNA sequencing reveals nascent transcription and widespread co-transcriptional splicing in the human brain. <i>Nature Structural and Molecular Biology</i> , 2011 , 18, 1435-40	17.6	217
73	Prepublication data sharing. <i>Nature</i> , 2009 , 461, 168-70	50.4	197
72	Structural variants: changing the landscape of chromosomes and design of disease studies. <i>Human Molecular Genetics</i> , 2006 , 15 Spec No 1, R57-66	5.6	190

71	Excessive genomic DNA copy number variation in the Li-Fraumeni cancer predisposition syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008 , 105, 11264-9	11.5	155
70	Genetic adaptation of fatty-acid metabolism: a human-specific haplotype increasing the biosynthesis of long-chain omega-3 and omega-6 fatty acids. <i>American Journal of Human Genetics</i> , 2012 , 90, 809-20	11	148
69	Absence of a paternally inherited FOXP2 gene in developmental verbal dyspraxia. <i>American Journal of Human Genetics</i> , 2006 , 79, 965-72	11	143
68	Genome assembly comparison identifies structural variants in the human genome. <i>Nature Genetics</i> , 2006 , 38, 1413-8	36.3	133
67	Lack of replication of association findings in complex disease: an analysis of 15 polymorphisms in prior candidate genes for sporadic Alzheimer's disease. <i>European Journal of Human Genetics</i> , 2001 , 9, 437-44	5.3	132
66	Discovery of human inversion polymorphisms by comparative analysis of human and chimpanzee DNA sequence assemblies. <i>PLoS Genetics</i> , 2005 , 1, e56	6	130
65	Accurate and reliable high-throughput detection of copy number variation in the human genome. <i>Genome Research</i> , 2006 , 16, 1566-74	9.7	122
64	Genome-wide association study of susceptibility loci for cervical cancer. <i>Journal of the National Cancer Institute</i> , 2013 , 105, 624-33	9.7	113
63	Linkage disequilibrium patterns vary substantially among populations. <i>European Journal of Human Genetics</i> , 2005 , 13, 677-86	5.3	113
62	SweGen: a whole-genome data resource of genetic variability in a cross-section of the Swedish population. <i>European Journal of Human Genetics</i> , 2017 , 25, 1253-1260	5.3	103
61	Haplotypes extending across ACE are associated with Alzheimer's disease. <i>Human Molecular Genetics</i> , 2003 , 12, 859-67	5.6	93
60	Genetic variation in a haplotype block spanning IDE influences Alzheimer disease. <i>Human Mutation</i> , 2003 , 22, 363-71	4.7	82
59	Welander distal myopathy caused by an ancient founder mutation in TIA1 associated with perturbed splicing. <i>Human Mutation</i> , 2013 , 34, 572-7	4.7	76
58	Diagnostic interpretation of array data using public databases and internet sources. <i>Human Mutation</i> , 2012 , 33, 930-40	4.7	74
57	Infantile cerebellar-retinal degeneration associated with a mutation in mitochondrial aconitase, ACO2. <i>American Journal of Human Genetics</i> , 2012 , 90, 518-23	11	72
56	Public data archives for genomic structural variation. <i>Nature Genetics</i> , 2010 , 42, 813-4	36.3	67
55	Global and unbiased detection of splice junctions from RNA-seq data. <i>Genome Biology</i> , 2010 , 11, R34	18.3	65
54	Elevated amyloid beta protein (Abeta42) and late onset Alzheimer's disease are associated with single nucleotide polymorphisms in the urokinase-type plasminogen activator gene. <i>Human Molecular Genetics</i> , 2005 , 14, 447-60	5.6	58

53	Variants of CYP46A1 may interact with age and APOE to influence CSF Abeta42 levels in Alzheimer's disease. <i>Human Genetics</i> , 2004 , 114, 581-7	6.3	57
52	Deleterious mutation in FDX1L gene is associated with a novel mitochondrial muscle myopathy. <i>European Journal of Human Genetics</i> , 2014 , 22, 902-6	5.3	56
51	Inversion variants in the human genome: role in disease and genome architecture. <i>Genome Medicine</i> , 2010 , 2, 11	14.4	47
50	Abolished InsP3R2 function inhibits sweat secretion in both humans and mice. <i>Journal of Clinical Investigation</i> , 2014 , 124, 4773-80	15.9	45
49	Sequence variants of IDE are associated with the extent of beta-amyloid deposition in the Alzheimer's disease brain. <i>Neurobiology of Aging</i> , 2005 , 26, 795-802	5.6	42
48	Mutations in HECW2 are associated with intellectual disability and epilepsy. <i>Journal of Medical Genetics</i> , 2016 , 53, 697-704	5.8	39
47	Exome RNA sequencing reveals rare and novel alternative transcripts. <i>Nucleic Acids Research</i> , 2013 , 41, e6	20.1	38
46	Towards compendia of negative genetic association studies: an example for Alzheimer disease. <i>Human Genetics</i> , 2006 , 119, 29-37	6.3	38
45	Detailed analysis of HTT repeat elements in human blood using targeted amplification-free long-read sequencing. <i>Human Mutation</i> , 2018 , 39, 1262-1272	4.7	37
44	Frequent appearance of novel protein-coding sequences by frameshift translation. <i>Genomics</i> , 2006 , 88, 690-697	4.3	36
43	apolipoprotein-E dependent role for the FAS receptor in early onset Alzheimer's disease: finding of a positive association for a polymorphism in the TNFRSF6 gene. <i>Human Genetics</i> , 2000 , 107, 391-6	6.3	36
42	Longitudinal memory performance during normal aging: twin association models of APOE and other Alzheimer candidate genes. <i>Behavior Genetics</i> , 2006 , 36, 185-94	3.2	34
41	Efficient cellular fractionation improves RNA sequencing analysis of mature and nascent transcripts from human tissues. <i>BMC Biotechnology</i> , 2013 , 13, 99	3.5	33
40	De Novo Assembly of Two Swedish Genomes Reveals Missing Segments from the Human GRCh38 Reference and Improves Variant Calling of Population-Scale Sequencing Data. <i>Genes</i> , 2018 , 9,	4.2	28
39	Identification of novel exons and transcribed regions by chimpanzee transcriptome sequencing. <i>Genome Biology</i> , 2010 , 11, R78	18.3	26
38	Mechanisms of formation of structural variation in a fully sequenced human genome. <i>Human Mutation</i> , 2013 , 34, 345-54	4.7	25
37	A Role for the Chromatin-Remodeling Factor BAZ1A in Neurodevelopment. <i>Human Mutation</i> , 2016 , 37, 964-75	4.7	22
36	Further evidence for role of a promoter variant in the TNFRSF6 gene in Alzheimer disease. <i>Human Mutation</i> , 2003 , 21, 53-60	4.7	20

35	Transcriptome Profiling Reveals Degree of Variability in Induced Pluripotent Stem Cell Lines: Impact for Human Disease Modeling. <i>Cellular Reprogramming</i> , 2015 , 17, 327-37	2.1	19
34	Exome sequencing reveals NAA15 and PUF60 as candidate genes associated with intellectual disability. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018 , 177, 10-20	3.5	19
33	Spatial sexual dimorphism of X and Y homolog gene expression in the human central nervous system during early male development. <i>Biology of Sex Differences</i> , 2016 , 7, 5	9.3	19
32	RNA-binding protein QKI regulates Glial fibrillary acidic protein expression in human astrocytes. <i>Human Molecular Genetics</i> , 2013 , 22, 1373-82	5.6	18
31	Reduced cell surface levels of GPI-linked markers in a new case with PIGG loss of function. <i>Human Mutation</i> , 2017 , 38, 1394-1401	4.7	17
30	Characterization of copy number-stable regions in the human genome. <i>Human Mutation</i> , 2011 , 32, 947-54	5.7	17
29	A 3-way hybrid approach to generate a new high-quality chimpanzee reference genome (Pan_tro_3.0). <i>GigaScience</i> , 2017 , 6, 1-6	7.6	16
28	Expression profiling and in situ screening of circular RNAs in human tissues. <i>Scientific Reports</i> , 2018 , 8, 16953	4.9	15
27	Splicing in the human brain. <i>International Review of Neurobiology</i> , 2014 , 116, 95-125	4.4	14
26	Intractable epilepsy of infancy due to homozygous mutation in the EFHC1 gene. <i>Epilepsia</i> , 2012 , 53, 1436-40	6.4	14
25	Amplification-free long-read sequencing reveals unforeseen CRISPR-Cas9 off-target activity. <i>Genome Biology</i> , 2020 , 21, 290	18.3	12
24	Copy number of pancreatic polypeptide receptor gene NPY4R correlates with body mass index and waist circumference. <i>PLoS ONE</i> , 2018 , 13, e0194668	3.7	11
23	Characterization of the nuclear and cytosolic transcriptomes in human brain tissue reveals new insights into the subcellular distribution of RNA transcripts. <i>Scientific Reports</i> , 2021 , 11, 4076	4.9	9
22	Whole genome sequencing of consanguineous families reveals novel pathogenic variants in intellectual disability. <i>Clinical Genetics</i> , 2019 , 95, 436-439	4	9
21	Analyzing DNA methylation patterns in subjects diagnosed with schizophrenia using machine learning methods. <i>Journal of Psychiatric Research</i> , 2019 , 114, 41-47	5.2	7
20	Transcriptome analysis of fibroblasts from schizophrenia patients reveals differential expression of schizophrenia-related genes. <i>Scientific Reports</i> , 2020 , 10, 630	4.9	7
19	Novel Y-Chromosome Long Non-Coding RNAs Expressed in Human Male CNS During Early Development. <i>Frontiers in Genetics</i> , 2019 , 10, 891	4.5	6
18	The TNFRSF6 gene is not implicated in familial early-onset Alzheimer's disease. <i>Human Genetics</i> , 2001 , 108, 552-3	6.3	6

17	CRISPR-Cas9 induces large structural variants at on-target and off-target sites in vivo that segregate across generations.. <i>Nature Communications</i> , 2022 , 13, 627	17.4	5
16	Amplification-free long read sequencing reveals unforeseen CRISPR-Cas9 off-target activity		5
15	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
14	R.ROSETTA: an interpretable machine learning framework. <i>BMC Bioinformatics</i> , 2021 , 22, 110	3.6	5
13	Linkage and exome analysis implicate multiple genes in non-syndromic intellectual disability in a large Swedish family. <i>BMC Medical Genomics</i> , 2019 , 12, 156	3.7	3
12	ASHG 2008 Annual Meeting: from enormous cohorts to individual genomes. <i>Genome Medicine</i> , 2009 , 1, 9	14.4	3
11	Exploring autoantibody signatures in brain tissue from patients with severe mental illness. <i>Translational Psychiatry</i> , 2020 , 10, 401	8.6	3
10	Evaluation of Single-Molecule Sequencing Technologies for Structural Variant Detection in Two Swedish Human Genomes. <i>Genes</i> , 2020 , 11,	4.2	3
9	Long-read whole genome analysis of human single cells		3
8	Interpretable Machine Learning Reveals Dissimilarities Between Subtypes of Autism Spectrum Disorder. <i>Frontiers in Genetics</i> , 2021 , 12, 618277	4.5	3
7	Hybrid sequencing resolves two germline ultra-complex chromosomal rearrangements consisting of 137 breakpoint junctions in a single carrier. <i>Human Genetics</i> , 2021 , 140, 775-790	6.3	2
6	Identification and rescue of a tRNA wobble inosine deficiency causing intellectual disability disorder. <i>Rna</i> , 2020 , 26, 1654-1666	5.8	2
5	One CNV Discordance in NRXN1 Observed Upon Genome-wide Screening in 38 Pairs of Adult Healthy Monozygotic Twins. <i>Twin Research and Human Genetics</i> , 2016 , 19, 97-103	2.2	2
4	Copy number determination of the gene for the human pancreatic polypeptide receptor NPY4R using read depth analysis and droplet digital PCR. <i>BMC Biotechnology</i> , 2019 , 19, 31	3.5	1
3	Whole genome sequencing of familial isolated oesophagus atresia uncover shared structural variants. <i>BMC Medical Genomics</i> , 2020 , 13, 85	3.7	1
2	Copy number variation in the autism genome. <i>Expert Opinion on Medical Diagnostics</i> , 2008 , 2, 417-28		1
1	DLG4-related synaptopathy: a new rare brain disorder. <i>Genetics in Medicine</i> , 2021 , 23, 888-899	8.1	0