Lars Feuk

List of Publications by Year
in descending order

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86 papers	24,620 citations	61977 43 h-index	51602 86 g-index
92 all docs	92 docs citations	92 times ranked	28083 citing authors

#	Article	IF	Citations
1	CRISPR-Cas9 induces large structural variants at on-target and off-target sites in vivo that segregate across generations. Nature Communications, 2022, 13, 627.	12.8	65
2	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
3	Interpretable Machine Learning Reveals Dissimilarities Between Subtypes of Autism Spectrum Disorder. Frontiers in Genetics, 2021, 12, 618277.	2.3	7
4	R.ROSETTA: an interpretable machine learning framework. BMC Bioinformatics, 2021, 22, 110.	2.6	16
5	Transcriptome Analysis of Post-Mortem Brain Tissue Reveals Up-Regulation of the Complement Cascade in a Subgroup of Schizophrenia Patients. Genes, 2021, 12, 1242.	2.4	12
6	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
7	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.	3.3	27
8	Exploring autoantibody signatures in brain tissue from patients with severe mental illness. Translational Psychiatry, 2020, 10, 401.	4.8	8
9	Amplification-free long-read sequencing reveals unforeseen CRISPR-Cas9 off-target activity. Genome Biology, 2020, 21, 290.	8.8	35
10	Identification and rescue of a tRNA wobble inosine deficiency causing intellectual disability disorder. Rna, 2020, 26, 1654-1666.	3.5	16
11	Evaluation of Single-Molecule Sequencing Technologies for Structural Variant Detection in Two Swedish Human Genomes. Genes, 2020, 11, 1444.	2.4	6
12	Whole genome sequencing of familial isolated oesophagus atresia uncover shared structural variants. BMC Medical Genomics, 2020, 13, 85.	1.5	2
13	Transcriptome analysis of fibroblasts from schizophrenia patients reveals differential expression of schizophrenia-related genes. Scientific Reports, 2020, 10, 630.	3.3	22
14	Novel Y-Chromosome Long Non-Coding RNAs Expressed in Human Male CNS During Early Development. Frontiers in Genetics, 2019, 10, 891.	2.3	8
15	Linkage and exome analysis implicate multiple genes in non-syndromic intellectual disability in a large Swedish family. BMC Medical Genomics, 2019, 12, 156.	1.5	3
16	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.	3.3	4
17	Analyzing DNA methylation patterns in subjects diagnosed with schizophrenia using machine learning methods. Journal of Psychiatric Research, 2019, 114, 41-47.	3.1	19
18	Whole genome sequencing of consanguineous families reveals novel pathogenic variants in intellectual disability. Clinical Genetics, 2019, 95, 436-439.	2.0	15

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19	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.	1.7	32
20	Expression profiling and in situ screening of circular RNAs in human tissues. Scientific Reports, 2018, 8, 16953.	3.3	21
21	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, 486.	2.4	50
22	Copy number of pancreatic polypeptide receptor gene NPY4R correlates with body mass index and waist circumference. PLoS ONE, 2018, 13, e0194668.	2.5	20
23	Detailed analysis of HTT repeat elements in human blood using targeted amplification-free long-read sequencing. Human Mutation, 2018, 39, 1262-1272.	2.5	62
24	Reduced cell surface levels of GPI-linked markers in a new case with PIGG loss of function. Human Mutation, 2017, 38, 1394-1401.	2.5	20
25	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
26	A 3-way hybrid approach to generate a new high-quality chimpanzee reference genome (Pan_tro_3.0). GigaScience, 2017, 6, 1-6.	6.4	17
27	A Role for the Chromatinâ€Remodeling FactorBAZ1Ain Neurodevelopment. Human Mutation, 2016, 37, 964-975.	2.5	29
28	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.	0.6	2
29	Mutations inHECW2are associated with intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 697-704.	3.2	55
30	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.	4.1	25
31	Transcriptome Profiling Reveals Degree of Variability in Induced Pluripotent Stem Cell Lines: Impact for Human Disease Modeling. Cellular Reprogramming, 2015, 17, 327-337.	0.9	21
32	Deleterious mutation in FDX1L gene is associated with a novel mitochondrial muscle myopathy. European Journal of Human Genetics, 2014, 22, 902-906.	2.8	65
33	The Database of Genomic Variants: a curated collection of structural variation in the human genome. Nucleic Acids Research, 2014, 42, D986-D992.	14.5	1,033
34	Splicing in the Human Brain. International Review of Neurobiology, 2014, 116, 95-125.	2.0	20
35	Abolished InsP3R2 function inhibits sweat secretion in both humans and mice. Journal of Clinical Investigation, 2014, 124, 4773-4780.	8.2	63
36	Welander Distal Myopathy Caused by an Ancient Founder Mutation inTIA1Associated with Perturbed Splicing. Human Mutation, 2013, 34, n/a-n/a.	2.5	91

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37	Efficient cellular fractionation improves RNA sequencing analysis of mature and nascent transcripts from human tissues. BMC Biotechnology, 2013, 13, 99.	3.3	47
38	Genome-wide Association Study of Susceptibility Loci for Cervical Cancer. Journal of the National Cancer Institute, 2013, 105, 624-633.	6.3	151
39	Mechanisms of Formation of Structural Variation in a Fully Sequenced Human Genome. Human Mutation, 2013, 34, 345-354.	2.5	34
40	Exome RNA sequencing reveals rare and novel alternative transcripts. Nucleic Acids Research, 2013, 41, e6-e6.	14.5	43
41	RNA-binding protein QKI regulates Glial fibrillary acidic protein expression in human astrocytes. Human Molecular Genetics, 2013, 22, 1373-1382.	2.9	21
42	Intractable epilepsy of infancy due to homozygous mutation in the EFHC1 gene. Epilepsia, 2012, 53, 1436-1440.	5.1	16
43	Diagnostic interpretation of array data using public databases and internet sources. Human Mutation, 2012, 33, 930-940.	2.5	87
44	Infantile Cerebellar-Retinal Degeneration Associated with a Mutation in Mitochondrial Aconitase, ACO2. American Journal of Human Genetics, 2012, 90, 518-523.	6.2	93
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, 809-820.	6.2	205
46	Total RNA sequencing reveals nascent transcription and widespread co-transcriptional splicing in the human brain. Nature Structural and Molecular Biology, 2011, 18, 1435-1440.	8.2	294
47	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. Nature Biotechnology, 2011, 29, 512-520.	17.5	384
48	Characterization of copy numberâ€stable regions in the human genome. Human Mutation, 2011, 32, 947-955.	2.5	19
49	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, 749-764.	6.2	2,325
50	Origins and functional impact of copy number variation in the human genome. Nature, 2010, 464, 704-712.	27.8	1,721
51	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. Nature, 2010, 464, 713-720.	27.8	737
52	Public data archives for genomic structural variation. Nature Genetics, 2010, 42, 813-814.	21.4	71
53	Identification of novel exons and transcribed regions by chimpanzee transcriptome sequencing. Genome Biology, 2010, 11, R78.	9.6	26
54	Global and unbiased detection of splice junctions from RNA-seq data. Genome Biology, 2010, 11, R34.	9.6	75

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55	Inversion variants in the human genome: role in disease and genome architecture. Genome Medicine, 2010, 2, 11.	8.2	60
56	Prepublication data sharing. Nature, 2009, 461, 168-170.	27.8	243
57	Multiple recurrent genetic events converge on control of histone lysine methylation in medulloblastoma. Nature Genetics, 2009, 41, 465-472.	21.4	391
58	ASHG 2008 Annual Meeting: from enormous cohorts to individual genomes. Genome Medicine, 2009, 1, 9.	8.2	3
59	Structural Variation of Chromosomes in Autism Spectrum Disorder. American Journal of Human Genetics, 2008, 82, 477-488.	6.2	1,641
60	Excessive genomic DNA copy number variation in the Liâ€“Fraumeni cancer predisposition syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 11264-11269.	7.1	192
61	Copy number variation in the autism genome. Expert Opinion on Medical Diagnostics, 2008, 2, 417-428.	1.6	2
62	The Diploid Genome Sequence of an Individual Human. PLoS Biology, 2007, 5, e254.	5.6	1,491
63	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	21.4	1,272
64	Challenges and standards in integrating surveys of structural variation. Nature Genetics, 2007, 39, S7-S15.	21.4	331
65	Absence of a Paternally Inherited FOXP2 Gene in Developmental Verbal Dyspraxia. American Journal of Human Genetics, 2006, 79, 965-972.	6.2	170
66	Frequent appearance of novel protein-coding sequences by frameshift translation. Genomics, 2006, 88, 690-697.	2.9	47
67	Genome assembly comparison identifies structural variants in the human genome. Nature Genetics, 2006, 38, 1413-1418.	21.4	150
68	Structural variation in the human genome. Nature Reviews Genetics, 2006, 7, 85-97.	16.3	1,745
69	Global variation in copy number in the human genome. Nature, 2006, 444, 444-454.	27.8	3,831
70	Longitudinal Memory Performance During Normal Aging: Twin Association Models of APOE and Other Alzheimer Candidate Genes. Behavior Genetics, 2006, 36, 185-194.	2.1	36
71	Towards compendia of negative genetic association studies: an example for Alzheimer disease. Human Genetics, 2006, 119, 29-37.	3.8	41
72	Accurate and reliable high-throughput detection of copy number variation in the human genome. Genome Research, 2006, 16, 1566-1574.	5.5	136

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73	Structural variants: changing the landscape of chromosomes and design of disease studies. Human Molecular Genetics, 2006, 15, R57-R66.	2.9	235
74	Copy number variation: New insights in genome diversity. Genome Research, 2006, 16, 949-961.	5.5	697
75	Linkage disequilibrium patterns vary substantially among populations. European Journal of Human Genetics, 2005, 13, 677-686.	2.8	138
76	Discovery of Human Inversion Polymorphisms by Comparative Analysis of Human and Chimpanzee DNA Sequence Assemblies. PLoS Genetics, 2005, 1, e56.	3.5	149
77	Elevated amyloid Î² protein (AÎ²42) and late onset Alzheimer's disease are associated with single nucleotide polymorphisms in the urokinase-type plasminogen activator gene. Human Molecular Genetics, 2005, 14, 447-460.	2.9	64
78	Sequence variants of IDE are associated with the extent of Î²-amyloid deposition in the Alzheimer's disease brain. Neurobiology of Aging, 2005, 26, 795-802.	3.1	47
79	Detection of large-scale variation in the human genome. Nature Genetics, 2004, 36, 949-951.	21.4	2,602
80	Variants of CYP46A1 may interact with age and APOE to influence CSF Aï¿½42 levels in Alzheimer?s disease. Human Genetics, 2004, 114, 581-587.	3.8	60
81	Further evidence for role of a promoter variant in the TNFRSF6 gene in Alzheimer disease. Human Mutation, 2003, 21, 53-60.	2.5	22
82	Genetic variation in a haplotype block spanningIDE influences Alzheimer disease. Human Mutation, 2003, 22, 363-371.	2.5	94
83	Haplotypes extending across ACE are associated with Alzheimer's disease. Human Molecular Genetics, 2003, 12, 859-867.	2.9	108
84	TheTNFRSF6 gene is not implicated in familial early-onset Alzheimer's disease. Human Genetics, 2001, 108, 552-553.	3.8	6
85	Lack of replication of association findings in complex disease: an analysis of 15 polymorphisms in prior candidate genes for sporadic Alzheimer's disease. European Journal of Human Genetics, 2001, 9, 437-444.	2.8	142
86	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. Human Genetics, 2000, 107, 391-396.	3.8	41