Jukka Moilanen

List of Publications by Year
in descending order

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72	4,304	186265	114465
papers	citations	28 h-index	63 g-index

76	76	76	9391
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A novel variant in SMG9 causes intellectual disability, confirming a role for nonsense-mediated decay components in neurocognitive development. European Journal of Human Genetics, 2022, 30, 619-627.	2.8	6
2	Clinical and Genetic Characteristics of Finnish Patients with Autosomal Recessive and Dominant Non-Syndromic Hearing Loss Due to Pathogenic TMC1 Variants. Journal of Clinical Medicine, 2022, 11, 1837.	2.4	2
3	Truncating TINF2 p.Tyr312Ter variant and inherited breast cancer susceptibility. Familial Cancer, 2022, , .	1.9	1
4	The ethical implications of genetic testing in neurodegenerative diseases: A systematic review. Scandinavian Journal of Caring Sciences, 2021, 35, 1057-1074.	2.1	2
5	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	1.2	17
6	Exome sequencing identifies a recurrent variant in SERPINA3 associating with hereditary susceptibility to breast cancer. European Journal of Cancer, 2021, 143, 46-51.	2.8	11
7	Epidemiological, clinical, and genetic characteristics of paediatric genetic white matter disorders in Northern Finland. Developmental Medicine and Child Neurology, 2021, 63, 1066-1074.	2.1	6
8	Evaluating the role of MLH3 p.Ser1188Ter variant in inherited breast cancer predisposition. Genetics in Medicine, 2020, 22, 663-664.	2.4	2
9	Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families. Journal of Clinical Oncology, 2020, 38, 674-685.	1.6	270
10	Recessive MYH3 variants cause â€œContractures, pterygia, and variable skeletal fusions syndrome 1B â€• mimicking Escobar variant multiple pterygium syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2605-2610.	1.2	5
11	X-linked myotubular myopathy mimics hereditary spastic paraplegia in two female manifesting carriers of pathogenic MTM1 variant. European Journal of Medical Genetics, 2020, 63, 104040.	1.3	1
12	Analysis of functional variants in mitochondrial DNA of Finnish athletes. BMC Genomics, 2019, 20, 784.	2.8	7
13	Contribution of rare and common variants to intellectual disability in a sub-isolate of Northern Finland. Nature Communications, 2019, 10, 410.	12.8	32
14	Exome sequencing in routine diagnostics: a generic test for 254 patients with primary immunodeficiencies. Genome Medicine, 2019, 11, 38.	8.2	49
15	Biallelic loss-of-function P4HTM gene variants cause hypotonia, hypoventilation, intellectual disability, dysautonomia, epilepsy, and eye abnormalities (HIDEA syndrome). Genetics in Medicine, 2019, 21, 2355-2363.	2.4	19
16	Predisposition to childhood acute lymphoblastic leukemia caused by a constitutional translocation disrupting ETV6. Blood Advances, 2019, 3, 2722-2731.	5.2	10
17	Clinical and genetic characteristics of late-onset Huntington's disease. Parkinsonism and Related Disorders, 2019, 61, 101-105.	2.2	17
18	Novel variants in Nordic patients referred for genetic testing of telomere-related disorders. European Journal of Human Genetics, 2018, 26, 858-867.	2.8	14

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19	NHLRC2 variants identified in patients with fibrosis, neurodegeneration, and cerebral angiomatosis (FINCA): characterisation of a novel cerebropulmonary disease. Acta Neuropathologica, 2018, 135, 727-742.	7.7	21
20	Haplotype analysis suggest that the MLH1 c.2059Câ€‰>â€‰T mutation is a Swedish founder mutation. Familial Cancer, 2018, 17, 531-537.	1.9	3
21	A homozygous <scp>I684T</scp> in <scp>GLE1</scp> as a novel cause of arthrogryposis and motor neuron loss. Clinical Genetics, 2018, 93, 173-177.	2.0	3
22	Bone marrow failure syndrome caused by homozygous frameshift mutation in the <scp>ERCC6L2</scp> gene. Clinical Genetics, 2018, 93, 392-395.	2.0	19
23	Neonatal Alexander Disease: Novel GFAP Mutation and Comparison to Previously Published Cases. Neuropediatrics, 2018, 49, 256-261.	0.6	6
24	Quantifying the Impact of Rare and Ultra-rare Coding Variation across the Phenotypic Spectrum. American Journal of Human Genetics, 2018, 102, 1204-1211.	6.2	102
25	Reduced Cancer Incidence in Huntington's Disease: Analysis in the Registry Study. Journal of Huntington's Disease, 2018, 7, 209-222.	1.9	14
26	Identification of C12orf4 as a gene for autosomal recessive intellectual disability. Clinical Genetics, 2017, 91, 100-105.	2.0	15
27	Gain-of-function SAMD9L mutations cause a syndrome of cytopenia, immunodeficiency, MDS, and neurological symptoms. Blood, 2017, 129, 2266-2279.	1.4	152
28	Novel homozygous PCK1 mutation causing cytosolic phosphoenolpyruvate carboxykinase deficiency presenting as childhood hypoglycemia, an abnormal pattern of urine metabolites and liver dysfunction. Molecular Genetics and Metabolism, 2017, 120, 337-341.	1.1	29
29	The genetic basis of classic nonketotic hyperglycinemia due to mutations in GLDC and AMT. Genetics in Medicine, 2017, 19, 104-111.	2.4	71
30	A novel MTTT mutation m.15933Gâ€‰>â€‰A revealed in analysis of mitochondrial DNA in patients with suspected mitochondrial disease. BMC Medical Genetics, 2017, 18, 14.	2.1	2
31	Novel compound heterozygous mutation in <scp>SACS</scp> gene leads to a milder autosomal recessive spastic ataxia of Charlevoixâ€Saguenay, <scp>ARSACS</scp>, in a Finnish family. Clinical Case Reports (discontinued), 2016, 4, 1151-1156.	0.5	12
32	Genetic aetiology of ophthalmological manifestations in children â€“ a focus on mitochondrial diseaseâ€related symptoms. Acta Ophthalmologica, 2016, 94, 83-91.	1.1	10
33	Phenotype and genotype in 52 patients with Rubinsteinâ€“Taybi syndrome caused by EP300 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3069-3082.	1.2	91
34	Rare loss-of-function variants in SETD1A are associated with schizophrenia and developmental disorders. Nature Neuroscience, 2016, 19, 571-577.	14.8	388
35	The Quality of Genetic Counseling and Connected Factors as Evaluated by Male BRCA1/2 Mutation Carriers in Finland. Journal of Genetic Counseling, 2016, 25, 413-421.	1.6	4
36	Cytopenia, Predisposition to Myelodysplastic Syndrome, Immunodeficiency, and Neurological Disease Caused By Gain-of-Function SAMD9L Mutations Is Frequently Ameliorated By Hematopoietic Revertant Mosaicism. Blood, 2016, 128, 4299-4299.	1.4	1

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37	A new family with autosomal recessive spastic ataxia of Charlevoixâ€“Saguenay (ARSACS). Neuromuscular Disorders, 2015, 25, S223.	0.6	0
38	Biallelic Mutations in BRCA1 Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142.	9.4	251
39	Epidemiology of early-onset Parkinson's disease in Finland. Parkinsonism and Related Disorders, 2015, 21, 938-942.	2.2	24
40	Human Chromosome Y and Haplogroups; introducing YDHS Database. Clinical and Translational Medicine, 2015, 4, 60.	4.0	5
41	The coâ€occurrence of mt<scp>DNA</scp> mutations on different oxidative phosphorylation subunits, not detected by haplogroup analysis, affects human longevity and is population specific. Aging Cell, 2014, 13, 401-407.	6.7	85
42	Breast-Cancer Risk in Families With Mutations in PALB2. Obstetrical and Gynecological Survey, 2014, 69, 659-660.	0.4	1
43	Targeted next-generation sequencing reveals further genetic heterogeneity in axonal Charcotâ€“Marieâ€“Tooth neuropathy and a mutation in HSPB1. European Journal of Human Genetics, 2014, 22, 522-527.	2.8	33
44	Clinical characterization, genetic mapping and whole-genome sequence analysis of a novel autosomal recessive intellectual disability syndrome. European Journal of Medical Genetics, 2014, 57, 543-551.	1.3	19
45	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	27.0	745
46	Mitochondrial DNA variant m.15218Aâ€‰>â€‰G in Finnish epilepsy patients who have maternal relatives with epilepsy, sensorineural hearing impairment or diabetes mellitus. BMC Medical Genetics, 2013, 14, 73.	2.1	7
47	Evaluation of the need for routine clinical testing of PALB2 c.1592delT mutation in BRCA negative Northern Finnish breast cancer families. BMC Medical Genetics, 2013, 14, 82.	2.1	12
48	The phenotype of Floating-Harbor syndrome: clinical characterization of 52 individuals with mutations in exon 34 of SRCAP. Orphanet Journal of Rare Diseases, 2013, 8, 63.	2.7	60
49	Mutations in sFRP1 or sFRP4 are not a common cause of craniotubular hyperostosis. Bone, 2013, 52, 292-295.	2.9	4
50	WFS1 variants in Finnish patients with diabetes mellitus, sensorineural hearing impairment or optic atrophy, and in suicide victims. Journal of Human Genetics, 2013, 58, 495-500.	2.3	7
51	De Novo STX16 Deletions: An Infrequent Cause of Pseudohypoparathyroidism Type Ib that Should Be Excluded in Sporadic Cases. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E2314-E2319.	3.6	32
52	Mitochondrial DNA sequence variation in Finnish patients with matrilineal diabetes mellitus. BMC Research Notes, 2012, 5, 350.	1.4	20
53	Mutations in SRCAP, Encoding SNF2-Related CREBBP Activator Protein, Cause Floating-Harbor Syndrome. American Journal of Human Genetics, 2012, 90, 308-313.	6.2	157
54	Mutations in STAT3 and diagnostic guidelines for hyper-IgE syndrome. Journal of Allergy and Clinical Immunology, 2010, 125, 424-432.e8.	2.9	247

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55	Homozygous W748S mutation in the POLG1 gene in patients with juvenileâ€onset Alpers syndrome and status epilepticus. Epilepsia, 2008, 49, 1038-1045.	5.1	75
56	Genotypeâ€“phenotype analysis in patients with giant axonal neuropathy (GAN). Neuromuscular Disorders, 2007, 17, 624-630.	0.6	29
57	Prevalence, segregation, and phenotype of the mitochondrial DNA 3243A>G mutation in children. Annals of Neurology, 2007, 62, 278-287.	5.3	91
58	Mitochondrial DNA sequence variation and mutation rate in patients with CADASIL. Neurogenetics, 2006, 7, 185-194.	1.4	18
59	Analysis of mitochondrial DNA sequences in patients with isolated or combined oxidative phosphorylation system deficiency. Journal of Medical Genetics, 2006, 43, 881-886.	3.2	21
60	A combination of three common inherited mitochondrial DNA polymorphisms promotes longevity in Finnish and Japanese subjects. European Journal of Human Genetics, 2005, 13, 166-170.	2.8	115
61	Sequence Variation in the tRNA Genes of Human Mitochondrial DNA. Journal of Molecular Evolution, 2005, 60, 587-597.	1.8	20
62	Prevalence of large-scale mitochondrial DNA deletions in an adult Finnish population. Neurology, 2005, 64, 976-981.	1.1	40
63	Mitochondrial DNA polymorphisms as risk factors for Parkinson?s disease and Parkinson?s disease dementia. Human Genetics, 2004, 115, 29-35.	3.8	113
64	Increased variation in mtDNA in patients with familial sensorineural hearing impairment. Human Genetics, 2003, 113, 220-227.	3.8	34
65	Lineage-Specific Selection in Human mtDNA: Lack of Polymorphisms in a Segment of MTND5 Gene in Haplogroup J. Molecular Biology and Evolution, 2003, 20, 2132-2142.	8.9	51
66	Epidemiology of the mitochondrial DNA 8344A>G mutation for the myoclonus epilepsy and ragged red fibres (MERRF) syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2003, 74, 1158-1159.	1.9	25
67	Phylogenetic Network and Physicochemical Properties of Nonsynonymous Mutations in the Protein-Coding Genes of Human Mitochondrial DNA. Molecular Biology and Evolution, 2003, 20, 1195-1210.	8.9	57
68	Complex segregation analysis of Parkinson's disease in the Finnish population. Human Genetics, 2001, 108, 184-189.	3.8	16
69	Hearing impairment in patients with 3243Aâ†'G mtDNA mutation: phenotype and rate of progression. Human Genetics, 2001, 108, 284-289.	3.8	56
70	Relative fitness of carriers of the mitochondrial DNA mutation 3243AÂ>ÂG. European Journal of Human Genetics, 2001, 9, 59-62.	2.8	11
71	Familial aggregation of Parkinson's disease in a Finnish population. Journal of Neurology, Neurosurgery and Psychiatry, 2000, 69, 107-109.	1.9	42
72	Epidemiology of A3243G, the Mutation for Mitochondrial Encephalomyopathy, Lactic Acidosis, and Strokelike Episodes: Prevalence of the Mutation in an Adult Population. American Journal of Human Genetics, 1998, 63, 447-454.	6.2	366