Minna PÃ¶yhÃ¶nen

List of Publications by Year
in descending order

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81	5,130	117625	88630
papers	citations	34 h-index	70 g-index

82	82	82	6240
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Platelet function and filamin A expression in two families with novel <scp>FLNA</scp> gene mutations associated with periventricular nodular heterotopia and panlobular emphysema. American Journal of Medical Genetics, Part A, 2022, , .	1.2	3
2	Genetic analysis reveals novel variants for vascular cognitive impairment. Acta Neurologica Scandinavica, 2022, 146, 42-50.	2.1	6
3	Haploinsufficiency of the NF1 gene is associated with protection against diabetes. Journal of Medical Genetics, 2021, 58, 378-384.	3.2	4
4	Whole-exome sequencing of Finnish patients with vascular cognitive impairment. European Journal of Human Genetics, 2021, 29, 663-671.	2.8	6
5	PHACTR1 genetic variability is not critical in small vessel ischemic disease patients and PcomA recruitment in C57BL/6J mice. Scientific Reports, 2021, 11, 6072.	3.3	2
6	COL4A1 and COL4A2 Duplication Causes Cerebral Small Vessel Disease With Recurrent Early Onset Ischemic Strokes. Stroke, 2021, 52, e624-e625.	2.0	9
7	APOE Îµ4 associates with increased risk of severe COVID-19, cerebral microhaemorrhages and post-COVID mental fatigue: a Finnish biobank, autopsy and clinical study. Acta Neuropathologica Communications, 2021, 9, 199.	5.2	55
8	Chorea-acanthocytosis associated with two novel heterozygous mutations in the VPS13A gene. Journal of the Neurological Sciences, 2020, 408, 116555.	0.6	3
9	Constitutional mosaicism for a BRCA2 mutation as a cause of early-onset breast cancer. Familial Cancer, 2020, 19, 307-310.	1.9	10
10	Oligogenic basis of sporadic ALS. Neurology: Genetics, 2019, 5, e335.	1.9	15
11	Neurofibromatosis type 1 of the child increases birth weight. American Journal of Medical Genetics, Part A, 2019, 179, 1173-1183.	1.2	6
12	Pediatric malignancies in neurofibromatosis type 1: A populationâ€based cohort study. International Journal of Cancer, 2019, 145, 2926-2932.	5.1	36
13	Novel TMEM173 Mutation and the Role of Disease Modifying Alleles. Frontiers in Immunology, 2019, 10, 2770.	4.8	45
14	Genetics of dementia in a Finnish cohort. European Journal of Human Genetics, 2018, 26, 827-837.	2.8	6
15	Breakpoint mapping and haplotype analysis of translocation t(1;12)(q43;q21.1) in two apparently independent families with vascular phenotypes. Molecular Genetics & Genomic Medicine, 2018, 6, 56-68.	1.2	8
16	Identification of OAF and PVRL1 as candidate genes for an ocular anomaly characterized by Peters anomaly type 2 and ectopia lentis. Experimental Eye Research, 2018, 168, 161-170.	2.6	5
17	Congenital anomalies in neurofibromatosis 1: a retrospective register-based total population study. Orphanet Journal of Rare Diseases, 2018, 13, 5.	2.7	23
18	Prevalence of neurofibromatosis type 1 in the Finnish population. Genetics in Medicine, 2018, 20, 1082-1086.	2.4	89

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19	Multi-infarct dementia of Swedish type is caused by a 3â€™UTR mutation of COL4A1. Brain, 2017, 140, e29-e29.	7.6	19
20	The HHID syndrome of hypertrichosis, hyperkeratosis, abnormal corpus callosum, intellectual disability, and minor anomalies is caused by mutations in ARID1B. American Journal of Medical Genetics, Part A, 2017, 173, 1440-1443.	1.2	11
21	Breast cancer in neurofibromatosis type 1: overrepresentation of unfavourable prognostic factors. British Journal of Cancer, 2017, 116, 211-217.	6.4	69
22	008 Congenital anomalies in neurofibromatosis 1: A retrospective register-based total population study. Journal of Investigative Dermatology, 2017, 137, S194.	0.7	0
23	The pregnancy in neurofibromatosis 1: A retrospective registerâ€based total population study. American Journal of Medical Genetics, Part A, 2017, 173, 2641-2648.	1.2	17
24	SNCA mutation p.Ala53Glu is derived from a common founder in the Finnish population. Neurobiology of Aging, 2017, 50, 168.e5-168.e8.	3.1	7
25	Primary familial brain calcification linked to deletion of 5â€™ noncoding region of SLC20A2. Acta Neurologica Scandinavica, 2017, 136, 59-63.	2.1	11
26	The Value of FLG Null Mutations in Predicting Treatment Response in Atopic Dermatitis: An Observational Study in Finnish Patients. Acta Dermato-Venereologica, 2017, 97, 456-463.	1.3	19
27	A Novel Loss-of-Function GRN Mutation p.(Tyr229*):â€ŠClinical and Neuropathological Features. Journal of Alzheimer's Disease, 2016, 55, 1167-1174.	2.6	5
28	Biallelic Variants in UBA5 Link Dysfunctional UFM1ÂUbiquitin-like Modifier Pathway to Severe Infantile-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 683-694.	6.2	72
29	Intrafamilial clinical variability in individuals carrying theCHCHD10mutation Gly66Val. Acta Neurologica Scandinavica, 2016, 133, 361-366.	2.1	21
30	Distinctive Cancer Associations in Patients With Neurofibromatosis Type 1. Journal of Clinical Oncology, 2016, 34, 1978-1986.	1.6	271
31	APOEandAGTin the Finnish p.Arg133Cys CADASIL population. Acta Neurologica Scandinavica, 2015, 132, 430-434.	2.1	2
32	Incidence and Mortality of Neurofibromatosis: A Total Population Study in Finland. Journal of Investigative Dermatology, 2015, 135, 904-906.	0.7	189
33	Neurofibromatosis Type 1 Gene Mutation Analysis Using Sequence Capture and High-throughput Sequencing. Acta Dermato-Venereologica, 2014, 94, 663-666.	1.3	8
34	<scp>CADASIL</scp> and <scp>CARASIL</scp>. Brain Pathology, 2014, 24, 525-544.	4.1	155
35	A novel Î±-synuclein mutation A53E associated with atypical multiple system atrophy and Parkinson's disease-type pathology. Neurobiology of Aging, 2014, 35, 2180.e1-2180.e5.	3.1	396
36	Experimental studies of mitochondrial function in CADASIL vascular smooth muscle cells. Experimental Cell Research, 2013, 319, 134-143.	2.6	24

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37	Neurofibromatosis 1-Related Osteopenia Often Progresses to Osteoporosis in 12ÂYears. Calcified Tissue International, 2013, 92, 23-27.	3.1	20
38	CADASIL Mutations and shRNA Silencing of NOTCH3 Affect Actin Organization in Cultured Vascular Smooth Muscle Cells. Journal of Cerebral Blood Flow and Metabolism, 2012, 32, 2171-2180.	4.3	26
39	A balanced translocation truncates Neurotrimin in a family with intracranial and thoracic aortic aneurysm. Journal of Medical Genetics, 2012, 49, 621-629.	3.2	15
40	Loss of SUFU Function in Familial Multiple Meningioma. American Journal of Human Genetics, 2012, 91, 520-526.	6.2	137
41	Diagnosing Vascular Dementia by Skin Biopsy - Uniqueness of CADASIL. , 2011, , .		1
42	Identification of SPRED1 deletions using RTâ€PCR, multiplex ligationâ€dependent probe amplification and quantitative PCR. American Journal of Medical Genetics, Part A, 2011, 155, 1352-1359.	1.2	15
43	Congruence between NOTCH3 mutations and GOM in 131 CADASIL patients. Brain, 2009, 132, 933-939.	7.6	166
44	Different Clinical Phenotypes in Monozygotic CADASIL Twins With a Novel NOTCH3 Mutation. Stroke, 2009, 40, 2215-2218.	2.0	33
45	Large genomic rearrangements and germline epimutations in Lynch syndrome. International Journal of Cancer, 2009, 124, 2333-2340.	5.1	80
46	Correspondence between the molecular genetic analysis and electron microscopic detection of granular osmiophilic material in CADASIL. Journal of the Neurological Sciences, 2009, 283, 293-294.	0.6	0
47	Molecular genetic and clinical findings in identical twins with CADASIL. Journal of the Neurological Sciences, 2009, 283, 294.	0.6	0
48	Genetic factors modify the clinical course of CADASIL. Journal of the Neurological Sciences, 2009, 283, 294.	0.6	0
49	CADASIL: the most common hereditary subcortical vascular dementia. Future Neurology, 2008, 3, 683-704.	0.5	13
50	Molecular analysis of the CHD7 gene in CHARGE syndrome: identification of 22 novel mutations and evidence for a low contribution of large CHD7 deletions. Genetics in Medicine, 2007, 9, 690-694.	2.4	42
51	Proteome Analysis of Cultivated Vascular Smooth Muscle Cells from a CADASIL Patient. Molecular Medicine, 2007, 13, 305-314.	4.4	36
52	Ophthalmologic Findings in Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy. Ophthalmology, 2006, 113, 1411-1417.e2.	5.2	40
53	Mitochondrial DNA sequence variation and mutation rate in patients with CADASIL. Neurogenetics, 2006, 7, 185-194.	1.4	18
54	Arterioles of the Lenticular Nucleus in CADASIL. Stroke, 2006, 37, 2242-2247.	2.0	56

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55	Decreased bone mineral density and content in neurofibromatosis type 1: Lowest local values are located in the load-carrying parts of the body. Osteoporosis International, 2005, 16, 928-936.	3.1	132
56	Neurologic symptoms are common during gestation and puerperium in CADASIL. Neurology, 2005, 64, 1441-1443.	1.1	31
57	Molecular Analysis of Familial Endometrial Carcinoma: A Manifestation of Hereditary Nonpolyposis Colorectal Cancer or a Separate Syndrome?. Journal of Clinical Oncology, 2005, 23, 4609-4616.	1.6	125
58	Insidious Cognitive Decline in CADASIL. Stroke, 2004, 35, 1598-1602.	2.0	88
59	Scanning Laser Doppler Flowmetry Shows Reduced Retinal Capillary Blood Flow in CADASIL. Stroke, 2004, 35, 2449-2452.	2.0	30
60	Detection of the founder effect in Finnish CADASIL families. European Journal of Human Genetics, 2004, 12, 813-819.	2.8	47
61	Positron Emission Tomography Examination of Cerebral Blood Flow and Glucose Metabolism in Young CADASIL Patients. Stroke, 2004, 35, 1063-1067.	2.0	78
62	Fibrosis and Stenosis of the Long Penetrating Cerebral Arteries: the Cause of the White Matter Pathology in Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy. Brain Pathology, 2004, 14, 358-364.	4.1	123
63	Independent NF1 mutations in two large families with spinal neurofibromatosis. Journal of Medical Genetics, 2003, 40, 122-126.	3.2	33
64	Molecular analysis of astrocytomas presenting after age 10 in individuals with NF1. Neurology, 2003, 61, 1397-1400.	1.1	85
65	Gliomas presenting after age 10 in individuals with neurofibromatosis type 1 (NF1). Neurology, 2002, 59, 759-761.	1.1	139
66	Phenotype of a Homozygous CADASIL Patient in Comparison to 9 Age-Matched Heterozygous Patients With the Same R133C Notch3 Mutation. Stroke, 2001, 32, 1767-1774.	2.0	79
67	Cardiovascular malformations and other cardiovascular abnormalities in neurofibromatosis 1. American Journal of Medical Genetics Part A, 2000, 95, 108-117.	2.4	214
68	New Function for NF1 Tumor Suppressor. Journal of Investigative Dermatology, 2000, 114, 473-479.	0.7	32
69	Mutations in the tyrosine phosphatase CD45 gene in a child with severe combined immunodeficiency disease. Nature Medicine, 2000, 6, 343-345.	30.7	276
70	Occult Neurofibroma and Increased S100 Protein in the Skin of Patients With Neurofibromatosis Type 1. Archives of Dermatology, 2000, 136, 1207-9.	1.4	27
71	Epidemiology of neurofibromatosis type 1 (NF1) in northern Finland. Journal of Medical Genetics, 2000, 37, 632-636.	3.2	56
72	A clinical assessment of neurofibromatosis type 1 (NF1) and segmental NF in northern Finland. Journal of Medical Genetics, 2000, 37, 43e-43.	3.2	17

#	ARTICLE	IF	CITATIONS
73	CADASIL: hereditary disease of arteries causing brain infarcts and dementia. Neuropathology and Applied Neurobiology, 1999, 25, 257-265.	3.2	73
74	Congenital pseudarthrosis of the tibia: Treatment and outcome at skeletal maturity in 10 children. Acta Orthopaedica, 1999, 70, 275-282.	1.4	29
75	A variant of Alzheimer's disease with spastic paraparesis and unusual plaques due to deletion of exon 9 of presenilin 1. Nature Medicine, 1998, 4, 452-455.	30.7	347
76	Evidence of Founder Mutations in Finnish BRCA1 and BRCA2 Families. American Journal of Human Genetics, 1998, 62, 1544-1548.	6.2	77
77	Hereditary spinal neurofibromatosis: a rare form of NF1?. Journal of Medical Genetics, 1997, 34, 184-187.	3.2	58
78	Neuro-ophthalmological findings in neurofibromatosis: Clinical and neuroradiological study of 125 patients. Neuro-Ophthalmology, 1997, 17, 117-126.	1.0	14
79	The structure of the presenilin 1 (S182) gene and identification of six novel mutations in early onset AD families. Nature Genetics, 1995, 11, 219-222.	21.4	461
80	Chromosome 14-encoded Alzheimer's disease: Genetic and clinicopathological description. Annals of Neurology, 1994, 36, 362-367.	5.3	95
81	National Neurofibromatosis Foundation International Database. American Journal of Medical Genetics Part A, 1993, 45, 88-91.	2.4	39