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	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
2	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
3	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
4	Mutations in the tyrosine phosphatase CD45 gene in a child with severe combined immunodeficiency disease. Nature Medicine, 2000, 6, 343-345.	30.7	276
5	Distinctive Cancer Associations in Patients With Neurofibromatosis Type 1. Journal of Clinical Oncology, 2016, 34, 1978-1986.	1.6	271
6	Cardiovascular malformations and other cardiovascular abnormalities in neurofibromatosis 1. American Journal of Medical Genetics Part A, 2000, 95, 108-117.	2.4	214
7	Incidence and Mortality of Neurofibromatosis: A Total Population Study in Finland. Journal of Investigative Dermatology, 2015, 135, 904-906.	0.7	189
8	Congruence between NOTCH3 mutations and GOM in 131 CADASIL patients. Brain, 2009, 132, 933-939.	7.6	166
9	<scp>CADASIL</scp> and <scp>CARASIL</scp>. Brain Pathology, 2014, 24, 525-544.	4.1	155
10	Gliomas presenting after age 10 in individuals with neurofibromatosis type 1 (NF1). Neurology, 2002, 59, 759-761.	1.1	139
11	Loss of SUFU Function in Familial Multiple Meningioma. American Journal of Human Genetics, 2012, 91, 520-526.	6.2	137
12	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
13	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
14	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
15	Chromosome 14-encoded Alzheimer's disease: Genetic and clinicopathological description. Annals of Neurology, 1994, 36, 362-367.	5.3	95
16	Prevalence of neurofibromatosis type 1 in the Finnish population. Genetics in Medicine, 2018, 20, 1082-1086.	2.4	89
17	Insidious Cognitive Decline in CADASIL. Stroke, 2004, 35, 1598-1602.	2.0	88
18	Molecular analysis of astrocytomas presenting after age 10 in individuals with NF1. Neurology, 2003, 61, 1397-1400.	1.1	85

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19	Large genomic rearrangements and germline epimutations in Lynch syndrome. International Journal of Cancer, 2009, 124, 2333-2340.	5.1	80
20	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
21	Positron Emission Tomography Examination of Cerebral Blood Flow and Glucose Metabolism in Young CADASIL Patients. Stroke, 2004, 35, 1063-1067.	2.0	78
22	Evidence of Founder Mutations in Finnish BRCA1 and BRCA2 Families. American Journal of Human Genetics, 1998, 62, 1544-1548.	6.2	77
23	CADASIL: hereditary disease of arteries causing brain infarcts and dementia. Neuropathology and Applied Neurobiology, 1999, 25, 257-265.	3.2	73
24	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
25	Breast cancer in neurofibromatosis type 1: overrepresentation of unfavourable prognostic factors. British Journal of Cancer, 2017, 116, 211-217.	6.4	69
26	Hereditary spinal neurofibromatosis: a rare form of NF1?. Journal of Medical Genetics, 1997, 34, 184-187.	3.2	58
27	Epidemiology of neurofibromatosis type 1 (NF1) in northern Finland. Journal of Medical Genetics, 2000, 37, 632-636.	3.2	56
28	Arterioles of the Lenticular Nucleus in CADASIL. Stroke, 2006, 37, 2242-2247.	2.0	56
29	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
30	Detection of the founder effect in Finnish CADASIL families. European Journal of Human Genetics, 2004, 12, 813-819.	2.8	47
31	Novel TMEM173 Mutation and the Role of Disease Modifying Alleles. Frontiers in Immunology, 2019, 10, 2770.	4.8	45
32	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
33	Ophthalmologic Findings in Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy. Ophthalmology, 2006, 113, 1411-1417.e2.	5.2	40
34	National Neurofibromatosis Foundation International Database. American Journal of Medical Genetics Part A, 1993, 45, 88-91.	2.4	39
35	Proteome Analysis of Cultivated Vascular Smooth Muscle Cells from a CADASIL Patient. Molecular Medicine, 2007, 13, 305-314.	4.4	36
36	Pediatric malignancies in neurofibromatosis type 1: A populationâ€based cohort study. International Journal of Cancer, 2019, 145, 2926-2932.	5.1	36

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37	Independent NF1 mutations in two large families with spinal neurofibromatosis. Journal of Medical Genetics, 2003, 40, 122-126.	3.2	33
38	Different Clinical Phenotypes in Monozygotic CADASIL Twins With a Novel NOTCH3 Mutation. Stroke, 2009, 40, 2215-2218.	2.0	33
39	New Function for NF1 Tumor Suppressor. Journal of Investigative Dermatology, 2000, 114, 473-479.	0.7	32
40	Neurologic symptoms are common during gestation and puerperium in CADASIL. Neurology, 2005, 64, 1441-1443.	1.1	31
41	Scanning Laser Doppler Flowmetry Shows Reduced Retinal Capillary Blood Flow in CADASIL. Stroke, 2004, 35, 2449-2452.	2.0	30
42	Congenital pseudarthrosis of the tibia: Treatment and outcome at skeletal maturity in 10 children. Acta Orthopaedica, 1999, 70, 275-282.	1.4	29
43	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
44	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
45	Experimental studies of mitochondrial function in CADASIL vascular smooth muscle cells. Experimental Cell Research, 2013, 319, 134-143.	2.6	24
46	Congenital anomalies in neurofibromatosis 1: a retrospective register-based total population study. Orphanet Journal of Rare Diseases, 2018, 13, 5.	2.7	23
47	Intrafamilial clinical variability in individuals carrying theCHCHD10mutation Gly66Val. Acta Neurologica Scandinavica, 2016, 133, 361-366.	2.1	21
48	Neurofibromatosis 1-Related Osteopenia Often Progresses to Osteoporosis in 12ÂYears. Calcified Tissue International, 2013, 92, 23-27.	3.1	20
49	Multi-infarct dementia of Swedish type is caused by a 3â€™UTR mutation of COL4A1. Brain, 2017, 140, e29-e29.	7.6	19
50	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
51	Mitochondrial DNA sequence variation and mutation rate in patients with CADASIL. Neurogenetics, 2006, 7, 185-194.	1.4	18
52	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
53	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
54	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

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55	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
56	Oligogenic basis of sporadic ALS. Neurology: Genetics, 2019, 5, e335.	1.9	15
57	Neuro-ophthalmological findings in neurofibromatosis: Clinical and neuroradiological study of 125 patients. Neuro-Ophthalmology, 1997, 17, 117-126.	1.0	14
58	CADASIL: the most common hereditary subcortical vascular dementia. Future Neurology, 2008, 3, 683-704.	0.5	13
59	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
60	Primary familial brain calcification linked to deletion of 5â€™ noncoding region of SLC20A2. Acta Neurologica Scandinavica, 2017, 136, 59-63.	2.1	11
61	Constitutional mosaicism for a BRCA2 mutation as a cause of early-onset breast cancer. Familial Cancer, 2020, 19, 307-310.	1.9	10
62	COL4A1 and COL4A2 Duplication Causes Cerebral Small Vessel Disease With Recurrent Early Onset Ischemic Strokes. Stroke, 2021, 52, e624-e625.	2.0	9
63	Neurofibromatosis Type 1 Gene Mutation Analysis Using Sequence Capture and High-throughput Sequencing. Acta Dermato-Venereologica, 2014, 94, 663-666.	1.3	8
64	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
65	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
66	Genetics of dementia in a Finnish cohort. European Journal of Human Genetics, 2018, 26, 827-837.	2.8	6
67	Neurofibromatosis type 1 of the child increases birth weight. American Journal of Medical Genetics, Part A, 2019, 179, 1173-1183.	1.2	6
68	Whole-exome sequencing of Finnish patients with vascular cognitive impairment. European Journal of Human Genetics, 2021, 29, 663-671.	2.8	6
69	Genetic analysis reveals novel variants for vascular cognitive impairment. Acta Neurologica Scandinavica, 2022, 146, 42-50.	2.1	6
70	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
71	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
72	Haploinsufficiency of the NF1 gene is associated with protection against diabetes. Journal of Medical Genetics, 2021, 58, 378-384.	3.2	4

#	ARTICLE	IF	CITATIONS
73	Chorea-acanthocytosis associated with two novel heterozygous mutations in the VPS13A gene. Journal of the Neurological Sciences, 2020, 408, 116555.	0.6	3
74	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
75	APOEandAGTin the Finnish p.Arg133Cys CADASIL population. Acta Neurologica Scandinavica, 2015, 132, 430-434.	2.1	2
76	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
77	Diagnosing Vascular Dementia by Skin Biopsy - Uniqueness of CADASIL. , 2011, , .		1
78	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
79	Molecular genetic and clinical findings in identical twins with CADASIL. Journal of the Neurological Sciences, 2009, 283, 294.	0.6	0
80	Genetic factors modify the clinical course of CADASIL. Journal of the Neurological Sciences, 2009, 283, 294.	0.6	0
81	008 Congenital anomalies in neurofibromatosis 1: A retrospective register-based total population study. Journal of Investigative Dermatology, 2017, 137, S194.	0.7	0