Mari Auranen

List of Publications by Year
in descending order

44 papers	2,444 citations	279798 23 h-index	243625 44 g-index
66 all docs	66 docs citations	66 times ranked	4205 citing authors

#	Article	IF	Citations
1	Effective treatment of mitochondrial myopathy by nicotinamide riboside, a vitamin <scp>B</scp>3. EMBO Molecular Medicine, 2014, 6, 721-731.	6.9	326
2	A Genomewide Screen for Autism-Spectrum Disorders: Evidence for a Major Susceptibility Locus on Chromosome 3q25-27. American Journal of Human Genetics, 2002, 71, 777-790.	6.2	217
3	Mutations in DNMT3B Modify Epigenetic Repression of the D4Z4 Repeat and the Penetrance of Facioscapulohumeral Dystrophy. American Journal of Human Genetics, 2016, 98, 1020-1029.	6.2	188
4	FGF21 is a biomarker for mitochondrial translation and mtDNA maintenance disorders. Neurology, 2016, 87, 2290-2299.	1.1	167
5	Fibroblast Growth Factor 21 Drives Dynamics of Local and Systemic Stress Responses in Mitochondrial Myopathy with mtDNA Deletions. Cell Metabolism, 2019, 30, 1040-1054.e7.	16.2	166
6	Niacin Cures Systemic NAD+ Deficiency and Improves Muscle Performance in Adult-Onset Mitochondrial Myopathy. Cell Metabolism, 2020, 31, 1078-1090.e5.	16.2	154
7	Search for autism loci by combined analysis of Autism Genetic Resource Exchange and Finnish families. Annals of Neurology, 2006, 59, 145-155.	5.3	152
8	Analysis of four neuroligin genes as candidates for autism. European Journal of Human Genetics, 2005, 13, 1285-1292.	2.8	136
9	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2016, 98, 1130-1145.	6.2	118
10	ATPase-deficient mitochondrial inner membrane protein ATAD3A disturbs mitochondrial dynamics in dominant hereditary spastic paraplegia. Human Molecular Genetics, 2017, 26, 1432-1443.	2.9	63
11	CHCHD10 variant p.(Gly66Val) causes axonal Charcot-Marie-Tooth disease. Neurology: Genetics, 2015, 1, e1.	1.9	62
12	Modified Atkins diet induces subacute selective raggedâ€redâ€fiber lysis in mitochondrial myopathyÂpatients. EMBO Molecular Medicine, 2016, 8, 1234-1247.	6.9	56
13	Analysis of autism susceptibility gene loci on chromosomes 1p, 4p, 6q, 7q, 13q, 15q, 16p, 17q, 19q and 22q in Finnish multiplex families. Molecular Psychiatry, 2000, 5, 320-322.	7.9	50
14	Loss of MICOS complex integrity and mitochondrial damage, but not TDP-43 mitochondrial localisation, are likely associated with severity of CHCHD10-related diseases. Neurobiology of Disease, 2018, 119, 159-171.	4.4	48
15	Dominant transmission of de novo KIF1A motor domain variant underlying pure spastic paraplegia. European Journal of Human Genetics, 2015, 23, 1427-1430.	2.8	44
16	Novel mutations in DNAJB6 gene cause a very severe early-onset limb-girdle muscular dystrophy 1D disease. Neuromuscular Disorders, 2015, 25, 835-842.	0.6	35
17	Diagnostic value of serum biomarkers <scp>FGF21</scp> and <scp>GDF15</scp> compared to muscle sample in mitochondrial disease. Journal of Inherited Metabolic Disease, 2021, 44, 469-480.	3.6	34
18	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

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19	Further evidence for linkage of autosomal-dominant medullary cystic kidney disease on chromosome 1q21. Kidney International, 2001, 60, 1225-1232.	5.2	31
20	CHCHD10 mutations p.R15L and p.G66V cause motoneuron disease by haploinsufficiency. Human Molecular Genetics, 2018, 27, 706-715.	2.9	30
21	Dominant GDAP1 founder mutation is a common cause of axonal Charcot-Marie-Tooth disease in Finland. Neurogenetics, 2013, 14, 123-132.	1.4	28
22	Clinical and metabolic consequences of L-serine supplementation in hereditary sensory and autonomic neuropathy type 1C. Journal of Physical Education and Sports Management, 2017, 3, a002212.	1.2	27
23	Truncated HSPB1 causes axonal neuropathy and impairs tolerance to unfolded protein stress. BBA Clinical, 2015, 3, 233-242.	4.1	26
24	Absence of NEFL in patient-specific neurons in early-onset Charcot-Marie-Tooth neuropathy. Neurology: Genetics, 2018, 4, e244.	1.9	25
25	Screening for late-onset Pompe disease in Finland. Neuromuscular Disorders, 2014, 24, 982-985.	0.6	24
26	Specific functional pathologies of Cx43 mutations associated with oculodentodigital dysplasia. Molecular Biology of the Cell, 2016, 27, 2172-2185.	2.1	20
27	CHCHD10 mutations and motor neuron disease: the distribution in Finnish patients. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 272-277.	1.9	19
28	The Variant p.(Arg183Trp) in SPTLC2 Causes Late-Onset Hereditary Sensory Neuropathy. NeuroMolecular Medicine, 2016, 18, 81-90.	3.4	18
29	IMPDH2: a new gene associated with dominant juvenile-onset dystonia-tremor disorder. European Journal of Human Genetics, 2021, 29, 1833-1837.	2.8	17
30	Effectiveness of clinical exome sequencing in adult patients with difficultâ€toâ€diagnose neurological disorders. Acta Neurologica Scandinavica, 2022, 145, 63-72.	2.1	16
31	Recessive PYROXD1 mutations cause adult-onset limb-girdle-type muscular dystrophy. Journal of Neurology, 2019, 266, 353-360.	3.6	15
32	Beneficial Effects of Ketogenic Diet on Phosphofructokinase Deficiency (Glycogen Storage Disease) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	2.4	15
33	Screening for Fabry disease and Hereditary ATTR amyloidosis in idiopathic smallâ€fiber and mixed neuropathy. Muscle and Nerve, 2019, 59, 354-357.	2.2	12
34	PFKMgene defect and glycogen storage disease GSDVII with misleading enzyme histochemistry. Neurology: Genetics, 2015, 1, e7.	1.9	11
35	Unique Exercise Lactate Profile in Muscle Phosphofructokinase Deficiency (Tarui Disease); Difference Compared with McArdle Disease. Frontiers in Neurology, 2016, 7, 82.	2.4	9
36	Dominant mutations in ITPR3 cause Charcotâ€Marieâ€Tooth disease. Annals of Clinical and Translational Neurology, 2020, 7, 1962-1972.	3.7	9

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37	Threshold of heteroplasmic truncating MT-ATP6 mutation in reprogramming, Notch hyperactivation and motor neuron metabolism. Human Molecular Genetics, 2022, 31, 958-974.	2.9	9
38	De novo SPTAN1mutation in axonal sensorimotor neuropathy and developmental disorder. Brain, 2020, 143, e104-e104.	7.6	8
39	Decreased Aerobic Capacity inÂANO5-Muscular Dystrophy. Journal of Neuromuscular Diseases, 2016, 3, 475-485.	2.6	7
40	Dominant Distal Myopathy 3 (MPD3) Caused by a Deletion in the HNRNPA1 Gene. Neurology: Genetics, 2021, 7, e632.	1.9	7
41	Bi-allelic loss-of-function OBSCN variants predispose individuals to severe recurrent rhabdomyolysis. Brain, 2022, 145, 3985-3998.	7.6	6
42	Serum Creatine, Not Neurofilament Light, Is Elevated in CHCHD10-Linked Spinal Muscular Atrophy. Frontiers in Neurology, 2022, 13, 793937.	2.4	4
43	Modified Atkins diet modifies cardiopulmonary exercise characteristics and promotes hyperventilation in healthy subjects. Journal of Functional Foods, 2021, 81, 104459.	3.4	1
44	Abnormal expression of Î²-dystroglycan in a patient with limb-girdle muscular dystrophy (LGMD). Neuromuscular Disorders, 1997, 7, 439.	0.6	0