

Mari Auranen

List of Publications by Year in descending order

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Version: 2024-02-01

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	Effectiveness of clinical exome sequencing in adult patients with difficult-to-diagnose neurological disorders. <i>Acta Neurologica Scandinavica</i> , 2022, 145, 63-72.	2.1	16
2	Threshold of heteroplasmic truncating MT-ATP6 mutation in reprogramming, Notch hyperactivation and motor neuron metabolism. <i>Human Molecular Genetics</i> , 2022, 31, 958-974.	2.9	9
3	Serum Creatine, Not Neurofilament Light, Is Elevated in CHCHD10-Linked Spinal Muscular Atrophy. <i>Frontiers in Neurology</i> , 2022, 13, 793937.	2.4	4
4	Bi-allelic loss-of-function OBSCN variants predispose individuals to severe recurrent rhabdomyolysis. <i>Brain</i> , 2022, 145, 3985-3998.	7.6	6
5	Diagnostic value of serum biomarkers <scp>FGF21</scp> and <scp>GDF15</scp> compared to muscle sample in mitochondrial disease. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 469-480.	3.6	34
6	Modified Atkins diet modifies cardiopulmonary exercise characteristics and promotes hyperventilation in healthy subjects. <i>Journal of Functional Foods</i> , 2021, 81, 104459.	3.4	1
7	IMPDH2: a new gene associated with dominant juvenile-onset dystonia-tremor disorder. <i>European Journal of Human Genetics</i> , 2021, 29, 1833-1837.	2.8	17
8	Dominant Distal Myopathy 3 (MPD3) Caused by a Deletion in the <i>HNRNPA1</i> Gene. <i>Neurology: Genetics</i> , 2021, 7, e632.	1.9	7
9	<i>De novo SPTAN1</i> mutation in axonal sensorimotor neuropathy and developmental disorder. <i>Brain</i> , 2020, 143, e104-e104.	7.6	8
10	Dominant mutations in ITPR3 cause Charcot-Marie-Tooth disease. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1962-1972.	3.7	9
11	Niacin Cures Systemic NAD+ Deficiency and Improves Muscle Performance in Adult-Onset Mitochondrial Myopathy. <i>Cell Metabolism</i> , 2020, 31, 1078-1090.e5.	16.2	154
12	Beneficial Effects of Ketogenic Diet on Phosphofructokinase Deficiency (Glycogen Storage Disease) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	2.4	15
13	Fibroblast Growth Factor 21 Drives Dynamics of Local and Systemic Stress Responses in Mitochondrial Myopathy with mtDNA Deletions. <i>Cell Metabolism</i> , 2019, 30, 1040-1054.e7.	16.2	166
14	Recessive PYROXD1 mutations cause adult-onset limb-girdle-type muscular dystrophy. <i>Journal of Neurology</i> , 2019, 266, 353-360.	3.6	15
15	Screening for Fabry disease and Hereditary ATTR amyloidosis in idiopathic small-fiber and mixed neuropathy. <i>Muscle and Nerve</i> , 2019, 59, 354-357.	2.2	12
16	CHCHD10 mutations p.R15L and p.G66V cause motoneuron disease by haploinsufficiency. <i>Human Molecular Genetics</i> , 2018, 27, 706-715.	2.9	30
17	Loss of MICOS complex integrity and mitochondrial damage, but not TDP-43 mitochondrial localisation, are likely associated with severity of CHCHD10-related diseases. <i>Neurobiology of Disease</i> , 2018, 119, 159-171.	4.4	48
18	Absence of NEFL in patient-specific neurons in early-onset Charcot-Marie-Tooth neuropathy. <i>Neurology: Genetics</i> , 2018, 4, e244.	1.9	25

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19	<i>CHCHD10</i> mutations and motor neuron disease: the distribution in Finnish patients. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 272-277.	1.9	19
20	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
21	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
22	Unique Exercise Lactate Profile in Muscle Phosphofructokinase Deficiency (Tarui Disease); Difference Compared with McArdle Disease. Frontiers in Neurology, 2016, 7, 82.	2.4	9
23	Modified Atkins diet induces subacute selective raggedâ€redâ€fiber lysis in mitochondrial myopathyâ€patients. EMBO Molecular Medicine, 2016, 8, 1234-1247.	6.9	56
24	Decreased Aerobic Capacity inâ€ANO5-Muscular Dystrophy. Journal of Neuromuscular Diseases, 2016, 3, 475-485.	2.6	7
25	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
26	FGF21 is a biomarker for mitochondrial translation and mtDNA maintenance disorders. Neurology, 2016, 87, 2290-2299.	1.1	167
27	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
28	Specific functional pathologies of Cx43 mutations associated with oculodentodigital dysplasia. Molecular Biology of the Cell, 2016, 27, 2172-2185.	2.1	20
29	The Variant p.(Arg183Trp) in SPTLC2 Causes Late-Onset Hereditary Sensory Neuropathy. NeuroMolecular Medicine, 2016, 18, 81-90.	3.4	18
30	<i>CHCHD10</i> variant p.(Gly66Val) causes axonal Charcot-Marie-Tooth disease. Neurology: Genetics, 2015, 1, e1.	1.9	62
31	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
32	Truncated HSPB1 causes axonal neuropathy and impairs tolerance to unfolded protein stress. BBA Clinical, 2015, 3, 233-242.	4.1	26
33	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
34	PFKM gene defect and glycogen storage disease GSDVII with misleading enzyme histochemistry. Neurology: Genetics, 2015, 1, e7.	1.9	11
35	Effective treatment of mitochondrial myopathy by nicotinamide riboside, a vitamin <sc>B</sc>3. EMBO Molecular Medicine, 2014, 6, 721-731.	6.9	326
36	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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37	Screening for late-onset Pompe disease in Finland. <i>Neuromuscular Disorders</i> , 2014, 24, 982-985.	0.6	24
38	Dominant GDAP1 founder mutation is a common cause of axonal Charcot-Marie-Tooth disease in Finland. <i>Neurogenetics</i> , 2013, 14, 123-132.	1.4	28
39	Search for autism loci by combined analysis of Autism Genetic Resource Exchange and Finnish families. <i>Annals of Neurology</i> , 2006, 59, 145-155.	5.3	152
40	Analysis of four neuroligin genes as candidates for autism. <i>European Journal of Human Genetics</i> , 2005, 13, 1285-1292.	2.8	136
41	A Genomewide Screen for Autism-Spectrum Disorders: Evidence for a Major Susceptibility Locus on Chromosome 3q25-27. <i>American Journal of Human Genetics</i> , 2002, 71, 777-790.	6.2	217
42	Further evidence for linkage of autosomal-dominant medullary cystic kidney disease on chromosome 1q21. <i>Kidney International</i> , 2001, 60, 1225-1232.	5.2	31
43	Analysis of autism susceptibility gene loci on chromosomes 1p, 4p, 6q, 7q, 13q, 15q, 16p, 17q, 19q and 22q in Finnish multiplex families. <i>Molecular Psychiatry</i> , 2000, 5, 320-322.	7.9	50
44	Abnormal expression of β 2-dystroglycan in a patient with limb-girdle muscular dystrophy (LGMD). <i>Neuromuscular Disorders</i> , 1997, 7, 439.	0.6	0