Johanna Palmio

List of Publications by Year in descending order

47 papers	1,606 citations	279798 23 h-index	315739 38 g-index
47 all docs	47 docs citations	47 times ranked	3081 citing authors

#	Article	IF	Citations
1	Mutations affecting the cytoplasmic functions of the co-chaperone DNAJB6 cause limb-girdle muscular dystrophy. Nature Genetics, 2012, 44, 450-455.	21.4	226
2	Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. Neurology, 2016, 86, 391-398.	1.1	107
3	TIA1 variant drives myodegeneration in multisystem proteinopathy with SQSTM1 mutations. Journal of Clinical Investigation, 2018, 128, 1164-1177.	8.2	75
4	Hereditary myopathy with early respiratory failure: occurrence in various populations. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 345-353.	1.9	65
5	Plasma and Cerebrospinal Fluid Amino Acids in Epileptic Patients. Neurochemical Research, 2004, 29, 319-324.	3.3	61
6	Speech deterioration in amyotrophic lateral sclerosis (ALS) after manifestation of bulbar symptoms. International Journal of Language and Communication Disorders, 2018, 53, 385-392.	1.5	60
7	Increase in Plasma Proinflammatory Cytokines After Electroconvulsive Therapy in Patients With Depressive Disorder. Journal of ECT, 2008, 24, 88-91.	0.6	59
8	Distinct distal myopathy phenotype caused by VCP gene mutation in a Finnish family. Neuromuscular Disorders, 2011, 21, 551-555.	0.6	58
9	Ubiquitin Carboxy-Terminal Hydrolase L1 (UCH-L1) is increased in cerebrospinal fluid and plasma of patients after epileptic seizure. BMC Neurology, 2012, 12, 85.	1.8	56
10	Normal CSF neuron-specific enolase and S-100 protein levels in patients with recent non-complicated tonicâ€“clonic seizures. Journal of the Neurological Sciences, 2001, 183, 27-31.	0.6	50
11	Cerebrospinal fluid tau as a marker of neuronal damage after epileptic seizure. Seizure: the Journal of the British Epilepsy Association, 2009, 18, 474-477.	2.0	50
12	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	7.6	45
13	Elevated serum neuron-specific enolase in patients with temporal lobe epilepsy: A videoâ€“EEG study. Epilepsy Research, 2008, 81, 155-160.	1.6	40
14	Diagnostically important muscle pathology in DNAJB6 mutated LGMD1D. Acta Neuropathologica Communications, 2016, 4, 9.	5.2	39
15	Electroconvulsive therapy and biomarkers of neuronal injury and plasticity: Serum levels of neuron-specific enolase and S-100b protein. Psychiatry Research, 2010, 177, 97-100.	3.3	38
16	Targeted Next-Generation Sequencing Reveals Novel TTN Mutations Causing Recessive Distal Titinopathy. Molecular Neurobiology, 2017, 54, 7212-7223.	4.0	38
17	Changes in plasma amino acids after electroconvulsive therapy of depressed patients. Psychiatry Research, 2005, 137, 183-190.	3.3	37
18	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

#	Article	IF	Citations
19	Expanding the importance of HMERF titinopathy: new mutations and clinical aspects. Journal of Neurology, 2019, 266, 680-690.	3.6	31
20	Immunological perspectives of temporal lobe seizures. Journal of Neuroimmunology, 2013, 263, 1-7.	2.3	30
21	A novel FLNC frameshift and an OBSCN variant in a family with distal muscular dystrophy. PLoS ONE, 2017, 12, e0186642.	2.5	29
22	Absence of NEFL in patient-specific neurons in early-onset Charcot-Marie-Tooth neuropathy. Neurology: Genetics, 2018, 4, e244.	1.9	25
23	Screening for late-onset Pompe disease in Finland. Neuromuscular Disorders, 2014, 24, 982-985.	0.6	24
24	Re-evaluation of the phenotype caused by the commonMATR3p.Ser85Cys mutation in a new family. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 448-450.	1.9	24
25	Myasthenic congenital myopathy from recessive mutations at a single residue in Na _V 1.4. Neurology, 2019, 92, e1405-e1415.	1.1	24
26	Neurofilament Light Regulates Axon Caliber, Synaptic Activity, and Organelle Trafficking in Cultured Human Motor Neurons. Frontiers in Cell and Developmental Biology, 2021, 9, 820105.	3.7	23
27	Four new Finnish families with LGMD1D; refinement of the clinical phenotype and the linked 7q36 locus. Neuromuscular Disorders, 2011, 21, 338-344.	0.6	22
28	Actininopathy: A new muscular dystrophy caused by ACTN2 dominant mutations. Annals of Neurology, 2019, 85, 899-906.	5.3	22
29	Predominantly myalgic phenotype caused by the c.3466G>A p.A1156T mutation in SCN4A gene. Neurology, 2017, 88, 1520-1527.	1.1	20
30	Novel valosin-containing protein mutations associated with multisystem proteinopathy. Neuromuscular Disorders, 2018, 28, 491-501.	0.6	20
31	Mutations in the J domain of DNAJB6 cause dominant distal myopathy. Neuromuscular Disorders, 2020, 30, 38-46.	0.6	20
32	CHCHD10 mutations and motor neuron disease: the distribution in Finnish patients. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 272-277.	1.9	19
33	Oral motor functions, speech and communication before a definitive diagnosis of amyotrophic lateral sclerosis. Journal of Communication Disorders, 2016, 61, 97-105.	1.5	16
34	Borderlines between Sarcopenia and Mild Late-Onset Muscle Disease. Frontiers in Aging Neuroscience, 2014, 6, 267.	3.4	15
35	Novel mutation in TNPO3 causes congenital limb-girdle myopathy with slow progression. Neurology: Genetics, 2019, 5, e337.	1.9	14
36	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

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37	PFKMgene defect and glycogen storage disease GSDVII with misleading enzyme histochemistry. Neurology: Genetics, 2015, 1, e7.	1.9	11
38	Late-onset limb-girdle muscular dystrophy caused by GMPPB mutations. Neuromuscular Disorders, 2017, 27, 627-630.	0.6	11
39	CSF and plasma adipokines after tonicâ€“clonic seizures. Seizure: the Journal of the British Epilepsy Association, 2016, 39, 10-12.	2.0	10
40	A novel COL6A2 mutation causing late-onset limb-girdle muscular dystrophy. Journal of Neurology, 2019, 266, 1649-1654.	3.6	10
41	An unusual phenotype of late-onset desminopathy. Neuromuscular Disorders, 2013, 23, 922-923.	0.6	9
42	Unique Exercise Lactate Profile in Muscle Phosphofructokinase Deficiency (Tarui Disease); Difference Compared with McArdle Disease. Frontiers in Neurology, 2016, 7, 82.	2.4	9
43	Comprehensive transcriptomic analysis shows disturbed calcium homeostasis and deregulation of T lymphocyte apoptosis in inclusion body myositis. Journal of Neurology, 2022, 269, 4161-4173.	3.6	8
44	Spontaneous activity in electromyography may differentiate certain benign lower motor neuron disease forms from amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2015, 355, 143-146.	0.6	5
45	Gluteus maximus hypertrophy: A diagnostic clue in four and a half LIM domain 1-mutated reducing body myopathy. Neuromuscular Disorders, 2017, 27, 962-963.	0.6	3
46	Variation in communication strategies in amyotrophic lateral sclerosis during a two-year follow-up. Speech, Language and Hearing, 2018, 21, 123-130.	1.0	1
47	Editorial: Current and Future Developments in the Therapeutic Management of Neuromuscular Diseases. Frontiers in Neurology, 2021, 12, 835839.	2.4	0