# Johanna Palmio

## List of Publications by Year in descending order



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|---|---|---|---|
| 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 | 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 |
| 2 | Editorial: Current and Future Developments in the Therapeutic Management of Neuromuscular Diseases. Frontiers in Neurology, 2021, 12, 835839. | 2.4 | 0 |
| 3 | 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 |
| 4 | Mutations in the J domain of DNAJB6 cause dominant distal myopathy. Neuromuscular Disorders, 2020, 30, 38-46. | 0.6 | 20 |
| 5 | New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708. | 7.6 | 45 |
| 6 | Expanding the importance of HMERF titinopathy: new mutations and clinical aspects. Journal of Neurology, 2019, 266, 680-690. | 3.6 | 31 |
| 7 | Novel mutation in TNPO3 causes congenital limb-girdle myopathy with slow progression. Neurology: Genetics, 2019, 5, e337. | 1.9 | 14 |
| 8 | Actininopathy: A new muscular dystrophy caused by ACTN2 dominant mutations. Annals of Neurology, 2019, 85, 899-906. | 5.3 | 22 |
| 9 | Myasthenic congenital myopathy from recessive mutations at a single residue in Na <sub>V</sub> 1.4. Neurology, 2019, 92, e1405-e1415. | 1.1 | 24 |
| 10 | A novel COL6A2 mutation causing late-onset limb-girdle muscular dystrophy. Journal of Neurology, 2019, 266, 1649-1654. | 3.6 | 10 |
| 11 | Novel valosin-containing protein mutations associated with multisystem proteinopathy. Neuromuscular Disorders, 2018, 28, 491-501. | 0.6 | 20 |
| 12 | 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 |
| 13 | 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 |
| 14 | Absence of NEFL in patient-specific neurons in early-onset Charcot-Marie-Tooth neuropathy. Neurology: Genetics, 2018, 4, e244. | 1.9 | 25 |
| 15 | TIA1 variant drives myodegeneration in multisystem proteinopathy with SQSTM1 mutations. Journal of Clinical Investigation, 2018, 128, 1164-1177. | 8.2 | 75 |
| 16 | CHCHD10 mutations and motor neuron disease: the distribution in Finnish patients. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 272-277. | 1.9 | 19 |
| 17 | Late-onset limb-girdle muscular dystrophy caused by GMPPB mutations. Neuromuscular Disorders, 2017, 27, 627-630. | 0.6 | 11 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Predominantly myalgic phenotype caused by the c.3466G>A p.A1156T mutation in SCN4A gene. Neurology, 2017, 88, 1520-1527. | 1.1 | 20 |
| 20 | Targeted Next-Generation Sequencing Reveals Novel TTN Mutations Causing Recessive Distal Titinopathy. Molecular Neurobiology, 2017, 54, 7212-7223. | 4.0 | 38 |
| 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 | 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 | 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 |
| 24 | CSF and plasma adipokines after tonicâ€“clonic seizures. Seizure: the Journal of the British Epilepsy Association, 2016, 39, 10-12. | 2.0 | 10 |
| 25 | 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 |
| 26 | Diagnostically important muscle pathology in DNAJB6 mutated LGMD1D. Acta Neuropathologica Communications, 2016, 4, 9. | 5.2 | 39 |
| 27 | Mutations in HSPB8 causing a new phenotype of distal myopathy and motor neuropathy. Neurology, 2016, 86, 391-398. | 1.1 | 107 |
| 28 | 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 |
| 29 | 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 |
| 30 | 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 |
| 31 | PFKMgene defect and glycogen storage disease GSDVII with misleading enzyme histochemistry. Neurology: Genetics, 2015, 1, e7. | 1.9 | 11 |
| 32 | Borderlines between Sarcopenia and Mild Late-Onset Muscle Disease. Frontiers in Aging Neuroscience, 2014, 6, 267. | 3.4 | 15 |
| 33 | Hereditary myopathy with early respiratory failure: occurrence in various populations. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 345-353. | 1.9 | 65 |
| 34 | Screening for late-onset Pompe disease in Finland. Neuromuscular Disorders, 2014, 24, 982-985. | 0.6 | 24 |
| 35 | Immunological perspectives of temporal lobe seizures. Journal of Neuroimmunology, 2013, 263, 1-7. | 2.3 | 30 |
| 36 | An unusual phenotype of late-onset desminopathy. Neuromuscular Disorders, 2013, 23, 922-923. | 0.6 | 9 |

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|---|---|---|---|
| 37 | Mutations affecting the cytoplasmic functions of the co-chaperone DNAJB6 cause limb-girdle muscular dystrophy. Nature Genetics, 2012, 44, 450-455. | 21.4 | 226 |
| 38 | 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 |
| 39 | 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 |
| 40 | Distinct distal myopathy phenotype caused by VCP gene mutation in a Finnish family. Neuromuscular Disorders, 2011, 21, 551-555. | 0.6 | 58 |
| 41 | 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 |
| 42 | 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 |
| 43 | Elevated serum neuron-specific enolase in patients with temporal lobe epilepsy: A videoâ€“EEG study. Epilepsy Research, 2008, 81, 155-160. | 1.6 | 40 |
| 44 | Increase in Plasma Proinflammatory Cytokines After Electroconvulsive Therapy in Patients With Depressive Disorder. Journal of ECT, 2008, 24, 88-91. | 0.6 | 59 |
| 45 | Changes in plasma amino acids after electroconvulsive therapy of depressed patients. Psychiatry Research, 2005, 137, 183-190. | 3.3 | 37 |
| 46 | Plasma and Cerebrospinal Fluid Amino Acids in Epileptic Patients. Neurochemical Research, 2004, 29, 319-324. | 3.3 | 61 |
| 47 | 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 |