Jodi Warman Chardon

List of Publications by Year
in descending order

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56 papers	1,045 citations	471371 17 h-index	454834 30 g-index
60 all docs	60 docs citations	60 times ranked	2348 citing authors

#	Article	IF	Citations
1	Missense mutations in ITPR1 cause autosomal dominant congenital nonprogressive spinocerebellar ataxia. Orphanet Journal of Rare Diseases, 2012, 7, 67.	1.2	124
2	Exome Sequencing as a Diagnostic Tool for Pediatricâ€Onset Ataxia. Human Mutation, 2014, 35, 45-49.	1.1	91
3	Clinical and genetic study of hereditary spastic paraplegia in Canada. Neurology: Genetics, 2017, 3, e122.	0.9	82
4	Abnormal fatty acid metabolism is a core component of spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1519-1532.	1.7	72
5	Identification of a methylation profile for DNMT1-associated autosomal dominant cerebellar ataxia, deafness, and narcolepsy. Clinical Epigenetics, 2016, 8, 91.	1.8	66
6	Targeted exome analysis identifies the genetic basis of disease in over 50% of patients with a wide range of ataxia-related phenotypes. Genetics in Medicine, 2019, 21, 195-206.	1.1	65
7	PDXK mutations cause polyneuropathy responsive to pyridoxal 5â€²â€phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	2.8	54
8	MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscular Disorders, 2019, 29, 827-841.	0.3	46
9	Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. Lancet Neurology, The, 2020, 19, 522-532.	4.9	36
10	Axons to Exons: the Molecular Diagnosis of Rare Neurological Diseases by Next-Generation Sequencing. Current Neurology and Neuroscience Reports, 2015, 15, 64.	2.0	29
11	Novel ELOVL4 mutation associated with erythrokeratodermia and spinocerebellar ataxia (SCA) Tj ETQq1 1 0.784314 rgBT /Ove	0.9	27
12	Autoimmune peripheral neuropathies. Clinica Chimica Acta, 2015, 449, 37-42.	0.5	26
13	Neurolymphomatosis of the Brachial Plexus and its Branches: Case Series and Literature Review. Canadian Journal of Neurological Sciences, 2018, 45, 137-143.	0.3	26
14	Whole-transcriptome sequencing in blood provides a diagnosis of spinal muscular atrophy with progressive myoclonic epilepsy. Human Mutation, 2017, 38, 611-614.	1.1	25
15	LIMS2 mutations are associated with a novel muscular dystrophy, severe cardiomyopathy and triangular tongues. Clinical Genetics, 2015, 88, 558-564.	1.0	23
16	Recent Advances in the Genetic Etiology of Brain Malformations. Current Neurology and Neuroscience Reports, 2013, 13, 364.	2.0	20
17	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a coâ€occurrence of multiple events. American Journal of Medical Genetics, Part A, 2016, 170, 1820-1825.	0.7	19
18	Novel Recessive TNNT1 Congenital Coreâ€Rod Myopathy in French Canadians. Annals of Neurology, 2020, 87, 568-583.	2.8	19

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19	The utility of exome sequencing for genetic diagnosis in a familial microcephaly epilepsy syndrome. BMC Neurology, 2014, 14, 22.	0.8	18
20	Neurolymphomatosis of the lumbosacral plexus and its branches: case series and literature review. BMC Cancer, 2019, 19, 1149.	1.1	16
21	Survival Motor Neuron Protein is Released from Cells in Exosomes: A Potential Biomarker for Spinal Muscular Atrophy. Scientific Reports, 2017, 7, 13859.	1.6	13
22	Deletion of AFG3L2 associated with spinocerebellar ataxia type 28 in the context of multiple genomic anomalies. American Journal of Medical Genetics, Part A, 2014, 164, 3209-3212.	0.7	11
23	The Role of Muscle Imaging in the Diagnosis and Assessment of Children with Genetic Muscle Disease. Neuropediatrics, 2017, 48, 233-241.	0.3	11
24	247th ENMC International Workshop: Muscle magnetic resonance imaging - Implementing muscle MRI as a diagnostic tool for rare genetic myopathy cohorts. Hoofddorp, The Netherlands, September 2019. Neuromuscular Disorders, 2020, 30, 938-947.	0.3	11
25	The Added Value of Cardiac Magnetic Resonance in Muscular Dystrophies. Journal of Neuromuscular Diseases, 2019, 6, 389-399.	1.1	10
26	Intrafamilial variability of limb-girdle muscular dystrophy, LGMD1D type. European Journal of Medical Genetics, 2020, 63, 103655.	0.7	10
27	Temporal evolution of nerve conduction study abnormalities in antiâ€myelinâ€associated glycoprotein neuropathy. Muscle and Nerve, 2021, 63, 401-404.	1.0	10
28	Combined isolated trigeminal and facial neuropathies from perineural invasion by squamous cell carcinoma: A case series and review of the literature. Journal of Clinical Neuroscience, 2017, 35, 5-12.	0.8	7
29	Age matters. Neurology: Neuroimmunology and NeuroInflammation, 2019, 6, e576.	3.1	7
30	A Canadian Adult Spinal Muscular Atrophy Outcome Measures Toolkit: Results of a National Consensus using a Modified Delphi Method. Journal of Neuromuscular Diseases, 2021, 8, 579-588.	1.1	7
31	Deletion of filamin A in two female patients with periventricular nodular heterotopia. American Journal of Medical Genetics, Part A, 2012, 158A, 1512-1516.	0.7	6
32	A National Spinal Muscular Atrophy Registry for Real-World Evidence. Canadian Journal of Neurological Sciences, 2020, 47, 810-815.	0.3	6
33	Laryngospasm in amyotrophic lateral sclerosis. Muscle and Nerve, 2022, 65, 400-404.	1.0	6
34	Supramaximal Stimulus Intensity as a Diagnostic Tool in Chronic Demyelinating Neuropathy. Neuroscience Journal, 2016, 2016, 1-5.	2.3	5
35	Impact of disuse muscular atrophy on the compound muscle action potential. Muscle and Nerve, 2020, 61, 58-62.	1.0	5
36	Very lateâ€onset Sandhoff disease presenting as Kennedy Disease. Muscle and Nerve, 2015, 52, 1135-1136.	1.0	4

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37	Amyloid Neuropathy Following Domino Liver Transplantation. JAMA Neurology, 2016, 73, 477.	4.5	4
38	Autosomal dominant cerebellar ataxia, deafness, and narcolepsy (ADCA-DN) associated with progressive cognitive and behavioral deterioration.. Neuropsychology, 2017, 31, 292-303.	1.0	4
39	Diffuse leukoencephalopathy with spheroids presenting as primary progressive aphasia. Neurology, 2016, 86, 1464-1465.	1.5	3
40	Autologous Hematopoietic Stem Cell Transplantation for Chronic Inflammatory Demyelinating Polyradiculoneuropathy. Canadian Journal of Neurological Sciences, 2021, , 1-7.	0.3	3
41	Whole genome sequencing reveals biallelic <scp>PLA2G6</scp> mutations in siblings with cerebellar atrophy and cap myopathy. Clinical Genetics, 2021, 99, 746-748.	1.0	3
42	A crucial first randomized controlled trial of thymectomy in non-thymomatous myasthenia gravis. Journal of Thoracic Disease, 2016, 8, E1375-E1378.	0.6	2
43	Cardiac Amyloidosis Phenotype Associated With a Glu89Lys Transthyretin Mutation. Canadian Journal of Cardiology, 2017, 33, 830.e5-830.e7.	0.8	2
44	A Survey of Cerebrospinal Fluid Total Protein Upper Limits in Canada: Time for an Update?. Canadian Journal of Neurological Sciences, 2019, 46, 283-286.	0.3	2
45	Distal Cervical Spondylotic Amyotrophy: Case Reports Demonstrating Clinical/Imaging Segmental Discrepancy. Journal of Clinical Neuromuscular Disease, 2019, 21, 107-111.	0.3	2
46	Marked enlargement of neck circumference from nerve hypertrophy in CIDP. Neurology, 2016, 87, 442-442.	1.5	1
47	Teaching Video NeuroImages: Rippling muscle disease with caveolin myopathy. Neurology, 2018, 91, e1726-e1727.	1.5	1
48	Intermittent undulating tongue as an involuntary movement in early amyotrophic lateral sclerosis. Parkinsonism and Related Disorders, 2019, 67, 1-2.	1.1	1
49	Myofibrillar Myopathy Mimicking Polyneuropathy. Case Reports in Neurology, 2020, 12, 97-102.	0.3	1
50	Systematic analysis of clinical deficits in unilateral hypoglossal nerve palsy. Muscle and Nerve, 2016, 54, 1055-1058.	1.0	0
51	Teaching Video Neuro Images : Trapezius muscle hypertrophy in multifocal motor neuropathy. Neurology, 2017, 89, e81-e82.	1.5	0
52	Intraneural Ganglion Cysts of the Fibular Nerve: A Cause of Fluctuating Painful Foot Drop. Canadian Journal of Neurological Sciences, 2018, 45, 601-603.	0.3	0
53	Dataset for worldwide survey of cerebrospinal total protein upper reference values. Data in Brief, 2019, 23, 103760.	0.5	0
54	Does Diabetes Alter CSF Total Protein Levels? A Retrospective Cohort Study. Neurohospitalist, The, 0, , 194187442110393.	0.3	0

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55	Pseudohypertrophy of the extensor digitorum brevis in diabetic polyneuropathy. Muscle and Nerve, 2021, 64, E20-E22.	1.0	0
56	MuSK not MNGIE: Atypical MuSK-antibody myasthenia presenting as a genetic disorder. Neuromuscular Disorders, 2021, , .	0.3	0