

Jodi Warman Chardon

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2484409/publications.pdf>

Version: 2024-02-01

56
papers

1,045
citations

471509

17
h-index

454955

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.	2.7	124
2	Exome Sequencing as a Diagnostic Tool for Pediatric Onset Ataxia. Human Mutation, 2014, 35, 45-49.	2.5	91
3	Clinical and genetic study of hereditary spastic paraplegia in Canada. Neurology: Genetics, 2017, 3, e122.	1.9	82
4	Abnormal fatty acid metabolism is a core component of spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 1519-1532.	3.7	72
5	Identification of a methylation profile for DNMT1-associated autosomal dominant cerebellar ataxia, deafness, and narcolepsy. Clinical Epigenetics, 2016, 8, 91.	4.1	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.	2.4	65
7	<i>PDXK</i> mutations cause polyneuropathy responsive to pyridoxal 5-phosphate supplementation. Annals of Neurology, 2019, 86, 225-240.	5.3	54
8	MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscular Disorders, 2019, 29, 827-841.	0.6	46
9	Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. Lancet Neurology, The, 2020, 19, 522-532.	10.2	36
10	Axons to Exons: the Molecular Diagnosis of Rare Neurological Diseases by Next-Generation Sequencing. Current Neurology and Neuroscience Reports, 2015, 15, 64.	4.2	29
11	Novel <i>ELOVL4</i> mutation associated with erythrokeratoderma and spinocerebellar ataxia (SCA) Tj ETQq1 1 0,784314 rgBT /Ov	1.9	27
12	Autoimmune peripheral neuropathies. Clinica Chimica Acta, 2015, 449, 37-42.	1.1	26
13	Neurolymphomatosis of the Brachial Plexus and its Branches: Case Series and Literature Review. Canadian Journal of Neurological Sciences, 2018, 45, 137-143.	0.5	26
14	Whole-transcriptome sequencing in blood provides a diagnosis of spinal muscular atrophy with progressive myoclonic epilepsy. Human Mutation, 2017, 38, 611-614.	2.5	25
15	LIMS2 mutations are associated with a novel muscular dystrophy, severe cardiomyopathy and triangular tongues. Clinical Genetics, 2015, 88, 558-564.	2.0	23
16	Recent Advances in the Genetic Etiology of Brain Malformations. Current Neurology and Neuroscience Reports, 2013, 13, 364.	4.2	20
17	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a co-occurrence of multiple events. American Journal of Medical Genetics, Part A, 2016, 170, 1820-1825.	1.2	19
18	Novel Recessive <i>TNNT1</i> Congenital Core Rod Myopathy in French Canadians. Annals of Neurology, 2020, 87, 568-583.	5.3	19

#	ARTICLE	IF	CITATIONS
19	The utility of exome sequencing for genetic diagnosis in a familial microcephaly epilepsy syndrome. BMC Neurology, 2014, 14, 22.	1.8	18
20	Neurolymphomatosis of the lumbosacral plexus and its branches: case series and literature review. BMC Cancer, 2019, 19, 1149.	2.6	16
21	Survival Motor Neuron Protein is Released from Cells in Exosomes: A Potential Biomarker for Spinal Muscular Atrophy. Scientific Reports, 2017, 7, 13859.	3.3	13
22	Deletion of <i>AFG3L2</i> associated with spinocerebellar ataxia type 28 in the context of multiple genomic anomalies. American Journal of Medical Genetics, Part A, 2014, 164, 3209-3212.	1.2	11
23	The Role of Muscle Imaging in the Diagnosis and Assessment of Children with Genetic Muscle Disease. Neuropediatrics, 2017, 48, 233-241.	0.6	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.	0.6	11
25	The Added Value of Cardiac Magnetic Resonance in Muscular Dystrophies. Journal of Neuromuscular Diseases, 2019, 6, 389-399.	2.6	10
26	Intrafamilial variability of limb-girdle muscular dystrophy, LGMD1D type. European Journal of Medical Genetics, 2020, 63, 103655.	1.3	10
27	Temporal evolution of nerve conduction study abnormalities in anti-Myelin-associated glycoprotein neuropathy. Muscle and Nerve, 2021, 63, 401-404.	2.2	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.	1.5	7
29	Age matters. Neurology: Neuroimmunology and NeuroInflammation, 2019, 6, e576.	6.0	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.	2.6	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.	1.2	6
32	A National Spinal Muscular Atrophy Registry for Real-World Evidence. Canadian Journal of Neurological Sciences, 2020, 47, 810-815.	0.5	6
33	Laryngospasm in amyotrophic lateral sclerosis. Muscle and Nerve, 2022, 65, 400-404.	2.2	6
34	Supramaximal Stimulus Intensity as a Diagnostic Tool in Chronic Demyelinating Neuropathy. Neuroscience Journal, 2016, 2016, 1-5.	2.5	5
35	Impact of disuse muscular atrophy on the compound muscle action potential. Muscle and Nerve, 2020, 61, 58-62.	2.2	5
36	Very late-onset Sandhoff disease presenting as Kennedy Disease. Muscle and Nerve, 2015, 52, 1135-1136.	2.2	4

#	ARTICLE	IF	CITATIONS
37	Amyloid Neuropathy Following Domino Liver Transplantation. <i>JAMA Neurology</i> , 2016, 73, 477.	9.0	4
38	Autosomal dominant cerebellar ataxia, deafness, and narcolepsy (ADCA-DN) associated with progressive cognitive and behavioral deterioration.. <i>Neuropsychology</i> , 2017, 31, 292-303.	1.3	4
39	Diffuse leukoencephalopathy with spheroids presenting as primary progressive aphasia. <i>Neurology</i> , 2016, 86, 1464-1465.	1.1	3
40	Autologous Hematopoietic Stem Cell Transplantation for Chronic Inflammatory Demyelinating Polyradiculoneuropathy. <i>Canadian Journal of Neurological Sciences</i> , 2021, , 1-7.	0.5	3
41	Whole genome sequencing reveals biallelic <i>PLA2G6</i> mutations in siblings with cerebellar atrophy and cap myopathy. <i>Clinical Genetics</i> , 2021, 99, 746-748.	2.0	3
42	A crucial first randomized controlled trial of thymectomy in non-thymomatous myasthenia gravis. <i>Journal of Thoracic Disease</i> , 2016, 8, E1375-E1378.	1.4	2
43	Cardiac Amyloidosis Phenotype Associated With a Glu89Lys Transthyretin Mutation. <i>Canadian Journal of Cardiology</i> , 2017, 33, 830.e5-830.e7.	1.7	2
44	A Survey of Cerebrospinal Fluid Total Protein Upper Limits in Canada: Time for an Update?. <i>Canadian Journal of Neurological Sciences</i> , 2019, 46, 283-286.	0.5	2
45	Distal Cervical Spondylotic Amyotrophy: Case Reports Demonstrating Clinical/Imaging Segmental Discrepancy. <i>Journal of Clinical Neuromuscular Disease</i> , 2019, 21, 107-111.	0.7	2
46	Marked enlargement of neck circumference from nerve hypertrophy in CIDP. <i>Neurology</i> , 2016, 87, 442-442.	1.1	1
47	Teaching Video NeuroImages: Rippling muscle disease with caveolin myopathy. <i>Neurology</i> , 2018, 91, e1726-e1727.	1.1	1
48	Intermittent undulating tongue as an involuntary movement in early amyotrophic lateral sclerosis. <i>Parkinsonism and Related Disorders</i> , 2019, 67, 1-2.	2.2	1
49	Myofibrillar Myopathy Mimicking Polyneuropathy. <i>Case Reports in Neurology</i> , 2020, 12, 97-102.	0.7	1
50	Systematic analysis of clinical deficits in unilateral hypoglossal nerve palsy. <i>Muscle and Nerve</i> , 2016, 54, 1055-1058.	2.2	0
51	Teaching Video Neuro Images : Trapezius muscle hypertrophy in multifocal motor neuropathy. <i>Neurology</i> , 2017, 89, e81-e82.	1.1	0
52	Intraneural Ganglion Cysts of the Fibular Nerve: A Cause of Fluctuating Painful Foot Drop. <i>Canadian Journal of Neurological Sciences</i> , 2018, 45, 601-603.	0.5	0
53	Dataset for worldwide survey of cerebrospinal total protein upper reference values. <i>Data in Brief</i> , 2019, 23, 103760.	1.0	0
54	Does Diabetes Alter CSF Total Protein Levels? A Retrospective Cohort Study. <i>Neurohospitalist</i> , The, 0, , 194187442110393.	0.8	0

#	ARTICLE	IF	CITATIONS
55	Pseudohypertrophy of the extensor digitorum brevis in diabetic polyneuropathy. <i>Muscle and Nerve</i> , 2021, 64, E20-E22.	2.2	0
56	MuSK not MNGIE: Atypical MuSK-antibody myasthenia presenting as a genetic disorder. <i>Neuromuscular Disorders</i> , 2021, , .	0.6	0