

Megan A Waldrop

List of Publications by Year in descending order

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Version: 2024-02-01

28
papers

650
citations

567281

15
h-index

610901

24
g-index

32
all docs

32
docs citations

32
times ranked

936
citing authors

#	ARTICLE	IF	CITATIONS
1	Intron mutations and early transcription termination in Duchenne and Becker muscular dystrophy. <i>Human Mutation</i> , 2022, 43, 511-528.	2.5	16
2	Phenotypic Spectrum of Dystrophinopathy Due to Duchenne Muscular Dystrophy Exon 2 Duplications. <i>Neurology</i> , 2022, 98, .	1.1	6
3	Overview of gene therapy in spinal muscular atrophy and Duchenne muscular dystrophy. <i>Pediatric Pulmonology</i> , 2021, 56, 710-720.	2.0	31
4	Spinal Muscular Atrophy. <i>Seminars in Pediatric Neurology</i> , 2021, 37, 100878.	2.0	35
5	Time is muscle: A recommendation for early treatment for preterm infants with spinal muscular atrophy. <i>Muscle and Nerve</i> , 2021, 64, 153-155.	2.2	11
6	Absence of Significant Off-Target Splicing Variation with a U7snRNA Vector Targeting <i>DMD</i> Exon 2 Duplications. <i>Human Gene Therapy</i> , 2021, 32, 1346-1359.	2.7	8
7	Validity and Reliability of the Neuromuscular Gross Motor Outcome. <i>Pediatric Neurology</i> , 2021, 122, 21-26.	2.1	5
8	Lack of Toxicity in Nonhuman Primates Receiving Clinically Relevant Doses of an AAV9.U7snRNA Vector Designed to Induce <i>DMD</i> Exon 2 Skipping. <i>Human Gene Therapy</i> , 2021, 32, 882-894.	2.7	29
9	Truncating variants in <i>UBAP1</i> associated with childhood-onset nonsyndromic hereditary spastic paraplegia. <i>Human Mutation</i> , 2020, 41, 632-640.	2.5	15
10	Homozygous variants in <i>AMPD2</i> and <i>COL11A1</i> lead to a complex phenotype of pontocerebellar hypoplasia type 9 and Stickler syndrome type 2. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 557-560.	1.2	5
11	Natural History of Steroid-Treated Young Boys With Duchenne Muscular Dystrophy Using the NSAA, 100m, and Timed Functional Tests. <i>Pediatric Neurology</i> , 2020, 113, 15-20.	2.1	14
12	Spinal Muscular Atrophy in the Treatment Era. <i>Neurologic Clinics</i> , 2020, 38, 505-518.	1.8	19
13	Gene Therapy for Spinal Muscular Atrophy: Safety and Early Outcomes. <i>Pediatrics</i> , 2020, 146, .	2.1	82
14	Clinical Phenotypes of <i>DMD</i> Exon 51 Skip Equivalent Deletions: A Systematic Review. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 217-229.	2.6	18
15	The Genotypic and Phenotypic Spectrum of <i>BICD2</i> Variants in Spinal Muscular Atrophy. <i>Annals of Neurology</i> , 2020, 87, 487-496.	5.3	18
16	A Novel De Novo Heterozygous <i>SCN4a</i> Mutation Causing Congenital Myopathy, Myotonia and Multiple Congenital Anomalies. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 467-473.	2.6	9
17	Diagnostic Utility of Whole Exome Sequencing in the Neuromuscular Clinic. <i>Neuropediatrics</i> , 2019, 50, 096-102.	0.6	28
18	Variants in <i>MED12L</i> , encoding a subunit of the mediator kinase module, are responsible for intellectual disability associated with transcriptional defect. <i>Genetics in Medicine</i> , 2019, 21, 2713-2722.	2.4	28

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19	Current Treatment Options in Neurology—SMA Therapeutics. Current Treatment Options in Neurology, 2019, 21, 25.	1.8	47
20	Update in Duchenne and Becker muscular dystrophy. Current Opinion in Neurology, 2019, 32, 722-727.	3.6	71
21	Low-level dystrophin expression attenuating the dystrophinopathy phenotype. Neuromuscular Disorders, 2018, 28, 116-121.	0.6	27
22	Low-level expression of EPG5 leads to an attenuated Vici syndrome phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 1207-1211.	1.2	9
23	In-frame de novo mutation in <i>BICD2</i> in two patients with muscular atrophy and arthrogryposis. Journal of Physical Education and Sports Management, 2018, 4, a003160.	1.2	14
24	Clinicopathologic Conference: A Newborn With Hypotonia, Cleft Palate, Micrognathia, and Bilateral Club Feet. Pediatric Neurology, 2017, 74, 11-14.	2.1	6
25	Expression of Neurexin, Neuroligin, and Their Cytoplasmic Binding Partners in the Pancreatic β -Cells and the Involvement of Neuroligin in Insulin Secretion. Endocrinology, 2008, 149, 6006-6017.	2.8	64
26	Release of Glutamate Decarboxylase-65 into the Circulation by Injured Pancreatic Islet β -Cells. Endocrinology, 2007, 148, 4572-4578.	2.8	19
27	A Highly Sensitive Immunoassay Resistant to Autoantibody Interference for Detection of the Diabetes-Associated Autoantigen Glutamic Acid Decarboxylase 65 in Blood and Other Biological Samples. Diabetes Technology and Therapeutics, 2006, 8, 207-218.	4.4	15
28	An evaluation of onasemnogene abeparvovec for Spinal Muscular Atrophy (SMN1). Expert Opinion on Orphan Drugs, 0, , .	0.8	0