

Centronuclear (myotubular) myopathy

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Citation Report

#	ARTICLE	IF	CITATIONS
1	T-tubule disorganization and defective excitation-contraction coupling in muscle fibers lacking myotubularin lipid phosphatase. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 18763-18768.	3.3	167
2	Phenotypic spectrum of dynamin 2 mutations in Charcot-Marie-Tooth neuropathy. Brain, 2009, 132, 1741-1752.	3.7	134
3	Loss of Myotubularin Function Results in T-Tubule Disorganization in Zebrafish and Human Myotubular Myopathy. PLoS Genetics, 2009, 5, e1000372.	1.5	201
4	Diverse roles of the actin cytoskeleton in striated muscle. Journal of Muscle Research and Cell Motility, 2009, 30, 187-197.	0.9	50
7	The role of muscle biopsy in the age of genetic testing. Current Opinion in Neurology, 2009, 22, 543-553.	1.8	9
9	Dynamin 2 and human diseases. Journal of Molecular Medicine, 2010, 88, 339-350.	1.7	112
10	Mutation studies in X-linked myotubular myopathy in three Indian families. Indian Journal of Pediatrics, 2010, 77, 431-433.	0.3	3
11	Ryanodine receptor calcium channels and their partners as drug targets. Biochemical Pharmacology, 2010, 79, 1535-1543.	2.0	69
12	STIM proteins: integrators of signalling pathways in development, differentiation and disease. Journal of Cellular and Molecular Medicine, 2010, 14, 1890-1903.	1.6	47
13	Vici syndrome associated with sensorineural hearing loss and evidence of neuromuscular involvement on muscle biopsy. American Journal of Medical Genetics, Part A, 2010, 152A, 741-747.	0.7	40
14	<i>RYR1</i> mutations are a common cause of congenital myopathies with central nuclei. Annals of Neurology, 2010, 68, 717-726.	2.8	230
15	Prenatal diagnosis of X-linked myotubular myopathy. Prenatal Diagnosis, 2010, 30, 177-179.	1.1	2
16	A centronuclear myopathy-dynamin 2 mutation impairs skeletal muscle structure and function in mice. Human Molecular Genetics, 2010, 19, 4820-4836.	1.4	107
17	Dynamin 2 Mutants Linked to Centronuclear Myopathies Form Abnormally Stable Polymers. Journal of Biological Chemistry, 2010, 285, 22753-22757.	1.6	71
18	Zebrafish MTMR14 is required for excitation-contraction coupling, developmental motor function and the regulation of autophagy. Human Molecular Genetics, 2010, 19, 2668-2681.	1.4	73
19	Ryanodinopathies. Current Topics in Membranes, 2010, 66, 139-167.	0.5	6
20	Case report of intrafamilial variability in autosomal recessive centronuclear myopathy associated to a novel BIN1 stop mutation. Orphanet Journal of Rare Diseases, 2010, 5, 35.	1.2	51
21	Centronuclear myopathy with cataracts due to a novel dynamin 2 (DNM2) mutation. Neuromuscular Disorders, 2010, 20, 49-52.	0.3	37

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22	Adult course in dynamin 2 dominant centronuclear myopathy with neonatal onset. <i>Neuromuscular Disorders</i> , 2010, 20, 53-56.	0.3	28
23	Novel molecular diagnostic approaches for X-linked centronuclear (myotubular) myopathy reveal intronic mutations. <i>Neuromuscular Disorders</i> , 2010, 20, 375-381.	0.3	33
24	Sporadic centronuclear myopathy with muscle pseudohypertrophy, neutropenia, and necklace fibers due to a DNMT2 mutation. <i>Neuromuscular Disorders</i> , 2010, 20, 801-804.	0.3	40
25	Inhibition of Activin Receptor Type IIB Increases Strength and Lifespan in Myotubularin-Deficient Mice. <i>American Journal of Pathology</i> , 2011, 178, 784-793.	1.9	63
26	Increased Expression of Wild-Type or a Centronuclear Myopathy Mutant of Dynamin 2 in Skeletal Muscle of Adult Mice Leads to Structural Defects and Muscle Weakness. <i>American Journal of Pathology</i> , 2011, 178, 2224-2235.	1.9	84
27	Dilated cardiomyopathy with centronuclear myopathy in a young male. <i>International Journal of Cardiology</i> , 2011, 150, 213-216.	0.8	4
28	Impaired neuromuscular transmission and response to acetylcholinesterase inhibitors in centronuclear myopathies. <i>Neuromuscular Disorders</i> , 2011, 21, 379-386.	0.3	96
29	Endplate structure and parameters of neuromuscular transmission in sporadic centronuclear myopathy associated with myasthenia. <i>Neuromuscular Disorders</i> , 2011, 21, 387-395.	0.3	39
31	Misregulated alternative splicing of BIN1 is associated with T tubule alterations and muscle weakness in myotonic dystrophy. <i>Nature Medicine</i> , 2011, 17, 720-725.	15.2	299
32	Muscle Imaging in Congenital Myopathies. <i>Seminars in Pediatric Neurology</i> , 2011, 18, 221-229.	1.0	70
33	Defects in amphiphysin 2 (BIN1) and triads in several forms of centronuclear myopathies. <i>Acta Neuropathologica</i> , 2011, 121, 253-266.	3.9	113
34	Phenotype variability and histopathological findings in centronuclear myopathy due to DNMT2 mutations. <i>Journal of Neurology</i> , 2011, 258, 1085-1090.	1.8	43
35	Cellular self-organization by autocatalytic alignment feedback. <i>Journal of Cell Science</i> , 2011, 124, 4213-4220.	1.2	49
36	Phosphatase-Dead Myotubularin Ameliorates X-Linked Centronuclear Myopathy Phenotypes in Mice. <i>PLoS Genetics</i> , 2012, 8, e1002965.	1.5	49
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38	Histopathological features in subsequent muscle biopsies in a warmblood mare with myotonic dystrophy. <i>Veterinary Quarterly</i> , 2012, 32, 187-192.	3.0	5
39	Modeling the human MTM1 p.R69C mutation in murine Mtm1 results in exon 4 skipping and a less severe myotubular myopathy phenotype. <i>Human Molecular Genetics</i> , 2012, 21, 811-825.	1.4	54
40	Myotubularins and associated neuromuscular diseases. <i>Clinical Lipidology</i> , 2012, 7, 151-162.	0.4	6

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41	Samaritan myopathy, an ultimately benign congenital myopathy, is caused by a RYR1 mutation. <i>Acta Neuropathologica</i> , 2012, 124, 575-581.	3.9	22
42	Establishing Clinical End Points of Respiratory Function in Large Animals for Clinical Translation. <i>Physical Medicine and Rehabilitation Clinics of North America</i> , 2012, 23, 75-94.	0.7	15
43	Myotubularin-Deficient Myoblasts Display Increased Apoptosis, Delayed Proliferation, and Poor Cell Engraftment. <i>American Journal of Pathology</i> , 2012, 181, 961-968.	1.9	37
44	Consensus Statement on Standard of Care for Congenital Myopathies. <i>Journal of Child Neurology</i> , 2012, 27, 363-382.	0.7	147
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49	Necklace fibers as histopathological marker in a patient with severe form of X-linked myotubular myopathy. <i>Neuromuscular Disorders</i> , 2012, 22, 541-545.	0.3	15
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54	Myotubular myopathy and the neuromuscular junction: a novel therapeutic approach from mouse models. <i>DMM Disease Models and Mechanisms</i> , 2012, 5, 852-9.	1.2	43
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58	Barfly: Sculpting membranes at the <i>Drosophila</i> neuromuscular junction. <i>Developmental Neurobiology</i> , 2012, 72, 33-56.	1.5	5
59	Congenital (Structural) Myopathies. , 2013, , 1-51.		0
60	Defective Autophagy and mTORC1 Signaling in Myotubularin Null Mice. <i>Molecular and Cellular Biology</i> , 2013, 33, 98-110.	1.1	74

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63	Role of phosphatidylinositol 5-phosphate 4-kinase $\hat{\pm}$ in zebrafish development. <i>International Journal of Biochemistry and Cell Biology</i> , 2013, 45, 1293-1301.	1.2	17
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68	A novel mutation in the DNM2 gene impairs dynamin 2 localization in skeletal muscle of a patient with late onset centronuclear myopathy. <i>Neuromuscular Disorders</i> , 2013, 23, 219-228.	0.3	23
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73	Enzyme replacement therapy rescues weakness and improves muscle pathology in mice with X-linked myotubular myopathy. <i>Human Molecular Genetics</i> , 2013, 22, 1525-1538.	1.4	71
74	Site-specific Mtm1 mutagenesis by an AAV-Cre vector reveals that myotubularin is essential in adult muscle. <i>Human Molecular Genetics</i> , 2013, 22, 1856-1866.	1.4	17
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80	Reducing dynamin 2 expression rescues X-linked centronuclear myopathy. <i>Journal of Clinical Investigation</i> , 2014, 124, 1350-1363.	3.9	115
82	Congenital (Structural) Myopathies. , 2014, , .		2
83	Novel excitation-contraction coupling related genes reveal aspects of muscle weakness beyond atrophy—new hopes for treatment of musculoskeletal diseases. <i>Frontiers in Physiology</i> , 2014, 5, 37.	1.3	37
84	Gene Therapy for Inherited Muscle Diseases. <i>American Journal of Physical Medicine and Rehabilitation</i> , 2014, 93, S97-S107.	0.7	19
85	Gene Therapy Prolongs Survival and Restores Function in Murine and Canine Models of Myotubular Myopathy. <i>Science Translational Medicine</i> , 2014, 6, 220ra10.	5.8	141
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93	Contracciã³n versus contractura y miopatÃa del nÃºcleo central versus miopatÃa de la parte central en hipertermia maligna. <i>Brazilian Journal of Anesthesiology (Edicion En Espanol)</i> , 2014, 64, 142-144.	0.0	0
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95	Differential Muscle Hypertrophy Is Associated with Satellite Cell Numbers and Akt Pathway Activation Following Activin Type IIB Receptor Inhibition in Mtm1 p.R69C Mice. <i>American Journal of Pathology</i> , 2014, 184, 1831-1842.	1.9	29
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101	Review of X-linked syndromes with arthrogryposis or early contractures—aid to diagnosis and pathway identification. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 931-973.	0.7	18
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104	Peripheral nerve and neuromuscular junction pathology in Pompe disease. <i>Human Molecular Genetics</i> , 2015, 24, 625-636.	1.4	63
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106	Phospholamban overexpression in mice causes a centronuclear myopathy-like phenotype. <i>DMM Disease Models and Mechanisms</i> , 2015, 8, 999-1009.	1.2	29
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108	Congenital and Other Structural Myopathies. , 2015, , 499-537.		3
109	Neuromuscular blocking effects of vecuronium in dogs with autosomal-recessive centronuclear myopathy. <i>American Journal of Veterinary Research</i> , 2015, 76, 302-307.	0.3	5
110	Clinical Phenotype of X-linked Myotubular Myopathy in Labrador Retriever Puppies. <i>Journal of Veterinary Internal Medicine</i> , 2015, 29, 254-260.	0.6	13
111	<i>HACD1</i> , a regulator of membrane composition and fluidity, promotes myoblast fusion and skeletal muscle growth. <i>Journal of Molecular Cell Biology</i> , 2015, 7, 429-440.	1.5	40
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117	Clinical, Electrophysiology, and Pathology Features of Dynamin Centronuclear Myopathy: A Case Report and Review of Literature. <i>Journal of Clinical Neuromuscular Disease</i> , 2016, 18, 84-88.	0.3	3
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122	Prenatal diagnosis of congenital myopathies and muscular dystrophies. <i>Clinical Genetics</i> , 2016, 90, 199-210.	1.0	20
123	Vici syndrome: a review. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 21.	1.2	55
124	Canine-Inherited Dystrophinopathies and Centronuclear Myopathies. <i>Pancreatic Islet Biology</i> , 2016, , 309-329.	0.1	5
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127	<i>EPG5</i> -related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. <i>Brain</i> , 2016, 139, 765-781.	3.7	99
128	A Study of a Cohort of X-Linked Myotubular Myopathy at the Clinical, Histologic, and Genetic Levels. <i>Pediatric Neurology</i> , 2016, 58, 107-112.	1.0	13
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131	Muscle MRI in pediatrics: clinical, pathological and genetic correlation. <i>Pediatric Radiology</i> , 2017, 47, 724-735.	1.1	12
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133	Microtubule motors involved in nuclear movement during skeletal muscle differentiation. <i>Molecular Biology of the Cell</i> , 2017, 28, 865-874.	0.9	43
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136	Long-term effects of systemic gene therapy in a canine model of myotubular myopathy. <i>Muscle and Nerve</i> , 2017, 56, 943-953.	1.0	50
137	Novel intronic mutation in <i>MTM1</i> detected by RNA analysis in a case of X-linked myotubular myopathy. <i>Neurology: Genetics</i> , 2017, 3, e182.	0.9	9
138	X-linked myotubular myopathy. <i>Neurology</i> , 2017, 89, 1316-1317.	1.5	1
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144	Single Intramuscular Injection of AAV-shRNA Reduces DNM2 and Prevents Myotubular Myopathy in Mice. <i>Molecular Therapy</i> , 2018, 26, 1082-1092.	3.7	35
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147	Nuclear positioning in skeletal muscle. <i>Seminars in Cell and Developmental Biology</i> , 2018, 82, 51-56.	2.3	143
148	A multicenter, retrospective medical record review of X-linked myotubular myopathy: The recensus study. <i>Muscle and Nerve</i> , 2018, 57, 550-560.	1.0	54
149	Reducing dynamin 2 (DNM2) rescues <i>DNM2</i> -related dominant centronuclear myopathy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 11066-11071.	3.3	50
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151	Neuromuscular Disorders in Newborns. , 2018, , 2323-2335.		0
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153	Centronuclear myopathy: advances in genetic understanding and potential for future treatments. <i>Expert Opinion on Orphan Drugs</i> , 2018, 6, 375-384.	0.5	0

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156	Novel Variants in Individuals with RYR1-Related Congenital Myopathies: Genetic, Laboratory, and Clinical Findings. Frontiers in Neurology, 2018, 9, 118.	1.1	11
157	Neonatal Neuromuscular Disorders. , 2018, , 952-960.e2.		3
158	Neonatal hypotonia and neuromuscular conditions. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2019, 162, 435-448.	1.0	13
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163	Molecular Basis of Muscle Disease. , 2019, , 13-39.		1
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165	Three novel MTM1 pathogenic variants identified in Japanese patients with X-linked myotubular myopathy. Molecular Genetics & Genomic Medicine, 2019, 7, e621.	0.6	4
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167	The expanding spectrum of neurological disorders of phosphoinositide metabolism. DMM Disease Models and Mechanisms, 2019, 12, .	1.2	23
168	Mutations in genes associated with either myopathy or noncompaction. Herz, 2019, 44, 756-758.	0.4	2
169	Mortality and respiratory support in X-linked myotubular myopathy: a RECENSUS retrospective analysis. Archives of Disease in Childhood, 2020, 105, 332-338.	1.0	24
170	Centronuclear Myopathy with Abundant Nemaline Rods in a Japanese Black and Hereford Crossbred Calf. Journal of Comparative Pathology, 2020, 174, 8-12.	0.1	0
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#	ARTICLE	IF	CITATIONS
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