

Recessive RYR1 mutations cause unusual congenital myopathy with
myofibrillar disorganization, sarcomeric internalization and large areas of myofibrillar disorganization

Neuropathology and Applied Neurobiology

37, 271-284

DOI: [10.1111/j.1365-2990.2010.01149.x](https://doi.org/10.1111/j.1365-2990.2010.01149.x)

Citation Report

#	ARTICLE	IF	CITATIONS
1	Identical de novo Mutation in the Type 1 Ryanodine Receptor Gene Associated with Fatal, Stress-induced Malignant Hyperthermia in Two Unrelated Families. <i>Anesthesiology</i> , 2011, 115, 938-945.	1.3	83
2	Core Myopathies. <i>Seminars in Pediatric Neurology</i> , 2011, 18, 239-249.	1.0	120
3	Centronuclear Myopathies. <i>Seminars in Pediatric Neurology</i> , 2011, 18, 250-256.	1.0	84
4	T-tubule biogenesis and triad formation in skeletal muscle and implication in human diseases. <i>Skeletal Muscle</i> , 2011, 1, 26.	1.9	143
5	Clinical utility gene card for: Multi-minicore disease. <i>European Journal of Human Genetics</i> , 2012, 20, 5-5.	1.4	2
6	Samaritan myopathy, an ultimately benign congenital myopathy, is caused by a RYR1 mutation. <i>Acta Neuropathologica</i> , 2012, 124, 575-581.	3.9	22
7	Dominant Mutation of CCDC78 in a Unique Congenital Myopathy with Prominent Internal Nuclei and Atypical Cores. <i>American Journal of Human Genetics</i> , 2012, 91, 365-371.	2.6	84
8	Mapping domains and mutations on the skeletal muscle ryanodine receptor channel. <i>Trends in Molecular Medicine</i> , 2012, 18, 644-657.	3.5	47
9	Clinical utility gene card for: Centronuclear and myotubular myopathies. <i>European Journal of Human Genetics</i> , 2012, 20, 1101-1101.	1.4	28
10	Centronuclear Myopathy in Labrador Retrievers: A Recent Founder Mutation in the PTPLA Gene Has Rapidly Disseminated Worldwide. <i>PLoS ONE</i> , 2012, 7, e46408.	1.1	21
11	Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. <i>Human Mutation</i> , 2012, 33, 981-988.	1.1	145
12	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. <i>Human Mutation</i> , 2012, 33, 949-959.	1.1	115
13	Congenital Myopathies: An Update. <i>Current Neurology and Neuroscience Reports</i> , 2012, 12, 165-174.	2.0	106
14	Swimming into prominence: the zebrafish as a valuable tool for studying human myopathies and muscular dystrophies. <i>FEBS Journal</i> , 2013, 280, 4187-4197.	2.2	63
15	Congenital (Structural) Myopathies. , 2013, , 1-51.		0
16	Congenital myopathy with focal loss of cross-striations revisited. <i>Neuromuscular Disorders</i> , 2013, 23, 160-164.	0.3	1
17	Muscle diseases with prominent joint contractures: Main entities and diagnostic strategy. <i>Revue Neurologique</i> , 2013, 169, 546-563.	0.6	13
18	198th ENMC International Workshop: 7th Workshop on Centronuclear (Myotubular) myopathies, 31st May - 2nd June 2013, Naarden, The Netherlands. <i>Neuromuscular Disorders</i> , 2013, 23, 1033-1043.	0.3	14

#	ARTICLE	IF	CITATIONS
19	Adult centronuclear myopathies: A hospital-based study. <i>Revue Neurologique</i> , 2013, 169, 625-631.	0.6	7
20	Congenital myopathies. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2013, 113, 1321-1336.	1.0	61
21	Core myopathies and malignant hyperthermia susceptibility: a review. <i>Paediatric Anaesthesia</i> , 2013, 23, 834-841.	0.6	48
22	Genotype-phenotype correlations in recessive RYR1-related myopathies. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 117.	1.2	99
23	The neuronal endopeptidase ECEL1 is associated with a distinct form of recessive distal arthropogryposis. <i>Human Molecular Genetics</i> , 2013, 22, 1483-1492.	1.4	66
24	Expanding the MTM1 mutational spectrum: novel variants including the first multi-exonic duplication and development of a locus-specific database. <i>European Journal of Human Genetics</i> , 2013, 21, 540-549.	1.4	29
25	Severe congenital <i>RYR1</i> -associated myopathy. <i>Neurology</i> , 2013, 80, 1584-1589.	1.5	91
26	<i>RYR1</i> Mutations as a Cause of Ophthalmoplegia, Facial Weakness, and Malignant Hyperthermia. <i>JAMA Ophthalmology</i> , 2013, 131, 1532.	1.4	26
27	An Integrated Diagnosis Strategy for Congenital Myopathies. <i>PLoS ONE</i> , 2013, 8, e67527.	1.1	53
28	Pathogenic Mechanisms in Centronuclear Myopathies. <i>Frontiers in Aging Neuroscience</i> , 2014, 6, 339.	1.7	89
29	Congenital (Structural) Myopathies. , 2014, , .		2
30	Bridging integrator 1 (Bin1) deficiency in zebrafish results in centronuclear myopathy. <i>Human Molecular Genetics</i> , 2014, 23, 3566-3578.	1.4	28
31	G.P.48. <i>Neuromuscular Disorders</i> , 2014, 24, 809.	0.3	0
32	SPEG Interacts with Myotubularin, and Its Deficiency Causes Centronuclear Myopathy with Dilated Cardiomyopathy. <i>American Journal of Human Genetics</i> , 2014, 95, 218-226.	2.6	143
33	Expanding genotype/phenotype of neuromuscular diseases by comprehensive target capture/NGS. <i>Neurology: Genetics</i> , 2015, 1, e14.	0.9	48
34	Malignant hyperthermia: a review. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 93.	1.2	392
35	<i>RYR1</i> -related myopathies: a wide spectrum of phenotypes throughout life. <i>European Journal of Neurology</i> , 2015, 22, 1094-1112.	1.7	111
36	DNM2 mutations in a cohort of sporadic patients with centronuclear myopathy. <i>Genetics and Molecular Biology</i> , 2015, 38, 147-151.	0.6	11

#	ARTICLE	IF	CITATIONS
37	Epigenetic changes as a common trigger of muscle weakness in congenital myopathies. <i>Human Molecular Genetics</i> , 2015, 24, 4636-4647.	1.4	44
38	A novel missense mutation of RYR1 in familial idiopathic hyper CK-emia. <i>Journal of the Neurological Sciences</i> , 2015, 356, 142-147.	0.3	8
39	Compound RYR1 heterozygosity resulting in a complex phenotype of malignant hyperthermia susceptibility and a core myopathy. <i>Neuromuscular Disorders</i> , 2015, 25, 567-576.	0.3	28
40	A novel dynamin-2 gene mutation associated with a late-onset centronuclear myopathy with necklace fibres. <i>Neuromuscular Disorders</i> , 2015, 25, 345-348.	0.3	15
41	Congenital and Other Structural Myopathies. , 2015, , 499-537.		3
42	Anesthetic considerations in myofibrillar myopathy. <i>Paediatric Anaesthesia</i> , 2015, 25, 231-238.	0.6	2
43	Next generation sequencing reveals ryanodine receptor 1 mutations in a Chinese central core disease cohort. <i>Muscle and Nerve</i> , 2016, 54, 432-438.	1.0	3
44	Congenital myopathies: not only a paediatric topic. <i>Current Opinion in Neurology</i> , 2016, 29, 642-650.	1.8	37
45	Review of RyR1 pathway and associated pathomechanisms. <i>Acta Neuropathologica Communications</i> , 2016, 4, 121.	2.4	66
46	Intra-familial variability associated with recessive RYR1 mutation diagnosed prenatally by exome sequencing. <i>Prenatal Diagnosis</i> , 2016, 36, 1020-1026.	1.1	13
47	Mouse myofibers lacking the SMYD1 methyltransferase are susceptible to atrophy, internalization of nuclei and myofibrillar disarray. <i>DMM Disease Models and Mechanisms</i> , 2016, 9, 347-359.	1.2	29
48	Gene Discovery in Congenital Myopathy. <i>Pancreatic Islet Biology</i> , 2016, , 39-83.	0.1	0
49	Common and variable clinical, histological, and imaging findings of recessive RYR1-related centronuclear myopathy patients. <i>Neuromuscular Disorders</i> , 2017, 27, 975-985.	0.3	34
50	Dihydropyridine receptor (DHPR, CACNA1S) congenital myopathy. <i>Acta Neuropathologica</i> , 2017, 133, 517-533.	3.9	97
51	Progressive Structural Defects in Canine Centronuclear Myopathy Indicate a Role for HACD1 in Maintaining Skeletal Muscle Membrane Systems. <i>American Journal of Pathology</i> , 2017, 187, 441-456.	1.9	13
52	<i>RYR1</i> causing distal myopathy. <i>Molecular Genetics & Genomic Medicine</i> , 2017, 5, 800-804.	0.6	14
53	Affected female carriers of MTM1 mutations display a wide spectrum of clinical and pathological involvement: delineating diagnostic clues. <i>Acta Neuropathologica</i> , 2017, 134, 889-904.	3.9	42
54	New era in genetics of early-onset muscle disease: Breakthroughs and challenges. <i>Seminars in Cell and Developmental Biology</i> , 2017, 64, 160-170.	2.3	24

#	ARTICLE	IF	CITATIONS
55	Congenital myopathies: clinical phenotypes and new diagnostic tools. Italian Journal of Pediatrics, 2017, 43, 101.	1.0	80
56	SPEG-deficient skeletal muscles exhibit abnormal triad and defective calcium handling. Human Molecular Genetics, 2018, 27, 1608-1617.	1.4	22
58	Hereditary Myopathies. , 0, , .		8
59	Ryanodine Receptor 1-Related Myopathies: Diagnostic and Therapeutic Approaches. Neurotherapeutics, 2018, 15, 885-899.	2.1	81
60	Characterization and genetic diagnosis of centronuclear myopathies in seven Chinese patients. Neurological Sciences, 2018, 39, 2043-2051.	0.9	1
61	Loss of Sarcomeric Scaffolding as a Common Baseline Histopathologic Lesion in Titin-Related Myopathies. Journal of Neuropathology and Experimental Neurology, 2018, 77, 1101-1114.	0.9	22
62	A Rare Case of Severe Congenital RYR1-Associated Myopathy. Case Reports in Genetics, 2018, 2018, 1-7.	0.1	6
63	Correlation of phenotype with genotype and protein structure in RYR1-related disorders. Journal of Neurology, 2018, 265, 2506-2524.	1.8	29
64	Centronuclear myopathy: advances in genetic understanding and potential for future treatments. Expert Opinion on Orphan Drugs, 2018, 6, 375-384.	0.5	0
65	Centronuclear myopathies under attack: A plethora of therapeutic targets. Journal of Neuromuscular Diseases, 2018, 5, 387-406.	1.1	34
66	6-minute walk test as a measure of disease progression and fatigability in a cohort of individuals with RYR1-related myopathies. Orphanet Journal of Rare Diseases, 2018, 13, 105.	1.2	19
67	Bayesian modeling to predict malignant hyperthermia susceptibility and pathogenicity of <i>RYR1</i>, <i>CACNA1S</i> and <i>STAC3</i> variants. Pharmacogenomics, 2019, 20, 989-1003.	0.6	1
68	Update on the Genetics of Congenital Myopathies. Seminars in Pediatric Neurology, 2019, 29, 12-22.	1.0	22
69	Diagnosis of "possible" mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	1.5	42
70	RYR1 Sequence Variants in Myopathies: Expression and Functional Studies in Two Families. BioMed Research International, 2019, 2019, 1-13.	0.9	7
71	Mouse model of severe recessive RYR1-related myopathy. Human Molecular Genetics, 2019, 28, 3024-3036.	1.4	22
72	An unusual ryanodine receptor 1 (RYR1) phenotype. Neurology, 2019, 92, e1600-e1609.	1.5	16
73	The histopathological spectrum of malignant hyperthermia and rhabdomyolysis due to RYR1 mutations. Journal of Neurology, 2019, 266, 876-887.	1.8	26

#	ARTICLE	IF	CITATIONS
74	Novel <i>SPEG</i> mutations in congenital myopathies: Genotype-phenotype correlations. <i>Muscle and Nerve</i> , 2019, 59, 357-362.	1.0	17
75	“Dusty core disease” (DuCD): expanding morphological spectrum of RYR1 recessive myopathies. <i>Acta Neuropathologica Communications</i> , 2019, 7, 3.	2.4	31
76	Novel <i>SPEG</i> variant cause centronuclear myopathy in China. <i>Journal of Clinical Laboratory Analysis</i> , 2020, 34, e23054.	0.9	13
77	Pathogenic Mutations and Putative Phenotype-Affecting Variants in Polish Myofibrillar Myopathy Patients. <i>Journal of Clinical Medicine</i> , 2021, 10, 914.	1.0	6
78	Ryanodine Receptor 1-Related Myopathies: Quantification of Intramuscular Fatty Infiltration from T1-Weighted MRI. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 657-668.	1.1	4
79	Marked Facial Weakness, Ptosis, and Hanging Jaw: A Case with RYR1-Related Congenital Centronuclear Myopathy. <i>Journal of Pediatric Genetics</i> , 2023, 12, 318-324.	0.3	0
80	Nonanesthetic Malignant Hyperthermia. <i>Anesthesiology</i> , 2011, 115, 915-917.	1.3	24
81	A rare case of centronuclear myopathy with <i>DNM2</i> mutation: genotype phenotype correlation. <i>Autopsy and Case Reports</i> , 2017, 7, 43-48.	0.2	4
82	Common Pathogenic Mechanisms in Centronuclear and Myotubular Myopathies and Latest Treatment Advances. <i>International Journal of Molecular Sciences</i> , 2021, 22, 11377.	1.8	21
86	An 8-year-old boy with delayed motor milestones and proximal leg muscle weakness. , 2020, , 269-274.		0
87	A 6-Week-Old Boy with Neonatal Hypotonia and Feeding and Respiratory Difficulties. , 2020, , 283-288.		0
88	Malignant Hyperthermia. <i>Missouri Medicine</i> , 2019, 116, 154-159.	0.3	6
89	<i>BIN1</i> modulation in vivo rescues dynamin-related myopathy. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2022, 119, .	3.3	10
90	Improvement of muscle strength in a mouse model for congenital myopathy treated with HDAC and DNA methyltransferase inhibitors. <i>ELife</i> , 2022, 11, .	2.8	7
91	Recessive RYR1-related centronuclear myopathy with congenital chylothorax in a Japanese male child. <i>Medicine, Case Reports and Study Protocols</i> , 2021, 2, e0190.	0.0	0
93	Mutations in proteins involved in E-C coupling and SOCE and congenital myopathies. <i>Journal of General Physiology</i> , 2022, 154, .	0.9	3
94	Gene Panel Sequencing Identifies a Novel RYR1 p.Ser2300Pro Variant as Candidate for Malignant Hyperthermia with Multi-Minicore Myopathy. <i>Genes</i> , 2022, 13, 1726.	1.0	1
95	Prenatal diagnosis identifies compound heterozygous variants in RYR1 that causes ultrasound abnormalities in a fetus. <i>BMC Medical Genomics</i> , 2022, 15, .	0.7	0

#	ARTICLE	IF	CITATIONS
---	---------	----	-----------