

CITATION REPORT

List of articles citing

RYR1 mutations are a common cause of congenital myopathies with central nuclei

DOI: 10.1002/ana.22119

Annals of Neurology, 2010, 68, 717-26.

Source: <https://exaly.com/paper-pdf/48126278/citation-report.pdf>

Version: 2024-04-28

This report has been generated based on the citations recorded by exaly.com for the above article. For the latest version of this publication list, visit the link given above.

The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

#	Paper	IF	Citations
216	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. 2011 , 178, 2224-35		65
215	Response. 2011 , 21, 148-149		1
214	Impaired neuromuscular transmission and response to acetylcholinesterase inhibitors in centronuclear myopathies. 2011 , 21, 379-86		78
213	Endplate structure and parameters of neuromuscular transmission in sporadic centronuclear myopathy associated with myasthenia. 2011 , 21, 387-95		36
212	King-Denborough syndrome with and without mutations in the skeletal muscle ryanodine receptor (RYR1) gene. 2011 , 21, 420-7		76
211	Identical de novo mutation in the type 1 ryanodine receptor gene associated with fatal, stress-induced malignant hyperthermia in two unrelated families. 2011 , 115, 938-45		63
210	Current world literature. 2011 , 23, 620-5		
209	Mining for mutations in malignant hyperthermia. 2011 , 113, 975-6		2
208	Misregulated alternative splicing of BIN1 is associated with T tubule alterations and muscle weakness in myotonic dystrophy. 2011 , 17, 720-5		228
207	Defects in Ca ²⁺ release associated with local expression of pathological ryanodine receptors in mouse muscle fibres. 2011 , 589, 5361-82		17
206	Clinical utility gene card for: malignant hyperthermia. 2011 , 19,		15
205	Core myopathies. 2011 , 18, 239-49		104
204	Centronuclear myopathies. 2011 , 18, 250-6		70
203	Congenital fiber-type disproportion. 2011 , 18, 264-71		47
202	T-tubule biogenesis and triad formation in skeletal muscle and implication in human diseases. 2011 , 1, 26		104
201	Prevalence of congenital myopathies in a representative pediatric united states population. <i>Annals of Neurology</i> , 2011 , 70, 662-5	9.4	93
200	Alterations of excitation-contraction coupling and excitation coupled Ca(2+) entry in human myotubes carrying CAV3 mutations linked to rippling muscle. 2011 , 32, 309-17		12

199	Further Delineation of the Phenotype of Congenital Disorder of Glycosylation DPAGT1-CDG (CDG-Ij) Identified by Homozygosity Mapping. 2012 , 2, 107-11	14
198	Enhanced excitation-coupled Ca(2+) entry induces nuclear translocation of NFAT and contributes to IL-6 release from myotubes from patients with central core disease. 2011 , 20, 589-600	19
197	Muscle magnetic resonance imaging in congenital myopathies due to ryanodine receptor type 1 gene mutations. 2011 , 68, 1171-9	72
196	Normal myofibrillar development followed by progressive sarcomeric disruption with actin accumulations in a mouse Cfl2 knockout demonstrates requirement of cofilin-2 for muscle maintenance. 2012 , 21, 2341-56	63
195	Clinical utility gene card for: Central core disease. 2012 , 20,	3
194	Clinical utility gene card for: Multi-minicore disease. 2012 , 20,	2
193	Samaritan myopathy, an ultimately benign congenital myopathy, is caused by a RYR1 mutation. 2012 , 124, 575-81	16
192	Consensus statement on standard of care for congenital myopathies. 2012 , 27, 363-82	104
191	X-linked myotubular myopathy due to a complex rearrangement involving a duplication of MTM1 exon 10. 2012 , 22, 384-8	10
190	182nd ENMC International Workshop: RYR1-related myopathies, 15-17th April 2011, Naarden, The Netherlands. 2012 , 22, 453-62	19
189	Myopathic causes of exercise intolerance with rhabdomyolysis. 2012 , 54, 886-91	47
188	The influence of DNA sequence on epigenome-induced pathologies. 2012 , 5, 11	16
187	Mapping domains and mutations on the skeletal muscle ryanodine receptor channel. 2012 , 18, 644-57	42
186	Clinical utility gene card for: Centronuclear and myotubular myopathies. 2012 , 20,	23
185	Myotubular myopathy and the neuromuscular junction: a novel therapeutic approach from mouse models. <i>DMM Disease Models and Mechanisms</i> , 2012 , 5, 852-9	4.1 39
184	Centronuclear myopathy in Labrador retrievers: a recent founder mutation in the PTPLA gene has rapidly disseminated worldwide. 2012 , 7, e46408	17
183	Oxidative stress and successful antioxidant treatment in models of RYR1-related myopathy. 2012 , 135, 1115-27	88
182	Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. 2012 , 33, 981-8	119

181	Mutation spectrum in the large GTPase dynamin 2, and genotype-phenotype correlation in autosomal dominant centronuclear myopathy. 2012 , 33, 949-59	91
180	Recessive RYR1 mutations in a patient with severe congenital nemaline myopathy with ophthalmoplegia identified through massively parallel sequencing. 2012 , 158A, 772-8	22
179	Congenital myopathies: an update. 2012 , 12, 165-74	94
178	Swimming into prominence: the zebrafish as a valuable tool for studying human myopathies and muscular dystrophies. 2013 , 280, 4187-97	50
177	Congenital (Structural) Myopathies. 2013 , 1-51	
176	A novel late-onset axial myopathy associated with mutations in the skeletal muscle ryanodine receptor (RYR1) gene. 2013 , 260, 1504-10	54
175	RyR1 deficiency in congenital myopathies disrupts excitation-contraction coupling. 2013 , 34, 986-96	33
174	Congenital myopathy with focal loss of cross-striations revisited. 2013 , 23, 160-4	1
173	Large duplication in MTM1 associated with myotubular myopathy. 2013 , 23, 214-8	8
172	198th ENMC International Workshop: 7th Workshop on Centronuclear (Myotubular) myopathies, 31st May - 2nd June 2013, Naarden, The Netherlands. 2013 , 23, 1033-43	11
171	Mutations in RYR1 are a common cause of exertional myalgia and rhabdomyolysis. 2013 , 23, 540-8	126
170	JP-45/JSRP1 variants affect skeletal muscle excitation-contraction coupling by decreasing the sensitivity of the dihydropyridine receptor. 2013 , 34, 184-90	8
169	Centronuclear myopathy related to dynamin 2 mutations: clinical, morphological, muscle imaging and genetic features of an Italian cohort. 2013 , 23, 229-38	45
168	Novel excitation-contraction uncoupled RYR1 mutations in patients with central core disease. 2013 , 23, 120-32	19
167	Adult centronuclear myopathies: A hospital-based study. 2013 , 169, 625-31	7
166	Congenital myopathies. 2013 , 113, 1321-36	49
165	Congenital myopathies--clinical features and frequency of individual subtypes diagnosed over a 5-year period in the United Kingdom. 2013 , 23, 195-205	91
164	Core myopathies and malignant hyperthermia susceptibility: a review. 2013 , 23, 834-41	37

163	Genotype-phenotype correlations in recessive RYR1-related myopathies. 2013 , 8, 117	75
162	Recessive truncating titin gene, TTN, mutations presenting as centronuclear myopathy. 2013 , 81, 1205-14	142
161	Expanding the MTM1 mutational spectrum: novel variants including the first multi-exonic duplication and development of a locus-specific database. 2013 , 21, 540-9	25
160	Severe congenital RYR1-associated myopathy: the expanding clinicopathologic and genetic spectrum. 2013 , 80, 1584-9	68
159	An integrated diagnosis strategy for congenital myopathies. 2013 , 8, e67527	42
158	Pathogenic mechanisms in centronuclear myopathies. 2014 , 6, 339	76
157	Clinical and Pathological Features of Korean Patients with DNM2-Related Centronuclear Myopathy. 2014 , 10, 24-31	8
156	Germline mutations in RYR1 are associated with foetal akinesia deformation sequence/lethal multiple pterygium syndrome. <i>Acta Neuropathologica Communications</i> , 2014 , 2, 148	7-3 18
155	Congenital Myopathies. 2014 , 195-209	
154	Bridging integrator 1 (Bin1) deficiency in zebrafish results in centronuclear myopathy. 2014 , 23, 3566-78	22
153	Respiratory assessment in centronuclear myopathies. 2014 , 50, 315-26	9
152	Congenital myopathies with secondary neuromuscular transmission defects; a case report and review of the literature. 2014 , 24, 1103-10	31
151	Triadopathies: an emerging class of skeletal muscle diseases. 2014 , 11, 773-85	49
150	Ryanodine myopathies without central cores--clinical, histopathologic, and genetic description of three cases. 2014 , 51, 275-8	8
149	Approach to the diagnosis of congenital myopathies. 2014 , 24, 97-116	178
148	RYR1-related congenital myopathy with fatigable weakness, responding to pyridostigimine. 2014 , 24, 707-12	31
147	Rhabdomyolysis: a genetic perspective. 2015 , 10, 51	73
146	RYR1-related myopathies: a wide spectrum of phenotypes throughout life. 2015 , 22, 1094-112	82

145	The kalaemic and neuromuscular effects of succinylcholine in centronuclear myopathy: A pilot investigation in a canine model. 2015 , 32, 666-71		1
144	Frequency and Phenotype of Myotubular Myopathy Amongst Danish Patients with Congenital Myopathy Older than 5 Years. 2015 , 2, 167-174		6
143	DNM2 mutations in a cohort of sporadic patients with centronuclear myopathy. 2015 , 38, 147-51		7
142	Epigenetic changes as a common trigger of muscle weakness in congenital myopathies. 2015 , 24, 4636-47		33
141	Pathophysiological concepts in the congenital myopathies: blurring the boundaries, sharpening the focus. 2015 , 138, 246-68		55
140	X-linked myotubular myopathy in Rottweiler dogs is caused by a missense mutation in Exon 11 of the MTM1 gene. 2015 , 5, 1		20
139	A novel missense mutation of RYR1 in familial idiopathic hyper CK-emia. 2015 , 356, 142-7		6
138	The Congenital Myopathies. 2015 , 1121-1129		1
137	Phospholamban overexpression in mice causes a centronuclear myopathy-like phenotype. <i>DMM Disease Models and Mechanisms</i> , 2015 , 8, 999-1009	4.1	21
136	Characterization of excitation-contraction coupling components in human extraocular muscles. 2015 , 466, 29-36		9
135	A diagnostic dilemma in a family with cystinuria type B resolved by muscle magnetic resonance. 2015 , 52, 548-51		3
134	Congenital and Other Structural Myopathies. 2015 , 499-537		2
133	Centronuclear myopathies: genotype-phenotype correlation and frequency of defined genetic forms in an Italian cohort. 2015 , 262, 1728-40		42
132	The neuromuscular differential diagnosis of joint hypermobility. 2015 , 169C, 23-42		19
131	Moving and positioning the nucleus in skeletal muscle - one step at a time. 2015 , 6, 373-81		56
130	Ryanodine Receptor Channelopathies in Skeletal and Cardiac Muscle. 2016 , 53-84		1
129	Severely Atrophic Human Muscle Fibers With Nuclear Misplacement Survive Many Years of Permanent Denervation. 2016 , 26, 5894		27
128	Excitation-Contraction Coupling Alterations in Myopathies. 2016 , 3, 443-453		13

127	Increasing Role of Titin Mutations in Neuromuscular Disorders. 2016 , 3, 293-308		75
126	Cellular, biochemical and molecular changes in muscles from patients with X-linked myotubular myopathy due to MTM1 mutations. 2017 , 26, 320-332		14
125	Congenital myopathies: not only a paediatric topic. 2016 , 29, 642-50		30
124	Review of RyR1 pathway and associated pathomechanisms. <i>Acta Neuropathologica Communications</i> , 2016 , 4, 121	7-3	34
123	Functional characterization of orbicularis oculi and extraocular muscles. 2016 , 147, 395-406		7
122	RYR1-related rhabdomyolysis: A common but probably underdiagnosed manifestation of skeletal muscle ryanodine receptor dysfunction. 2016 , 172, 546-558		21
121	Intra-familial variability associated with recessive RYR1 mutation diagnosed prenatally by exome sequencing. 2016 , 36, 1020-1026		11
120	Prenatal diagnosis of congenital myopathies and muscular dystrophies. 2016 , 90, 199-210		15
119	217th ENMC International Workshop: RYR1-related myopathies, Naarden, The Netherlands, 29-31 January 2016. 2016 , 26, 624-33		28
118	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. 2016 , 26, 292-9		20
117	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, 347-59	4-1	23
116	Gene Discovery in Congenital Myopathy. 2016 , 39-83		
115	219th ENMC International Workshop Titinopathies International database of titin mutations and phenotypes, Heemskerk, The Netherlands, 29 April-1 May 2016. 2017 , 27, 396-407		23
114	Myopathology in times of modern imaging. 2017 , 43, 24-43		23
113	Resequencing array for gene variant detection in malignant hyperthermia and butyrylcholinesterase deficiency. 2017 , 27, 492-499		2
112	RYR1-Related Myopathies: Clinical, Histopathologic and Genetic Heterogeneity Among 17 Patients from a Portuguese Tertiary Centre. 2017 , 4, 67-76		8
111	Atrophy, ultra-structural disorders, severe atrophy and degeneration of denervated human muscle in SCI and Aging. Implications for their recovery by Functional Electrical Stimulation, updated 2017. 2017 , 39, 660-666		40
110	Common and variable clinical, histological, and imaging findings of recessive RYR1-related centronuclear myopathy patients. 2017 , 27, 975-985		24

109	A chemical chaperone improves muscle function in mice with a RyR1 mutation. 2017 , 8, 14659	40
108	Myopathology in congenital myopathies. 2017 , 43, 5-23	41
107	Effect of local cooling on excitation-contraction coupling in myasthenic muscle: Another mechanism of ice-pack test in myasthenia gravis. 2017 , 128, 2309-2317	8
106	RYR1 causing distal myopathy. 2017 , 5, 800-804	11
105	Affected female carriers of MTM1 mutations display a wide spectrum of clinical and pathological involvement: delineating diagnostic clues. 2017 , 134, 889-904	30
104	A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome. 2017 , 8, 16077	51
103	New era in genetics of early-onset muscle disease: Breakthroughs and challenges. 2017 , 64, 160-170	20
102	Current and future therapeutic approaches to the congenital myopathies. 2017 , 64, 191-200	26
101	Congenital myopathies: clinical phenotypes and new diagnostic tools. 2017 , 43, 101	43
100	Parental mosaicism in RYR1-related Central Core Disease. 2018 , 28, 422-426	3
99	Characterization of congenital myopathies at a Korean neuromuscular center. 2018 , 58, 235-244	5
98	Congenital myopathies: disorders of excitation-contraction coupling and muscle contraction. 2018 , 14, 151-167	133
97	Congenital Myopathies. 2018 , 173-182	1
96	Atypical periodic paralysis and myalgia: A novel phenotype. 2018 , 90, e412-e418	27
95	Whole exome sequencing discloses a pathogenic MTM1 gene mutation and ends the diagnostic odyssey in an older woman with a progressive and seemingly sporadic myopathy: Case report and literature review of MTM1 manifesting female carriers. 2018 , 28, 339-345	2
94	Malignant Hyperthermia Susceptibility and Related Diseases. 2018 , 128, 159-167	56
93	Compound heterozygous RYR1 mutations in a preterm with arthrogryposis multiplex congenita and prenatal CNS bleeding. 2018 , 28, 54-58	4
92	Hereditary Myopathies. 2018 ,	2

91	Ryanodine Receptor 1-Related Myopathies: Diagnostic and Therapeutic Approaches. 2018 , 15, 885-899		43
90	Dynamin 2 (DNM2) as Cause of, and Modifier for, Human Neuromuscular Disease. 2018 , 15, 966-975		24
89	Characterization and genetic diagnosis of centronuclear myopathies in seven Chinese patients. 2018 , 39, 2043-2051		1
88	A Rare Case of Severe Congenital RYR1-Associated Myopathy. 2018 , 2018, 6184185		5
87	Correlation of phenotype with genotype and protein structure in RYR1-related disorders. 2018 , 265, 2506-2524		13
86	Centronuclear myopathy: advances in genetic understanding and potential for future treatments. 2018 , 6, 375-384		
85	Muscular MRI-based algorithm to differentiate inherited myopathies presenting with spinal rigidity. 2018 , 28, 5293-5303		19
84	New variant of necklace fibres display peculiar lysosomal structures and mitophagy. 2018 , 28, 846-856		0
83	An integrated modelling methodology for estimating the prevalence of centronuclear myopathy. 2018 , 28, 766-777		15
82	Ryanodine receptor dysfunction in human disorders. 2018 , 1865, 1687-1697		53
81	Centronuclear myopathies under attack: A plethora of therapeutic targets. 2018 , 5, 387-406		23
80	Novel Variants in Individuals with -Related Congenital Myopathies: Genetic, Laboratory, and Clinical Findings. <i>Frontiers in Neurology</i> , 2018 , 9, 118	4.1	11
79	Clinical Application of Epilepsy Genetics in Africa: Is Now the Time?. <i>Frontiers in Neurology</i> , 2018 , 9, 276	4.1	9
78	The genetics of congenital myopathies. 2018 , 148, 549-564		43
77	Abnormal Excitation-Contraction Coupling and Calcium Homeostasis in Myopathies and Cardiomyopathies. 2019 , 6, 289-305		8
76	Single-Cell Transcriptomics and Proteomics of Skeletal Muscle: Technology and Applications. 2019 , 253-281		
75	Update on the Genetics of Congenital Myopathies. 2019 , 29, 12-22		12
74	Diagnosis of Possible mitochondrial disease: an existential crisis. 2019 , 56, 123-130		27

73	Quantitative reduction of RyR1 protein caused by a single-allele frameshift mutation in RYR1 ex36 impairs the strength of adult skeletal muscle fibres. 2019 , 28, 1872-1884		5
72	Therapeutic Aspects in Congenital Myopathies. 2019 , 29, 71-82		5
71	Mouse model of severe recessive RYR1-related myopathy. 2019 , 28, 3024-3036		13
70	Quantitative RyR1 reduction and loss of calcium sensitivity of RyR1Q1970fsX16+A4329D cause cores and loss of muscle strength. 2019 , 28, 2987-2999		13
69	Panel-Based Nuclear and Mitochondrial Next-Generation Sequencing Outcomes of an Ethnically Diverse Pediatric Patient Cohort with Mitochondrial Disease. 2019 , 21, 503-513		9
68	Myopathology of Congenital Myopathies: Bridging the Old and the New. 2019 , 29, 55-70		6
67	The histopathological spectrum of malignant hyperthermia and rhabdomyolysis due to RYR1 mutations. 2019 , 266, 876-887		10
66	Reliability and Validity of Self-Report Questionnaires as Indicators of Fatigue in RYR1-Related Disorders. 2019 , 6, 133-141		2
65	Interactions among ryanodine receptor isoforms contribute to muscle fiber type development and function. <i>DMM Disease Models and Mechanisms</i> , 2019 , 13,	4.1	3
64	Cored in the act: the use of models to understand core myopathies. <i>DMM Disease Models and Mechanisms</i> , 2019 , 12,	4.1	11
63	Homozygous/compound heterozygote RYR1 gene variants: Expanding the clinical spectrum. 2019 , 179, 386-396		10
62	Quasi core disease (DuCD): expanding morphological spectrum of RYR1 recessive myopathies. <i>Acta Neuropathologica Communications</i> , 2019 , 7, 3	7.3	15
61	Signs and Symptoms in Congenital Myopathies. 2019 , 29, 3-11		2
60	Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for the Use of Potent Volatile Anesthetic Agents and Succinylcholine in the Context of RYR1 or CACNA1S Genotypes. 2019 , 105, 1338-1344		35
59	Congenital Myopathies and Related Diseases. 2019 , 201-216		
58	Bi-allelic expression of the RyR1 p.A4329D mutation decreases muscle strength in slow-twitch muscles in mice. 2020 , 295, 10331-10339		1
57	MyoSight-semi-automated image analysis of skeletal muscle cross sections. 2020 , 10, 33		4
56	Ryanodine receptor 1-related disorders: an historical perspective and proposal for a unified nomenclature. 2020 , 10, 32		12

55	X-linked myotubular myopathy mimics hereditary spastic paraplegia in two female manifesting carriers of pathogenic MTM1 variant. 2020 , 63, 104040		
54	The intragenic microRNA in the dynamin 2 gene contributes to the pathology of X-linked centronuclear myopathy. 2020 , 295, 8656-8667		7
53	Update on Congenital Myopathies in Adulthood. 2020 , 21,		2
52	Myostatin: a Circulating Biomarker Correlating with Disease in Myotubular Myopathy Mice and Patients. 2020 , 17, 1178-1189		6
51	RyR1-targeted drug discovery pipeline integrating FRET-based high-throughput screening and human myofiber dynamic Ca assays. 2020 , 10, 1791		14
50	Consequences of mutations in the genes of the ER export machinery COPII in vertebrates. 2020 , 25, 199-209		2
49	New Compound Heterozygous Splice Site Mutations of the Skeletal Muscle Ryanodine Receptor () Gene Manifest Fetal Akinesia: A Linkage with Congenital Myopathies. 2020 , 11, 104-109		
48	Congenital Myopathies and Related Disorders. 2021 , 312-360		
47	Cardiac RyR N-terminal region biosensors for FRET-based high-throughput screening.		
46	Genetics of the Congenital Myopathies. 2, 1-9		
45	Striated Preferentially Expressed Protein Kinase (SPEG) in Muscle Development, Function, and Disease. 2021 , 22,		0
44	Ion Channel Gene Mutations Causing Skeletal Muscle Disorders: Pathomechanisms and Opportunities for Therapy. 2021 , 10,		4
43	Early Findings in Neonatal Cases of -Related Congenital Myopathies. <i>Frontiers in Neurology</i> , 2021 , 12, 664618	4.1	0
42	Ryanodine Receptor 1-Related Myopathies: Quantification of Intramuscular Fatty Infiltration from T1-Weighted MRI. 2021 , 8, 657-668		0
41	Marked Facial Weakness, Ptosis, and Hanging Jaw: A Case with RYR1-Related Congenital Centronuclear Myopathy.		
40	Molecular and cellular basis of genetically inherited skeletal muscle disorders. 2021 , 22, 713-732		11
39	A recurrent RYR1 mutation associated with early-onset hypotonia and benign disease course. <i>Acta Neuropathologica Communications</i> , 2021 , 9, 155	7.3	1
38	Clinical, genetic, and histological features of centronuclear myopathy in the Netherlands. 2021 , 100, 692-702		1

37	Mutational and clinical spectrum of centronuclear myopathy in 9 cases and a literature review of Chinese patients. 2021 , 1		
36	A review of core myopathy: central core disease, multimimicore disease, dusty core disease, and core-rod myopathy. 2021 , 31, 968-977		2
35	Skeletal Muscle Biopsy Evaluation. 2020 , 3-48		1
34	Ryanodine Receptor 1 and Associated Pathologies. 2014 , 167-187		1
33	SPEG binds with desmin and its deficiency causes defects in triad and focal adhesion proteins. 2021 , 29, 3882-3891		1
32	Severe Neonatal RYR1 Myopathy With Pathological Features of Congenital Muscular Dystrophy. 2019 , 78, 283-287		3
31	Variable myopathic presentation in a single family with novel skeletal RYR1 mutation. 2013 , 8, e69296		11
30	Sarcoplipin deletion exacerbates soleus muscle atrophy and weakness in phospholamban overexpressing mice. 2017 , 12, e0173708		16
29	Potassium dependent rescue of a myopathy with core-like structures in mouse. <i>ELife</i> , 2015 , 4,	8.9	7
28	Common Pathogenic Mechanisms in Centronuclear and Myotubular Myopathies and Latest Treatment Advances. 2021 , 22,		1
27	Whole-exome sequencing identifies biosignatures that predict adverse survival outcomes in surgically treated patients with oral cavity squamous cell carcinoma. 2021 , 122, 105547		1
26	Congenital Myopathies and Related Disorders. 2013 , 358-405		1
25	Core Myopathies, Malignant Hyperthermia Susceptibility, and Brody Disease. 214-224		
24	Centronuclear Myopathies. 134-144		0
23	Aberrant Epigenetic Silencing is Triggered by a Transient Reduction in Gene Expression. 2013 , 197-224		
22	SH3KBP1 scaffolds endoplasmic reticulum and controls skeletal myofibers architecture and integrity.		
21	The congenital myopathies. 2020 , 451-461		
20	Dominant or recessive mutations in the gene causing central core myopathy in Brazilian patients. 2020 , 39, 274-282		0

19	Improvement of muscle strength in a mouse model for congenital myopathy treated with HDAC and DNA methyltransferase inhibitors.		
18	Cardiac ryanodine receptor N-terminal region biosensors identify novel inhibitors via FRET-based high-throughput screening. 2021 , 101412		1
17	BIN1 modulation in vivo rescues dynamin-related myopathy.. 2022 , 119,		1
16	Improvement of muscle strength in a mouse model for congenital myopathy treated with HDAC and DNA methyltransferase inhibitors.. <i>ELife</i> , 2022 , 11,	8.9	0
15	Characterization of a novel zebrafish model of SPEG-related centronuclear myopathy.. <i>DMM Disease Models and Mechanisms</i> , 2022 ,	4.1	1
14	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	
13	A novel zebrafish model of SPEG-related centronuclear myopathy (CNM): characterization and comparison with other CNM model zebrafish.		
12	Expanding the clinical-pathological and genetic spectrum of RYR1-related congenital myopathies with cores and minicores: an Italian population study.. <i>Acta Neuropathologica Communications</i> , 2022 , 10, 54	7.3	1
11	presentation_1.PDF. 2018 ,		
10	Correlation of PhenotypeGenotype and Protein Structure in RYR1-Related Myopathy. <i>Frontiers in Neurology</i> , 13,	4.1	0
9	A review of major causative genes in congenital myopathies. <i>Journal of Human Genetics</i> ,	4.3	0
8	Mutations in proteins involved in E-C coupling and SOCE and congenital myopathies. 2022 , 154,		0
7	Phenotypic Spectrum ofDNM2-Related Centronuclear Myopathy. 2022 , 8, e200027		0
6	A case for genomic medicine in South African paediatric patients with neuromuscular disease. 10,		0
5	RYR-1-Related Diseases International Research Workshop: From Mechanisms to Treatments Pittsburgh, PA, U.S.A., 21-22 July 2022. 2022 , 1-20		0
4	A Large-Scale High-Throughput Screen for Modulators of SERCA Activity. 2022 , 12, 1789		0
3	Skeletal and cardiac muscle calcium transport regulation in health and disease. 2022 , 42,		2
2	Using Cluster Analysis to Overcome the Limits of Traditional PhenotypeGenotype Correlations: The Example of RYR1-Related Myopathies. 2023 , 14, 298		1

1 Titin Related Myopathy with Ophthalmoplegia. A Novel Phenotype.. **2023**,

o