

Approach to the diagnosis of congenital myopathies

Neuromuscular Disorders

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Expanding the Phenotype Associated With the <i>NEFL</i> Mutation. <i>JAMA Neurology</i> , 2014, 71, 1413.	4.5	30
2	Congenital (Structural) Myopathies. , 2014, , .		2
3	Loss of Tropomodulin4 in the zebrafish mutant <i>trAge</i> causes cytoplasmic rod formation and muscle weakness reminiscent of nemaline myopathy. <i>DMM Disease Models and Mechanisms</i> , 2014, 7, 1407-15.	1.2	32
4	Kelch proteins: emerging roles in skeletal muscle development and diseases. <i>Skeletal Muscle</i> , 2014, 4, 11.	1.9	119
5	Congenital myopathies: Characteristics and subtypes in Hong Kong. <i>Neuromuscular Disorders</i> , 2015, 25, S278.	0.3	1
6	Expanding genotype/phenotype of neuromuscular diseases by comprehensive target capture/NGS. <i>Neurology: Genetics</i> , 2015, 1, e14.	0.9	48
7	<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
8	Feeding and Swallowing Disorders in Pediatric Neuromuscular Diseases: An Overview. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, 357-369.	1.1	58
9	Skeletal Muscle MicroRNA and Messenger RNA Profiling in Cofilin-2 Deficient Mice Reveals Cell Cycle Dysregulation Hindering Muscle Regeneration. <i>PLoS ONE</i> , 2015, 10, e0123829.	1.1	9
10	Neonatal Hypotonia. <i>Clinics in Perinatology</i> , 2015, 42, 363-371.	0.8	27
11	A diagnostic approach to recurrent myalgia and rhabdomyolysis in children. <i>Archives of Disease in Childhood</i> , 2015, 100, 793-797.	1.0	29
12	Impaired tropomyosin-troponin interactions reduce activation of the actin thin filament. <i>Biochimica Et Biophysica Acta - Proteins and Proteomics</i> , 2015, 1854, 381-390.	1.1	12
13	Congenital myopathies. <i>Neurology</i> , 2015, 84, 28-35.	1.5	106
14	X-linked myotubular myopathy in Rottweiler dogs is caused by a missense mutation in Exon 11 of the <i>MTM1</i> gene. <i>Skeletal Muscle</i> , 2015, 5, 1.	1.9	46
15	The Congenital Myopathies. , 2015, , 1121-1129.		2
16	Congenital myopathies. <i>Neurology</i> , 2015, 84, 15-16.	1.5	0
18	Congenital and Other Structural Myopathies. , 2015, , 499-537.		3
19	Centronuclear myopathies: genotype-phenotype correlation and frequency of defined genetic forms in an Italian cohort. <i>Journal of Neurology</i> , 2015, 262, 1728-1740.	1.8	51

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20	Bilateral foot-drop as predominant symptom in nebulin (NEB) gene related "core-rod" congenital myopathy. <i>European Journal of Medical Genetics</i> , 2015, 58, 556-561.	0.7	12
21	Evaluation of the floppy infant. <i>Paediatrics and Child Health (United Kingdom)</i> , 2015, 25, 498-504.	0.2	5
22	Increasing Role of Titin Mutations in Neuromuscular Disorders. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 293-308.	1.1	120
23	Aerobic Training in Patients with Congenital Myopathy. <i>PLoS ONE</i> , 2016, 11, e0146036.	1.1	17
24	<sc><i>ADSSL</i></sc><i>1</i> mutation relevant to autosomal recessive adolescent onset distal myopathy. <i>Annals of Neurology</i> , 2016, 79, 231-243.	2.8	32
25	Clinical features and therapeutic strategies for managing the striated muscle laminopathies. <i>Expert Opinion on Orphan Drugs</i> , 2016, 4, 631-638.	0.5	1
28	Congenital myopathies: not only a paediatric topic. <i>Current Opinion in Neurology</i> , 2016, 29, 642-650.	1.8	37
29	Muscle magnetic resonance imaging in congenital myasthenic syndromes. <i>Muscle and Nerve</i> , 2016, 54, 211-219.	1.0	24
30	Muscle dysfunction caused by loss of <i>Magel2</i> in a mouse model of Prader-Willi and Schaaf-Yang syndromes. <i>Human Molecular Genetics</i> , 2016, 25, 3798-3809.	1.4	38
32	Prenatal diagnosis of congenital myopathies and muscular dystrophies. <i>Clinical Genetics</i> , 2016, 90, 199-210.	1.0	20
33	Anesthetic management of 877 pediatric patients undergoing muscle biopsy for neuromuscular disorders: a 20-year review. <i>Paediatric Anaesthesia</i> , 2016, 26, 710-721.	0.6	97
34	The genetic basis of undiagnosed muscular dystrophies and myopathies. <i>Neurology</i> , 2016, 87, 71-76.	1.5	92
36	KLHL40-related nemaline myopathy with a sustained, positive response to treatment with acetylcholinesterase inhibitors. <i>Journal of Neurology</i> , 2016, 263, 517-523.	1.8	30
37	Pediatric laminopathies: Whole-body magnetic resonance imaging fingerprint and comparison with <i>Sepn1</i> myopathy. <i>Muscle and Nerve</i> , 2016, 54, 192-202.	1.0	31
38	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. <i>Neuromuscular Disorders</i> , 2016, 26, 292-299.	0.3	25
39	New massive parallel sequencing approach improves the genetic characterization of congenital myopathies. <i>Journal of Human Genetics</i> , 2016, 61, 497-505.	1.1	15
40	The transcription coactivator ASC-1 is a regulator of skeletal myogenesis, and its deficiency causes a novel form of congenital muscle disease. <i>Human Molecular Genetics</i> , 2016, 25, 1559-1573.	1.4	25
41	Gene Discovery in Congenital Myopathy. <i>Pancreatic Islet Biology</i> , 2016, , 39-83.	0.1	0

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42	Next Generation Sequencing in Neurology and Psychiatry. , 2016, , 97-136.		0
43	Myo18b is essential for sarcomere assembly in fast skeletal muscle. Human Molecular Genetics, 2017, 26, ddx025.	1.4	24
44	Myopathology in times of modern imaging. Neuropathology and Applied Neurobiology, 2017, 43, 24-43.	1.8	34
45	Muscle MRI in pediatrics: clinical, pathological and genetic correlation. Pediatric Radiology, 2017, 47, 724-735.	1.1	12
46	Central core myopathy with autophagy. Muscle and Nerve, 2017, 56, E8-E9.	1.0	2
47	Targeted massively parallel sequencing and histological assessment of skeletal muscles for the molecular diagnosis of inherited muscle disorders. Journal of Medical Genetics, 2017, 54, 104-110.	1.5	51
48	Mutations in INPP5K , Encoding a Phosphoinositide 5-Phosphatase, Cause Congenital Muscular Dystrophy with Cataracts and Mild Cognitive Impairment. American Journal of Human Genetics, 2017, 100, 523-536.	2.6	67
49	Distal myopathy with ADSSL1 mutations in Korean patients. Neuromuscular Disorders, 2017, 27, 465-472.	0.3	15
50	Whole exome sequencing of a patient with suspected mitochondrial myopathy reveals novel compound heterozygous variants in <i>RYR1</i> . Molecular Genetics & Genomic Medicine, 2017, 5, 295-302.	0.6	6
51	Establishing prevalence in rare neuromuscular diseases. Neurology: Genetics, 2017, 3, e146.	0.9	1
52	The Applications and Challenges of Next-Generation Sequencing in Diagnosing Neuromuscular Disorders. , 2017, , 177-200.		2
53	Phenotypes, genotypes, and prevalence of congenital myopathies older than 5 years in Denmark. Neurology: Genetics, 2017, 3, e140.	0.9	34
54	Inherited Neuromuscular Disorders: Presentation, Diagnosis, and Advances in Treatment. Current Pediatrics Reports, 2017, 5, 36-44.	1.7	0
55	Advances in neuromuscular disorders – an update. Paediatrics and Child Health (United Kingdom), 2017, 27, 271-275.	0.2	2
56	Myopathology in congenital myopathies. Neuropathology and Applied Neurobiology, 2017, 43, 5-23.	1.8	54
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58	Novel intronic mutation in <i>MTM1</i> detected by RNA analysis in a case of X-linked myotubular myopathy. Neurology: Genetics, 2017, 3, e182.	0.9	9
59	X-linked myotubular myopathy. Neurology, 2017, 89, 1316-1317.	1.5	1

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61	Cost-effectiveness of massively parallel sequencing for diagnosis of paediatric muscle diseases. <i>Npj Genomic Medicine</i> , 2017, 2, .	1.7	67
62	The Role of Muscle Imaging in the Diagnosis and Assessment of Children with Genetic Muscle Disease. <i>Neuropediatrics</i> , 2017, 48, 233-241.	0.3	11
63	Congenital Muscular Dystrophies and Myopathies: An Overview and Update. <i>Neuropediatrics</i> , 2017, 48, 247-261.	0.3	40
64	A recessive mutation in beta-IV-spectrin (SPTBN4) associates with congenital myopathy, neuropathy, and central deafness. <i>Human Genetics</i> , 2017, 136, 903-910.	1.8	51
65	Recessive mutations in the kinase ZAK cause a congenital myopathy with fibre type disproportion. <i>Brain</i> , 2017, 140, 37-48.	3.7	28
66	Current and future therapeutic approaches to the congenital myopathies. <i>Seminars in Cell and Developmental Biology</i> , 2017, 64, 191-200.	2.3	29
67	Muscle redox disturbances and oxidative stress as pathomechanisms and therapeutic targets in early-onset myopathies. <i>Seminars in Cell and Developmental Biology</i> , 2017, 64, 213-223.	2.3	51
68	Muscular dystrophies and myopathies: the spectrum of mutated genes in the Czech Republic. <i>Clinical Genetics</i> , 2017, 91, 463-469.	1.0	32
69	Discovery of pathogenic variants in a large Korean cohort of inherited muscular disorders. <i>Clinical Genetics</i> , 2017, 91, 403-410.	1.0	24
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71	Congenital Myopathies. , 2017, , 1123-1130.		0
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73	The relative frequency of common neuromuscular diagnoses in a reference center. <i>Arquivos De Neuro-Psiquiatria</i> , 2017, 75, 789-795.	0.3	8
74	The Floppy Infant. , 2017, , 1051-1056.		0
75	SPEG-deficient skeletal muscles exhibit abnormal triad and defective calcium handling. <i>Human Molecular Genetics</i> , 2018, 27, 1608-1617.	1.4	22
76	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. <i>Annals of Neurology</i> , 2018, 83, 1105-1124.	2.8	93
77	Characterization of congenital myopathies at a Korean neuromuscular center. <i>Muscle and Nerve</i> , 2018, 58, 235-244.	1.0	8

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78	Congenital myopathies: disorders of excitation-contraction coupling and muscle contraction. <i>Nature Reviews Neurology</i> , 2018, 14, 151-167.	4.9	212
79	Genetic and functional analysis of the RYR1 mutation p.Thr84Met revealed a susceptibility to malignant hyperthermia. <i>Journal of Anesthesia</i> , 2018, 32, 174-181.	0.7	7
80	In Vivo Function of the Chaperonin TRiC in α -Actin Folding during Sarcomere Assembly. <i>Cell Reports</i> , 2018, 22, 313-322.	2.9	29
82	Inositol trisphosphate receptor-mediated Ca ²⁺ signalling stimulates mitochondrial function and gene expression in core myopathy patients. <i>Human Molecular Genetics</i> , 2018, 27, 2367-2382.	1.4	14
83	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. <i>Neuromuscular Disorders</i> , 2018, 28, 339-345.	0.3	4
84	Treating pediatric neuromuscular disorders: The future is now. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 804-841.	0.7	93
85	Congenital fiber-type disproportion in an ambulatory rehabilitation setting. <i>Wiener Medizinische Wochenschrift</i> , 2018, 168, 367-373.	0.5	0
86	Hypotonia, Weakness, and Stroke. , 2018, , 455-507.e4.		0
87	Ryanodine Receptor 1-Related Myopathies: Diagnostic and Therapeutic Approaches. <i>Neurotherapeutics</i> , 2018, 15, 885-899.	2.1	81
88	A novel SPEG mutation causes non-compaction cardiomyopathy and neuropathy in a floppy infant with centronuclear myopathy. <i>Acta Neuropathologica Communications</i> , 2018, 6, 83.	2.4	17
90	Neuromuscular Disorders in Newborns. , 2018, , 2323-2335.		0
91	Clinical and Pathologic Findings of Korean Patients with RYR1-Related Congenital Myopathy. <i>Journal</i>		

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98	Novel Variants in Individuals with RYR1-Related Congenital Myopathies: Genetic, Laboratory, and Clinical Findings. <i>Frontiers in Neurology</i> , 2018, 9, 118.	1.1	11
99	Knockout of myomaker results in defective myoblast fusion, reduced muscle growth and increased adipocyte infiltration in zebrafish skeletal muscle. <i>Human Molecular Genetics</i> , 2018, 27, 3542-3554.	1.4	25
100	Muscle Involvement and Restricted Disorders. , 2018, , 922-970.e15.		3
101	The genetics of congenital myopathies. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2018, 148, 549-564.	1.0	68
102	Essentials of Neurology and Neuromuscular Disorders. , 2019, , 561-580.e4.		2
103	214th ENMC International Workshop: Establishing an international consortium for gene discovery and clinical research for Congenital Muscle Disease, Heemskerk, the Netherlands, 6â€“18 October 2015. <i>Neuromuscular Disorders</i> , 2019, 29, 644-650.	0.3	2
104	Neonatal hypotonia and neuromuscular conditions. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2019, 162, 435-448.	1.0	13
105	<i>MYL2</i>-associated congenital fiber-type disproportion and cardiomyopathy with variants in additional neuromuscular disease genes; the dilemma of panel testing. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a004184.	0.5	5
106	Novel <i>TTN</i> mutations and muscle imaging characteristics in congenital titinopathy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1311-1318.	1.7	16
107	Neuromuscular Diseases of the Newborn. <i>Seminars in Pediatric Neurology</i> , 2019, 32, 100771.	1.0	6
108	Congenital myopathy with a novel SELN missense mutation and the challenge to differentiate it from congenital muscular dystrophy. <i>Journal of Clinical Neuroscience</i> , 2019, 62, 238-239.	0.8	4
109	Dihydropyridine Receptor Congenital Myopathy In A Consanguineous Turkish Family. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 377-384.	1.1	12
110	Congenital myopathies in the adult neuromuscular clinic. <i>Neurology: Genetics</i> , 2019, 5, e341.	0.9	22
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112	Role of Nerve and Muscle Biopsies in Pediatric Patients in the Era of Genetic Testing. <i>Journal of Surgical Research</i> , 2019, 243, 27-32.	0.8	5
114	Recessive mutations in proximal I-band of TTN gene cause severe congenital multi-minicore disease without cardiac involvement. <i>Neuromuscular Disorders</i> , 2019, 29, 350-357.	0.3	10
115	Pregnancy and Delivery in Women With Congenital Myopathies. <i>Seminars in Pediatric Neurology</i> , 2019, 29, 23-29.	1.0	5
116	Molecular Basis of Muscle Disease. , 2019, , 13-39.		1

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118	Congenital myopathies are mainly associated with a mild cardiac phenotype. <i>Journal of Neurology</i> , 2019, 266, 1367-1375.	1.8	10
119	Myoimaging in Congenital Myopathies. <i>Seminars in Pediatric Neurology</i> , 2019, 29, 30-43.	1.0	18
120	Common pathogenic mechanism in patients with dropped head syndrome caused by different mutations in the MYH7 gene. <i>Gene</i> , 2019, 697, 159-164.	1.0	4
121	Myopathology of Congenital Myopathies: Bridging the Old and the New. <i>Seminars in Pediatric Neurology</i> , 2019, 29, 55-70.	1.0	8
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124	The neuro-ophthalmology of inherited myopathies. <i>Current Opinion in Ophthalmology</i> , 2019, 30, 476-483.	1.3	4
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126	A pattern-based approach to the interpretation of skeletal muscle biopsies. <i>Modern Pathology</i> , 2019, 32, 462-483.	2.9	14
127	The Use of Muscle Ultrasound in the Diagnosis and Differential Diagnosis of Congenital Disorders of Muscle in the Age of Next Generation Genetics. <i>Seminars in Pediatric Neurology</i> , 2019, 29, 44-54.	1.0	6
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130	Congenital myopathies: an update. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 297-302.	1.1	50
131	Exercise Training as Part of Musculoskeletal Management for Congenital Myopathy: Where Are We Now?. <i>Pediatric Neurology</i> , 2020, 104, 13-18.	1.0	8
132	ASC β Is a Cell Cycle Regulator Associated with Severe and Mild Forms of Myopathy. <i>Annals of Neurology</i> , 2020, 87, 217-232.	2.8	12
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134	Ryanodine receptor 1-related disorders: an historical perspective and proposal for a unified nomenclature. <i>Skeletal Muscle</i> , 2020, 10, 32.	1.9	45

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135	X-Linked Myotubular Myopathy and Duchenne Muscular Dystrophy in a Preterm Infant: A Rare Combination. <i>Pediatrics</i> , 2020, 146, e20182879.	1.0	2
136	Ovine congenital progressive muscular dystrophy (OCPMD) is a model of TNNT1 congenital myopathy. <i>Acta Neuropathologica Communications</i> , 2020, 8, 142.	2.4	4
137	Preclinical model systems of ryanodine receptor 1-related myopathies and malignant hyperthermia: a comprehensive scoping review of works published 1990–2019. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 113.	1.2	19
138	Update on Congenital Myopathies in Adulthood. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3694.	1.8	9
139	A new 1p36.13–p36.12 microdeletion syndrome characterized by learning disability, behavioral abnormalities, and ptosis. <i>Clinical Genetics</i> , 2020, 97, 927-932.	1.0	6
140	Distal Myopathies. <i>Neurologic Clinics</i> , 2020, 38, 637-659.	0.8	4
141	LAMA2 Neuropathies: Human Findings and Pathomechanisms From Mouse Models. <i>Frontiers in Molecular Neuroscience</i> , 2020, 13, 60.	1.4	18
142	The Phenotype and Genotype of Congenital Myopathies Based on a Large Pediatric Cohort. <i>Pediatric Neurology</i> , 2021, 115, 50-65.	1.0	11
143	Whole-exome analyses of congenital muscular dystrophy and congenital myopathy patients from India reveal a wide spectrum of known and novel mutations. <i>European Journal of Neurology</i> , 2021, 28, 992-1003.	1.7	9
144	Muscle biopsy essential diagnostic advice for pathologists. <i>Surgical and Experimental Pathology</i> , 2021, 4, .	0.2	4
145	l-Carnitine ameliorates congenital myopathy in a tropomyosin 3 de novo mutation transgenic zebrafish. <i>Journal of Biomedical Science</i> , 2021, 28, 8.	2.6	8
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147	MR imaging of inherited myopathies: a review and proposal of imaging algorithms. <i>European Radiology</i> , 2021, 31, 8498-8512.	2.3	10
148	Profound Hypotonia and Respiratory Failure due to Suspected Nemaline Myopathy in a Preterm Infant. <i>AJP Reports</i> , 2021, 11, e91-e94.	0.4	1
149	Neuromuscular disease and respiratory failure. , 2021, , 231-244.		0
150	Guidelines for genetic testing of muscle and neuromuscular junction disorders. <i>Muscle and Nerve</i> , 2021, 64, 255-269.	1.0	8
151	Inherited Defects of the ASC-1 Complex in Congenital Neuromuscular Diseases. <i>International Journal of Molecular Sciences</i> , 2021, 22, 6039.	1.8	6
152	Novel TNNT1 mutation and mild nemaline myopathy phenotype in an Italian patient. <i>Neuromuscular Disorders</i> , 2021, 31, 532-538.	0.3	12

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153	A case of dermatomyositis in a patient with central core disease: unusual association with autoimmunity and genetic muscle disease. <i>Pediatric Rheumatology</i> , 2021, 19, 100.	0.9	0
154	Noteworthy Cardiovascular Involvement with Sporadic Late-onset Nemaline Myopathy. <i>Internal Medicine</i> , 2021, 60, 2327-2332.	0.3	1
155	New diagnostic and therapeutic modalities in neuromuscular disorders in children. <i>Current Problems in Pediatric and Adolescent Health Care</i> , 2021, 51, 101033.	0.8	9
156	Nemaline Myopathy: A Case Report. <i>Case Reports in Neurology</i> , 2021, 13, 499-503.	0.3	5
157	Progression or Not – A Small Natural History Study of Genetical Confirmed Congenital Myopathies. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 647-655.	1.1	1
158	Troponin Variants in Congenital Myopathies: How They Affect Skeletal Muscle Mechanics. <i>International Journal of Molecular Sciences</i> , 2021, 22, 9187.	1.8	6
159	A review of core myopathy: central core disease, multiminicore disease, dusty core disease, and core-rod myopathy. <i>Neuromuscular Disorders</i> , 2021, 31, 968-977.	0.3	13
160	Treatment and Management of Spinal Muscular Atrophy and Congenital Myopathies. , 2022, , 261-277.		0
161	Differentiating Moebius syndrome and other congenital facial weakness disorders with electrodiagnostic studies. <i>Muscle and Nerve</i> , 2021, 63, 516-524.	1.0	6
162	Skeletal Muscle and Peripheral Nerves. , 2015, , 767-787.		1
163	An Overview of Congenital Myopathies. <i>CONTINUUM Lifelong Learning in Neurology</i> , 2016, 22, 1932-1953.	0.4	11
164	Congenital Muscular Dystrophy and Congenital Myopathy. <i>CONTINUUM Lifelong Learning in Neurology</i> , 2019, 25, 1640-1661.	0.4	24
165	Early-Onset Myopathies: Clinical Findings, Prevalence of Subgroups and Diagnostic Approach in a Single Neuromuscular Referral Center in Germany. <i>Journal of Neuromuscular Diseases</i> , 2017, 4, 315-325.	1.1	17
166	KLHL40 mutation associated with severe nemaline myopathy, fetal akinesia, and cleft palate. <i>Journal of Pediatric Neurosciences</i> , 2019, 14, 222.	0.2	8
167	Integrated diagnostic approach of pediatric neuromuscular disorders. <i>Journal of Genetic Medicine</i> , 2018, 15, 55-63.	0.1	3
168	Do surgimento à extinção: a trajetória de um serviço ambulatorial de genética médica no Brasil. <i>Revista De Atenção À Saúde</i> , 2021, 19, .	0.0	0
169	Two decades of advances in muscle imaging in children: from pattern recognition of muscle diseases to quantification and machine learning approaches. <i>Neuromuscular Disorders</i> , 2021, 31, 1038-1050.	0.3	2
170	Myocardial and Arrhythmic Spectrum of Neuromuscular Disorders in Children. <i>Biomolecules</i> , 2021, 11, 1578.	1.8	5

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171	Neuromuscular Disorders in Newborns. , 2017, , 1-13.		0
172	Myopathies and Myotonic Disorders. , 2017, , 327-354.		0
173	Muscle Imaging. , 2017, , 119-130.		0
175	Early Diagnosis and Differential Diagnosis of Cerebral Palsy. , 2018, , 89-99.		0
176	Hypotonia at Birth: A Case Study of ACTA-1 Mutation, a Congenital Myopathy. Neonatal Network: NN, 2018, 37, 212-217.	0.1	3
177	A 6-Year-Old Boy with Respiratory and Feeding Difficulties at Birth, Delayed Gross Motor Milestones, and Facial and Proximal Lower Limb Weakness. , 2020, , 289-296.		0
178	A Systematic Review and Meta-Analysis of the Prevalence of Congenital Myopathy. Frontiers in Neurology, 2021, 12, 761636.	1.1	12
179	Muskelerkrankungen. , 2020, , 123-151.		0
180	An 8-year-old boy with delayed motor milestones and proximal leg muscle weakness. , 2020, , 269-274.		0
181	Neonatal Hypotonia. , 2020, , 71-101.		0
182	The congenital myopathies. , 2020, , 451-461.		0
183	Cardiac Complications Associated with Neuromuscular Diseases. , 2020, , 55-93.		0
184	Core myopathies - a short review. Acta Myologica, 2020, 39, 266-273.	1.5	1
185	Phenotypic Variability of MEGF10 Variants Causing Congenital Myopathy: Report of Two Unrelated Patients from a Highly Consanguineous Population. Genes, 2021, 12, 1783.	1.0	3
186	Myocardial strain assessment by 2D speckle-tracking echocardiography in patients with congenital myopathy. Journal of Cardiovascular Echography, 2021, 31, 214.	0.1	0
188	Clinical and genetic features of infancy-onset congenital myopathies from a Chinese paediatric centre. BMC Pediatrics, 2022, 22, 65.	0.7	3
189	Does specificity of electrodiagnostic test referrals predict for test outcome in children?. Muscle and Nerve, 2022, , .	1.0	1
190	Myopathy with oval inclusions in a domestic shorthair cat. Journal of Feline Medicine and Surgery Open Reports, 2022, 8, 205511692210814.	0.1	0

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