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Congenital myopathies: Natural history of a large pediatric cohort

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#	Paper	IF	Citations
96	Congenital myopathies: Rebuilding the natural history, one gene at a time. <i>Neurology</i> , 2015 , 84, 15-6	6.5	
95	Muscular Dystrophies. 2015 , 469-480		
94	Prognosis of Neurological Diseases. 2015 ,		1
93	Ryanodine Receptor Channelopathies in Skeletal and Cardiac Muscle. 2016 , 53-84		1
92	Clinical course of growth in patients with congenital neuromuscular disease in a single multidisciplinary neuromuscular clinic. <i>Journal of Pediatric Rehabilitation Medicine</i> , 2016 , 9, 13-21	1.4	
91	Letter to the Editor: Posterior spinal instrumented fusion for idiopathic scoliosis in patients with multisystemic neurodegenerative disorder: a report of two cases. <i>Journal of Orthopaedic Surgery</i> , 2016 , 24, 428	1.4	
90	Congenital myopathies: not only a paediatric topic. <i>Current Opinion in Neurology</i> , 2016 , 29, 642-50	7.1	30
89	Noninvasive Assessment of Neuromuscular Disease in Dogs: Use of the 6-minute Walk Test to Assess Submaximal Exercise Tolerance in Dogs with Centronuclear Myopathy. <i>Journal of Veterinary Internal Medicine</i> , 2016 , 30, 808-12	3.1	5
88	An RYR1 mutation associated with malignant hyperthermia is also associated with bleeding abnormalities. <i>Science Signaling</i> , 2016 , 9, ra68	8.8	26
87	Prenatal diagnosis of congenital myopathies and muscular dystrophies. <i>Clinical Genetics</i> , 2016 , 90, 199-210		15
86	KLHL40-related nemaline myopathy with a sustained, positive response to treatment with acetylcholinesterase inhibitors. <i>Journal of Neurology</i> , 2016 , 263, 517-23	5.5	21
85	Mutation-specific effects on thin filament length in thin filament myopathy. <i>Annals of Neurology</i> , 2016 , 79, 959-69	9.4	42
84	Safe and sound? A systematic literature review of seizure detection methods for personal use. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2016 , 36, 4-15	3.2	59
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81	Sarcomere Dysfunction in Nemaline Myopathy. <i>Journal of Neuromuscular Diseases</i> , 2017 , 4, 99-113	5	33
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74	Characterization of congenital myopathies at a Korean neuromuscular center. <i>Muscle and Nerve</i> , 2018 , 58, 235-244	3.4	5
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72	Treating pediatric neuromuscular disorders: The future is now. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 804-841	2.5	49
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70	Recent advances in understanding congenital myopathies. <i>F1000Research</i> , 2018 , 7,	3.6	15
69	Ryanodine Receptor 1-Related Myopathies: Diagnostic and Therapeutic Approaches. <i>Neurotherapeutics</i> , 2018 , 15, 885-899	6.4	43
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67	Correlation of phenotype with genotype and protein structure in RYR1-related disorders. <i>Journal of Neurology</i> , 2018 , 265, 2506-2524	5.5	13
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39	A Cross-Sectional Study of Nemaline Myopathy. <i>Neurology</i> , 2021 , 96, e1425-e1436	6.5	3
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- 3 La grande variabilité phénotypique des mutations du gène RYR1. **2022**, 38, 46-48
- 2 Clinical Manifestation of Nebulin-Associated Nemaline Myopathy. **2023**, 9, e200056
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