

# Adele Damico

## List of Publications by Year in Descending Order

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

141  
papers

5,218  
citations

40  
h-index

65  
g-index

195  
ext. papers

6,191  
ext. citations

3  
avg, IF

4.66  
L-index

#	Paper	IF	Citations
141	Body mass index in type 2 spinal muscular atrophy: a longitudinal study.. <i>European Journal of Pediatrics</i> , <b>2022</b> , 1	4.1	0
140	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> , <b>2022</b> , 10, 54	7.3	1
139	Genetic modifiers of upper limb function in Duchenne muscular dystrophy.. <i>Journal of Neurology</i> , <b>2022</b> , 1	5.5	1
138	Hepatobiliary disease in XLMTM: a common comorbidity with potential impact on treatment strategies. <i>Orphanet Journal of Rare Diseases</i> , <b>2021</b> , 16, 425	4.2	2
137	International retrospective natural history study of -related congenital muscular dystrophy. <i>Brain Communications</i> , <b>2021</b> , 3, fcab075	4.5	2
136	Spinal muscular atrophy: state of the art and new therapeutic strategies. <i>Neurological Sciences</i> , <b>2021</b> , 1	3.5	5
135	Nusinersen in pediatric and adult patients with type III spinal muscular atrophy. <i>Annals of Clinical and Translational Neurology</i> , <b>2021</b> , 8, 1622-1634	5.3	6
134	North Star Ambulatory Assessment changes in ambulant Duchenne boys amenable to skip exons 44, 45, 51, and 53: A 3 year follow up. <i>PLoS ONE</i> , <b>2021</b> , 16, e0253882	3.7	1
133	Circadian Genes as Exploratory Biomarkers in DMD: Results From Both the Mouse Model and Patients. <i>Frontiers in Physiology</i> , <b>2021</b> , 12, 678974	4.6	0
132	Respiratory Trajectories in Type 2 and 3 Spinal Muscular Atrophy in the iSMAC Cohort Study. <i>Neurology</i> , <b>2021</b> , 96, e587-e599	6.5	12
131	Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. <i>Neuromuscular Disorders</i> , <b>2021</b> , 31, 596-602	2.9	9
130	Age-related sensory neuropathy in patients with spinal muscular atrophy type 1. <i>Muscle and Nerve</i> , <b>2021</b> , 64, 599-603	3.4	
129	Growth patterns in children with spinal muscular atrophy. <i>Orphanet Journal of Rare Diseases</i> , <b>2021</b> , 16, 375	4.2	3
128	Long-Term Safety and Usefulness of Mexiletine in a Large Cohort of Patients Affected by Non-dystrophic Myotonias. <i>Frontiers in Neurology</i> , <b>2020</b> , 11, 300	4.1	8
127	Predictive energy equations for spinal muscular atrophy type I children. <i>American Journal of Clinical Nutrition</i> , <b>2020</b> , 111, 983-996	7	4
126	Diagnostic journey in Spinal Muscular Atrophy: Is it still an odyssey?. <i>PLoS ONE</i> , <b>2020</b> , 15, e0230677	3.7	15
125	Cytokine Profile in Striated Muscle Laminopathies: New Promising Biomarkers for Disease Prediction. <i>Cells</i> , <b>2020</b> , 9,	7.9	2

124	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. <i>Annals of Clinical and Translational Neurology</i> , <b>2020</b> , 7, 786-798	5.3	14
123	Duchenne Dilated Cardiomyopathy: Cardiac Management from Prevention to Advanced Cardiovascular Therapies. <i>Journal of Clinical Medicine</i> , <b>2020</b> , 9,	5.1	10
122	A Recurrent Pathogenic Variant of Underlies Autosomal Recessive Congenital Muscular Dystrophy With Cataracts and Intellectual Disability: Evidence for a Founder Effect in Southern Italy. <i>Frontiers in Genetics</i> , <b>2020</b> , 11, 565868	4.5	2
121	Clinical Variability in Spinal Muscular Atrophy Type III. <i>Annals of Neurology</i> , <b>2020</b> , 88, 1109-1117	9.4	14
120	Heart rate reduction strategy using ivabradine in end-stage Duchenne cardiomyopathy. <i>International Journal of Cardiology</i> , <b>2019</b> , 280, 99-103	3.2	9
119	Long-term natural history data in Duchenne muscular dystrophy ambulant patients with mutations amenable to skip exons 44, 45, 51 and 53. <i>PLoS ONE</i> , <b>2019</b> , 14, e0218683	3.7	21
118	X-linked myotubular myopathy: A prospective international natural history study. <i>Neurology</i> , <b>2019</b> , 92, e1852-e1867	6.5	33
117	Clinical-genetic features and peculiar muscle histopathology in infantile DNM1L-related mitochondrial epileptic encephalopathy. <i>Human Mutation</i> , <b>2019</b> , 40, 601-618	4.7	22
116	Samish Nemaline Myopathy Sin 2 Italian siblings harbouring a novel homozygous mutation in Troponin-I gene. <i>Neuromuscular Disorders</i> , <b>2019</b> , 29, 766-770	2.9	8
115	Evaluation of gait in Duchenne Muscular Dystrophy: Relation of 3D gait analysis to clinical assessment. <i>Neuromuscular Disorders</i> , <b>2019</b> , 29, 920-929	2.9	4
114	Development of an academic disease registry for spinal muscular atrophy. <i>Neuromuscular Disorders</i> , <b>2019</b> , 29, 794-799	2.9	17
113	Longitudinal natural history in young boys with Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , <b>2019</b> , 29, 857-862	2.9	12
112	Cardiac and Neuromuscular Features of Patients With LMNA-Related Cardiomyopathy. <i>Annals of Internal Medicine</i> , <b>2019</b> , 171, 458-463	8	22
111	Italian recommendations for diagnosis and management of congenital myasthenic syndromes. <i>Neurological Sciences</i> , <b>2019</b> , 40, 457-468	3.5	16
110	Upper limb function in Duchenne muscular dystrophy: 24 month longitudinal data. <i>PLoS ONE</i> , <b>2018</b> , 13, e0199223	3.7	28
109	An observational study of functional abilities in infants, children, and adults with type 1 SMA. <i>Neurology</i> , <b>2018</b> , 91, e696-e703	6.5	12
108	Implicit learning deficit in children with Duchenne muscular dystrophy: Evidence for a cerebellar cognitive impairment?. <i>PLoS ONE</i> , <b>2018</b> , 13, e0191164	3.7	13
107	Broad phenotypic spectrum and genotype-phenotype correlations in GMPPB-related dystroglycanopathies: an Italian cross-sectional study. <i>Orphanet Journal of Rare Diseases</i> , <b>2018</b> , 13, 170	4.2	21

106	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. <i>Genes</i> , <b>2018</b> , 9,	4.2	4
105	Diagnosis of Duchenne Muscular Dystrophy in Italy in the last decade: Critical issues and areas for improvements. <i>Neuromuscular Disorders</i> , <b>2017</b> , 27, 447-451	2.9	24
104	Muscle MRI in neutral lipid storage disease (NLS). <i>Journal of Neurology</i> , <b>2017</b> , 264, 1334-1342	5.5	11
103	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. <i>European Journal of Paediatric Neurology</i> , <b>2017</b> , 21, 873-883	3.8	4
102	Neutral Lipid Storage Diseases: clinical/genetic features and natural history in a large cohort of Italian patients. <i>Orphanet Journal of Rare Diseases</i> , <b>2017</b> , 12, 90	4.2	34
101	Integrated care of muscular dystrophies in Italy. Part 1. Pharmacological treatment and rehabilitative interventions. <i>Acta Myologica</i> , <b>2017</b> , 36, 19-24	1.6	2
100	A novel AIFM1 mutation expands the phenotype to an infantile motor neuron disease. <i>European Journal of Human Genetics</i> , <b>2016</b> , 24, 463-6	5.3	36
99	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. <i>Neuromuscular Disorders</i> , <b>2016</b> , 26, 292-9	2.9	20
98	Patterns of disease progression in type 2 and 3 SMA: Implications for clinical trials. <i>Neuromuscular Disorders</i> , <b>2016</b> , 26, 126-31	2.9	105
97	Health-related quality of life and functional changes in DMD: A 12-month longitudinal cohort study. <i>Neuromuscular Disorders</i> , <b>2016</b> , 26, 189-96	2.9	20
96	Revised North Star Ambulatory Assessment for Young Boys with Duchenne Muscular Dystrophy. <i>PLoS ONE</i> , <b>2016</b> , 11, e0160195	3.7	27
95	Timed Rise from Floor as a Predictor of Disease Progression in Duchenne Muscular Dystrophy: An Observational Study. <i>PLoS ONE</i> , <b>2016</b> , 11, e0151445	3.7	22
94	New Mutations in NEB Gene Discovered by Targeted Next-Generation Sequencing in Nemaline Myopathy Italian Patients. <i>Journal of Molecular Neuroscience</i> , <b>2016</b> , 59, 351-9	3.3	13
93	Deep RNA profiling identified CLOCK and molecular clock genes as pathophysiological signatures in collagen VI myopathy. <i>Journal of Cell Science</i> , <b>2016</b> , 129, 1671-84	5.3	12
92	Translational approach to address therapy in myotonia permanens due to a new SCN4A mutation. <i>Neurology</i> , <b>2016</b> , 86, 2100-8	6.5	15
91	Histological effects of givinostat in boys with Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , <b>2016</b> , 26, 643-649	2.9	96
90	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 974-983	11	37
89	Benefits of glucocorticoids in non-ambulant boys/men with Duchenne muscular dystrophy: A multicentric longitudinal study using the Performance of Upper Limb test. <i>Neuromuscular Disorders</i> , <b>2015</b> , 25, 749-53	2.9	35

88	Cardiac function in types II and III spinal muscular atrophy: should we change standards of care?. <i>Neuropediatrics</i> , <b>2015</b> , 46, 33-6	1.6	9
87	Functional and Morphological Improvement of Dystrophic Muscle by Interleukin 6 Receptor Blockade. <i>EBioMedicine</i> , <b>2015</b> , 2, 285-93	8.8	47
86	Long term follow-up to evaluate the efficacy of miglustat treatment in Italian patients with Niemann-Pick disease type C. <i>Orphanet Journal of Rare Diseases</i> , <b>2015</b> , 10, 22	4.2	45
85	Muscle imaging in fibrodysplasia ossificans progressiva: The neurologist's perspective. <i>Neuromuscular Disorders</i> , <b>2015</b> , 25, 672-3	2.9	1
84	Centronuclear myopathies: genotype-phenotype correlation and frequency of defined genetic forms in an Italian cohort. <i>Journal of Neurology</i> , <b>2015</b> , 262, 1728-40	5.5	42
83	Histologic muscular history in steroid-treated and untreated patients with Duchenne dystrophy. <i>Neurology</i> , <b>2015</b> , 85, 1886-93	6.5	29
82	Developmental lag of visuospatial attention in Duchenne muscular dystrophy. <i>Research in Developmental Disabilities</i> , <b>2015</b> , 36C, 55-61	2.7	5
81	Spinal muscular atrophy associated with progressive myoclonic epilepsy: A rare condition caused by mutations in <i>ASAH1</i> . <i>Epilepsia</i> , <b>2015</b> , 56, 692-8	6.4	25
80	Old measures and new scores in spinal muscular atrophy patients. <i>Muscle and Nerve</i> , <b>2015</b> , 52, 435-7	3.4	5
79	Early Neurodevelopmental Findings Predict School Age Cognitive Abilities in Duchenne Muscular Dystrophy: A Longitudinal Study. <i>PLoS ONE</i> , <b>2015</b> , 10, e0133214	3.7	16
78	Muscle imaging in patients with tubular aggregate myopathy caused by mutations in <i>STIM1</i> . <i>Neuromuscular Disorders</i> , <b>2015</b> , 25, 898-903	2.9	9
77	Suitability of North Star Ambulatory Assessment in young boys with Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , <b>2015</b> , 25, 14-8	2.9	24
76	Prevalence of congenital muscular dystrophy in Italy: a population study. <i>Neurology</i> , <b>2015</b> , 84, 904-11	6.5	49
75	Effect of mexiletine on transitory depression of compound motor action potential in recessive myotonia congenita. <i>Clinical Neurophysiology</i> , <b>2015</b> , 126, 399-403	4.3	18
74	Childhood onset tubular aggregate myopathy associated with de novo <i>STIM1</i> mutations. <i>Journal of Neurology</i> , <b>2014</b> , 261, 870-6	5.5	41
73	Hammersmith Functional Motor Scale and Motor Function Measure-20 in non ambulant SMA patients. <i>Neuromuscular Disorders</i> , <b>2014</b> , 24, 347-52	2.9	32
72	Persistent pulmonary arterial hypertension in the newborn (PPHN): a frequent manifestation of <i>TMEM70</i> defective patients. <i>Molecular Genetics and Metabolism</i> , <b>2014</b> , 111, 353-359	3.7	28
71	Muscle magnetic resonance imaging and histopathology in <i>ACTA1</i> -related congenital nemaline myopathy. <i>Muscle and Nerve</i> , <b>2014</b> , 50, 1011-6	3.4	11

70	Long term natural history data in ambulant boys with Duchenne muscular dystrophy: 36-month changes. <i>PLoS ONE</i> , <b>2014</b> , 9, e108205	3.7	82
69	LMNA-associated myopathies: the Italian experience in a large cohort of patients. <i>Neurology</i> , <b>2014</b> , 83, 1634-44	6.5	45
68	6 Minute walk test in Duchenne MD patients with different mutations: 12 month changes. <i>PLoS ONE</i> , <b>2014</b> , 9, e83400	3.7	56
67	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , <b>2014</b> , 24, 201-6	2.9	68
66	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. <i>Journal of Clinical Investigation</i> , <b>2014</b> , 124, 4693-708	15.9	118
65	The 6 minute walk test and performance of upper limb in ambulant duchenne muscular dystrophy boys. <i>PLOS Currents</i> , <b>2014</b> , 6,		21
64	Characterization of a rare case of Ullrich congenital muscular dystrophy due to truncating mutations within the COL6A1 gene C-terminal domain: a case report. <i>BMC Medical Genetics</i> , <b>2013</b> , 14, 59	2.1	9
63	Six minute walk test in type III spinal muscular atrophy: a 12month longitudinal study. <i>Neuromuscular Disorders</i> , <b>2013</b> , 23, 624-8	2.9	35
62	Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients. <i>Journal of Inherited Metabolic Disease</i> , <b>2013</b> , 36, 43-53	5.4	62
61	Centronuclear myopathy related to dynamin 2 mutations: clinical, morphological, muscle imaging and genetic features of an Italian cohort. <i>Neuromuscular Disorders</i> , <b>2013</b> , 23, 229-38	2.9	45
60	A new de novo missense mutation in MYH2 expands clinical and genetic findings in hereditary myosin myopathies. <i>Neuromuscular Disorders</i> , <b>2013</b> , 23, 437-40	2.9	15
59	Duchenne muscular dystrophy and epilepsy. <i>Neuromuscular Disorders</i> , <b>2013</b> , 23, 313-5	2.9	42
58	Efficacy of miglustat in Niemann-Pick C disease: a single centre experience. <i>Molecular Genetics and Metabolism</i> , <b>2013</b> , 110, 329-35	3.7	16
57	Natural history of pulmonary function in collagen VI-related myopathies. <i>Brain</i> , <b>2013</b> , 136, 3625-33	11.2	69
56	Riboflavin transporter 3 involvement in infantile Brown-Vialetto-Van Laere disease: two novel mutations. <i>Journal of Medical Genetics</i> , <b>2013</b> , 50, 104-7	5.8	27
55	24 month longitudinal data in ambulant boys with Duchenne muscular dystrophy. <i>PLoS ONE</i> , <b>2013</b> , 8, e52512	3.7	75
54	The use of muscle biopsy in the diagnosis of undefined ataxia with cerebellar atrophy in children. <i>European Journal of Paediatric Neurology</i> , <b>2012</b> , 16, 248-56	3.8	26
53	Rituximab in a childhood-onset idiopathic refractory chronic inflammatory demyelinating polyneuropathy. <i>European Journal of Paediatric Neurology</i> , <b>2012</b> , 16, 301-3	3.8	15

52	Allelic and phenotypic heterogeneity in 49 Italian patients with the muscle form of CPT-II deficiency. <i>Clinical Genetics</i> , <b>2012</b> , 82, 232-9	4	33
51	Attention deficit hyperactivity disorder and cognitive function in Duchenne muscular dystrophy: phenotype-genotype correlation. <i>Journal of Pediatrics</i> , <b>2012</b> , 161, 705-9.e1	3.6	82
50	Fetal acetylcholine receptor inactivation syndrome and maternal myasthenia gravis: a case report. <i>Neuromuscular Disorders</i> , <b>2012</b> , 22, 546-8	2.9	13
49	Respiratory and cardiac function in congenital muscular dystrophies with alpha dystroglycan deficiency. <i>Neuromuscular Disorders</i> , <b>2012</b> , 22, 685-9	2.9	26
48	Increased muscle expression of interleukin-17 in Duchenne muscular dystrophy. <i>Neurology</i> , <b>2012</b> , 78, 1309-14	6.5	34
47	Genetic characterization in symptomatic female DMD carriers: lack of relationship between X-inactivation, transcriptional DMD allele balancing and phenotype. <i>BMC Medical Genetics</i> , <b>2012</b> , 13, 73	2.1	42
46	The empowerment of translational research: lessons from laminopathies. <i>Orphanet Journal of Rare Diseases</i> , <b>2012</b> , 7, 37	4.2	5
45	Exon 45 skipping through U1-snRNA antisense molecules recovers the Dys-nNOS pathway and muscle differentiation in human DMD myoblasts. <i>Molecular Therapy</i> , <b>2012</b> , 20, 2134-42	11.7	37
44	Brown-Vialetto-van Laere and Fazio-Londe overlap syndromes: a clinical, biochemical and genetic study. <i>Neuromuscular Disorders</i> , <b>2012</b> , 22, 1075-82	2.9	32
43	Cardiomyopathy in patients with POMT1-related congenital and limb-girdle muscular dystrophy. <i>European Journal of Human Genetics</i> , <b>2012</b> , 20, 1234-9	5.3	23
42	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. <i>Neurology</i> , <b>2012</b> , 79, 159-62	6.5	62
41	GM1 gangliosidosis and Morquio B disease: an update on genetic alterations and clinical findings. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , <b>2011</b> , 1812, 782-90	6.9	95
40	Mosaic caveolin-3 expression in acquired rippling muscle disease without evidence of myasthenia gravis or acetylcholine receptor autoantibodies. <i>Neuromuscular Disorders</i> , <b>2011</b> , 21, 194-203	2.9	15
39	Low-rate repetitive nerve stimulation protocol in an Italian cohort of patients affected by recessive myotonia congenita. <i>Journal of Clinical Neurophysiology</i> , <b>2011</b> , 28, 39-44	2.2	17
38	Congenital muscular dystrophies: a brief review. <i>Seminars in Pediatric Neurology</i> , <b>2011</b> , 18, 277-88	2.9	68
37	A new form of alpha-dystroglycanopathy associated with severe drug-resistant epilepsy and unusual EEG features. <i>Epileptic Disorders</i> , <b>2011</b> , 13, 259-62	1.9	4
36	Spinal muscular atrophy. <i>Orphanet Journal of Rare Diseases</i> , <b>2011</b> , 6, 71	4.2	223
35	miRNAs as serum biomarkers for Duchenne muscular dystrophy. <i>EMBO Molecular Medicine</i> , <b>2011</b> , 3, 258-65	201	

34	SMN transcript levels in leukocytes of SMA patients determined by absolute real-time PCR. <i>European Journal of Human Genetics</i> , <b>2010</b> , 18, 52-8	5.3	36
33	Salbutamol increases survival motor neuron (SMN) transcript levels in leukocytes of spinal muscular atrophy (SMA) patients: relevance for clinical trial design. <i>Journal of Medical Genetics</i> , <b>2010</b> , 47, 856-8	5.8	54
32	Inheritance patterns and phenotypic features of myofibrillar myopathy associated with a BAG3 mutation. <i>Neuromuscular Disorders</i> , <b>2010</b> , 20, 438-42	2.9	77
31	North Star Ambulatory Assessment, 6-minute walk test and timed items in ambulant boys with Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , <b>2010</b> , 20, 712-6	2.9	134
30	Mitochondrial encephalomyopathies and related syndromes: brief review. <i>Cerebrovascular Diseases</i> , <b>2009</b> , 14, 38-52	3.2	3
29	Identification and characterization of novel collagen VI non-canonical splicing mutations causing Ullrich congenital muscular dystrophy. <i>Human Mutation</i> , <b>2009</b> , 30, E662-72	4.7	29
28	Molecular analysis of NPC1 and NPC2 gene in 34 Niemann-Pick C Italian patients: identification and structural modeling of novel mutations. <i>Neurogenetics</i> , <b>2009</b> , 10, 229-39	3	33
27	Reliability of the North Star Ambulatory Assessment in a multicentric setting. <i>Neuromuscular Disorders</i> , <b>2009</b> , 19, 458-61	2.9	125
26	Heart transplantation in a child with LGMD2I presenting as isolated dilated cardiomyopathy. <i>Neuromuscular Disorders</i> , <b>2008</b> , 18, 153-5	2.9	25
25	POMT1 and POMT2 mutations in CMD patients: a multicentric Italian study. <i>Neuromuscular Disorders</i> , <b>2008</b> , 18, 565-71	2.9	28
24	Daily salbutamol in young patients with SMA type II. <i>Neuromuscular Disorders</i> , <b>2008</b> , 18, 536-40	2.9	83
23	Clinical and genetic characterization of Chanarin-Dorfman syndrome. <i>Biochemical and Biophysical Research Communications</i> , <b>2008</b> , 369, 1125-8	3.4	60
22	Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. <i>Brain</i> , <b>2008</b> , 131, 747-59	11.2	112
21	Congenital myopathies. <i>Current Neurology and Neuroscience Reports</i> , <b>2008</b> , 8, 73-9	6.6	22
20	De novo LMNA mutations cause a new form of congenital muscular dystrophy. <i>Annals of Neurology</i> , <b>2008</b> , 64, 177-86	9.4	213
19	Molecular mechanisms and phenotypic variation in RYR1-related congenital myopathies. <i>Brain</i> , <b>2007</b> , 130, 2024-36	11.2	138
18	Randomized, double-blind, placebo-controlled trial of phenylbutyrate in spinal muscular atrophy. <i>Neurology</i> , <b>2007</b> , 68, 51-5	6.5	141
17	Gene expression profiling in the early phases of DMD: a constant molecular signature characterizes DMD muscle from early postnatal life throughout disease progression. <i>FASEB Journal</i> , <b>2007</b> , 21, 1210-26 <sup>0.9</sup>		168

16	The Hammersmith functional score correlates with the SMN2 copy number: a multicentric study. <i>Neuromuscular Disorders</i> , <b>2007</b> , 17, 400-3	2.9	39
15	Ullrich myopathy phenotype with secondary ColVI defect identified by confocal imaging and electron microscopy analysis. <i>Neuromuscular Disorders</i> , <b>2007</b> , 17, 587-96	2.9	20
14	Neonatal-onset nemaline myopathy mimicking congenital diaphragmatic hernia. <i>Journal of Pediatric Surgery</i> , <b>2007</b> , 42, E19-22	2.6	3
13	POMGnT1 mutations in congenital muscular dystrophy: genotype-phenotype correlation and expanded clinical spectrum. <i>Archives of Neurology</i> , <b>2006</b> , 63, 1491-5		30
12	Expanding the clinical spectrum of POMT1 phenotype. <i>Neurology</i> , <b>2006</b> , 66, 1564-7; discussion 1461	6.5	50
11	Reliability of the Hammersmith functional motor scale for spinal muscular atrophy in a multicentric study. <i>Neuromuscular Disorders</i> , <b>2006</b> , 16, 93-8	2.9	51
10	POMT2 mutation in a patient with SMEB-like phenotype. <i>Neuromuscular Disorders</i> , <b>2006</b> , 16, 446-8	2.9	35
9	Fatal hypertrophic cardiomyopathy and nemaline myopathy associated with ACTA1 K336E mutation. <i>Neuromuscular Disorders</i> , <b>2006</b> , 16, 548-52	2.9	68
8	Hypertrophic cardiomyopathy, cataract, developmental delay, lactic acidosis: a novel subtype of 3-methylglutaconic aciduria. <i>Journal of Inherited Metabolic Disease</i> , <b>2006</b> , 29, 546-50	5.4	22
7	Two patients with Dropped head syndrome due to mutations in LMNA or SEPN1 genes. <i>Neuromuscular Disorders</i> , <b>2005</b> , 15, 521-4	2.9	51
6	Major myofibrillar changes in early onset myopathy due to de novo heterozygous missense mutation in lamin A/C gene. <i>Neuromuscular Disorders</i> , <b>2005</b> , 15, 847-50	2.9	16
5	Acute quadriplegic myopathy in a 16-month-old child. <i>Paediatric Anaesthesia</i> , <b>2005</b> , 15, 611-5	1.8	
4	Phenotypic heterogeneity in two unrelated Danon patients associated with the same LAMP-2 gene mutation. <i>Neuropediatrics</i> , <b>2005</b> , 36, 309-13	1.6	22
3	Insulin-like growth factor I in inclusion-body myositis and human muscle cultures. <i>Journal of Neuropathology and Experimental Neurology</i> , <b>2004</b> , 63, 650-9	3.1	17
2	Genomic rearrangements at the IGHMBP2 gene locus in two patients with SMARD1. <i>Human Genetics</i> , <b>2004</b> , 115, 319-26	6.3	28
1	Pilot trial of phenylbutyrate in spinal muscular atrophy. <i>Neuromuscular Disorders</i> , <b>2004</b> , 14, 130-5	2.9	109