Eugenio Mercuri

List of Publications by Year
in descending order

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602 papers	33,331 citations	3515 90 h-index	7496 151 g-index
621 all docs	621 docs citations	621 times ranked	18996 citing authors

#	Article	IF	Citations
1	Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2017, 377, 1723-1732.	13.9	1,533
2	Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2018, 378, 625-635.	13.9	977
3	Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. Neuromuscular Disorders, 2018, 28, 103-115.	0.3	584
4	Mutations in the Fukutin-Related Protein Gene (FKRP) Cause a Form of Congenital Muscular Dystrophy with Secondary Laminin Î±2 Deficiency and Abnormal Glycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2001, 69, 1198-1209.	2.6	563
5	Origin and timing of brain lesions in term infants with neonatal encephalopathy. Lancet, The, 2003, 361, 736-742.	6.3	544
6	Duchenne muscular dystrophy. Nature Reviews Disease Primers, 2021, 7, 13.	18.1	448
7	Clinical and molecular genetic spectrum of autosomal dominant Emery-Dreifuss muscular dystrophy due to mutations of the lamin A/C gene. Annals of Neurology, 2000, 48, 170-180.	2.8	440
8	Longitudinal effect of eteplirsen versus historical control on ambulation in <scp>D</scp>uchenne muscular dystrophy. Annals of Neurology, 2016, 79, 257-271.	2.8	428
9	Diagnosis and management of spinal muscular atrophy: Part 2: Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics. Neuromuscular Disorders, 2018, 28, 197-207.	0.3	421
10	Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. Brain, 2007, 130, 2725-2735.	3.7	385
11	Ataluren in patients with nonsense mutation Duchenne muscular dystrophy (ACT DMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. Lancet, The, 2017, 390, 1489-1498.	6.3	365
12	Spinal muscular atrophy. Orphanet Journal of Rare Diseases, 2011, 6, 71.	1.2	363
13	Abnormal Magnetic Resonance Signal in the Internal Capsule Predicts Poor Neurodevelopmental Outcome in Infants With Hypoxic-Ischemic Encephalopathy. Pediatrics, 1998, 102, 323-328.	1.0	360
14	Muscle MRI in inherited neuromuscular disorders: Past, present, and future. Journal of Magnetic Resonance Imaging, 2007, 25, 433-440.	1.9	357
15	Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle and Nerve, 2014, 50, 477-487.	1.0	357
16	Muscular dystrophies. Lancet, The, 2013, 381, 845-860.	6.3	352
17	Childhood spinal muscular atrophy: controversies and challenges. Lancet Neurology, The, 2012, 11, 443-452.	4.9	297
18	Early Prognostic Indicators of Outcome in Infants With Neonatal Cerebral Infarction: A Clinical, Electroencephalogram, and Magnetic Resonance Imaging Study. Pediatrics, 1999, 103, 39-46.	1.0	289

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19	Muscular dystrophies. Lancet, The, 2019, 394, 2025-2038.	6.3	276
20	Optimality score for the neurologic examination of the infant at 12 and 18 months of age. Journal of Pediatrics, 1999, 135, 153-161.	0.9	262
21	The Childrenâ€™s Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP INTEND): Test development and reliability. Neuromuscular Disorders, 2010, 20, 155-161.	0.3	239
22	Risdiplam in Type 1 Spinal Muscular Atrophy. New England Journal of Medicine, 2021, 384, 915-923.	13.9	229
23	Phenotypic spectrum associated with mutations in the fukutin-related protein gene. Annals of Neurology, 2003, 53, 537-542.	2.8	219
24	Neurodevelopmental, emotional, and behavioural problems in Duchenne muscular dystrophy in relation to underlying dystrophin gene mutations. Developmental Medicine and Child Neurology, 2016, 58, 77-84.	1.1	213
25	Gene expression profiling in the early phases of DMD: a constant molecular signature characterizes DMD muscle from early postnatal life throughout disease progression. FASEB Journal, 2007, 21, 1210-1226.	0.2	209
26	Increased dystrophin production with golodirsen in patients with Duchenne muscular dystrophy. Neurology, 2020, 94, e2270-e2282.	1.5	207
27	The Hammersmith Functional Motor Scale for Children with Spinal Muscular Atrophy: a Scale to Test Ability and Monitor Progress in Children with Limited Ambulation. European Journal of Paediatric Neurology, 2003, 7, 155-159.	0.7	187
28	Clinical and imaging findings in six cases of congenital muscular dystrophy with rigid spine syndrome linked to chromosome 1p (RSMD1). Neuromuscular Disorders, 2002, 12, 631-638.	0.3	176
29	Development of the <scp>P</scp>erformance of the <scp>U</scp>pper <scp>L</scp>imb module for <scp>D</scp>uchenne muscular dystrophy. Developmental Medicine and Child Neurology, 2013, 55, 1038-1045.	1.1	173
30	Neonatal Cerebral Infarction and Neuromotor Outcome at School Age. Pediatrics, 2004, 113, 95-100.	1.0	172
31	Brain involvement in muscular dystrophies with defective dystroglycan glycosylation. Annals of Neurology, 2008, 64, 573-582.	2.8	172
32	Reliability of the North Star Ambulatory Assessment in a multicentric setting. Neuromuscular Disorders, 2009, 19, 458-461.	0.3	171
33	North Star Ambulatory Assessment, 6-minute walk test and timed items in ambulant boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2010, 20, 712-716.	0.3	171
34	Probabilistic diffusion tractography of the optic radiations and visual function in preterm infants at term equivalent age. Brain, 2008, 131, 573-582.	3.7	167
35	Congenital muscular dystrophies with defective glycosylation of dystroglycan. Neurology, 2009, 72, 1802-1809.	1.5	166
36	Revised upper limb module for spinal muscular atrophy: Development of a new module. Muscle and Nerve, 2017, 55, 869-874.	1.0	166

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37	An optimality score for the neurologic examination of the term newborn. Journal of Pediatrics, 1998, 133, 406-416.	0.9	162
38	Prothrombotic Disorders and Abnormal Neurodevelopmental Outcome in Infants With Neonatal Cerebral Infarction. Pediatrics, 2001, 107, 1400-1404.	1.0	162
39	Randomized, double-blind, placebo-controlled trial of phenylbutyrate in spinal muscular atrophy. Neurology, 2007, 68, 51-55.	1.5	159
40	Magnetic Resonance Image Correlates of Hemiparesis After Neonatal and Childhood Middle Cerebral Artery Stroke. Pediatrics, 2005, 115, 321-326.	1.0	158
41	Autosomal recessive inheritance of RYR1 mutations in a congenital myopathy with cores. Neurology, 2002, 59, 284-287.	1.5	157
42	Neurological and Perceptual-Motor Outcome at 5 - 6 Years of Age in Children with Neonatal Encephalopathy: Relationship with Neonatal Brain MRI. Neuropediatrics, 2002, 33, 242-248.	0.3	156
43	Muscle MRI in Ullrich congenital muscular dystrophy and Bethlem myopathy. Neuromuscular Disorders, 2005, 15, 303-310.	0.3	154
44	Functional changes in Duchenne muscular dystrophy. Neurology, 2011, 77, 250-256.	1.5	151
45	Phenylbutyrate increases SMN gene expression in spinal muscular atrophy patients. European Journal of Human Genetics, 2005, 13, 256-259.	1.4	148
46	Mutations in KANSL1 cause the 17q21.31 microdeletion syndrome phenotype. Nature Genetics, 2012, 44, 636-638.	9.4	148
47	Use of the Hammersmith Infant Neurological Examination in infants with cerebral palsy: a critical review of the literature. Developmental Medicine and Child Neurology, 2016, 58, 240-245.	1.1	145
48	Histological effects of givinostat in boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, 643-649.	0.3	144
49	Development of Exon Skipping Therapies for Duchenne Muscular Dystrophy: A Critical Review and a Perspective on the Outstanding Issues. Nucleic Acid Therapeutics, 2017, 27, 251-259.	2.0	144
50	Muscle magnetic resonance imaging involvement in muscular dystrophies with rigidity of the spine. Annals of Neurology, 2010, 67, 201-208.	2.8	143
51	Patterns of disease progression in type 2 and 3 SMA: Implications for clinical trials. Neuromuscular Disorders, 2016, 26, 126-131.	0.3	142
52	Natural history of Ullrich congenital muscular dystrophy. Neurology, 2009, 73, 25-31.	1.5	141
53	Muscle MRI findings in patients with limb girdle muscular dystrophy with calpain 3 deficiency (LGMD2A) and early contractures. Neuromuscular Disorders, 2005, 15, 164-171.	0.3	140
54	Neurofilament as a potential biomarker for spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 932-944.	1.7	137

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55	Magnetic resonance imaging of muscle in congenital myopathies associated with RYR1 mutations. Neuromuscular Disorders, 2004, 14, 785-790.	0.3	135
56	Antepartum and Intrapartum Factors Preceding Neonatal Hypoxic-Ischemic Encephalopathy. Pediatrics, 2013, 132, e952-e959.	1.0	130
57	Pilot trial of phenylbutyrate in spinal muscular atrophy. Neuromuscular Disorders, 2004, 14, 130-135.	0.3	128
58	Head Growth in Infants With Hypoxicâ€“Ischemic Encephalopathy: Correlation With Neonatal Magnetic Resonance Imaging. Pediatrics, 2000, 106, 235-243.	1.0	127
59	General Movements Detect Early Signs of Hemiplegia in Term Infants with Neonatal Cerebral Infarction. Neuropediatrics, 2003, 34, 61-66.	0.3	126
60	Fracture prevalence in Duchenne muscular dystrophy. Developmental Medicine and Child Neurology, 2002, 44, 695-698.	1.1	123
61	DPM2â€CDG: A muscular dystrophyâ€“dystroglycanopathy syndrome with severe epilepsy. Annals of Neurology, 2012, 72, 550-558.	2.8	121
62	Attention Deficit Hyperactivity Disorder and Cognitive Function in Duchenne Muscular Dystrophy: Phenotype-Genotype Correlation. Journal of Pediatrics, 2012, 161, 705-709.e1.	0.9	121
63	Does cranial ultrasound imaging identify arterial cerebral infarction in term neonates?. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2005, 90, F252-f256.	1.4	118
64	Skeletal muscle pathology in autosomal dominant Emery-Dreifuss muscular dystrophy with lamin A/C mutations. Neuropathology and Applied Neurobiology, 2001, 27, 281-290.	1.8	117
65	Combined Use of Electroencephalogram and Magnetic Resonance Imaging in Full-Term Neonates With Acute Encephalopathy. Pediatrics, 2001, 107, 461-468.	1.0	115
66	Extreme Variability of Phenotype in Patients With an Identical Missense Mutation in the Lamin A/C Gene. Archives of Neurology, 2004, 61, 690.	4.9	114
67	Disease severity in dominant Emery Dreifuss is increased by mutations in both emerin and desmin proteins. Brain, 2006, 129, 1260-1268.	3.7	114
68	Onasemnogene abeparvovec gene therapy for symptomatic infantile-onset spinal muscular atrophy type 1 (STR1VE-EU): an open-label, single-arm, multicentre, phase 3 trial. Lancet Neurology, The, 2021, 20, 832-841.	4.9	112
69	Minor neurological signs and perceptual-motor difficulties in prematurely born children. Archives of Disease in Childhood: Fetal and Neonatal Edition, 1997, 76, F9-F14.	1.4	111
70	Pilot trial of albuterol in spinal muscular atrophy. Neurology, 2002, 59, 609-610.	1.5	111
71	Novel mutations expand the clinical spectrum of DYNC1H1 -associated spinal muscular atrophy. Neurology, 2015, 84, 668-679.	1.5	106
72	A short protocol for muscle MRI in children with muscular dystrophies. European Journal of Paediatric Neurology, 2002, 6, 305-7.	0.7	105

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73	Motor and perceptualâ€“motor competence in children with Down syndrome: variation in performance with age. European Journal of Paediatric Neurology, 1999, 3, 7-14.	0.7	104
74	Collagen VI involvement in Ullrich syndrome. Neurology, 2002, 58, 1354-1359.	1.5	103
75	Content validity and clinical meaningfulness of the HFMSE in spinal muscular atrophy. BMC Neurology, 2017, 17, 39.	0.8	102
76	A randomized placebo-controlled phase 3 trial of an antisense oligonucleotide, drisapersen, in Duchenne muscular dystrophy. Neuromuscular Disorders, 2018, 28, 4-15.	0.3	102
77	Selective Muscle Involvement on Magnetic Resonance Imaging in Autosomal Dominant Emery-Dreifuss Muscular Dystrophy. Neuropediatrics, 2002, 33, 10-14.	0.3	101
78	The NorthStar Ambulatory Assessment in Duchenne muscular dystrophy: considerations for the design of clinical trials. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, jnnp-2014-309405.	0.9	99
79	24 Month Longitudinal Data in Ambulant Boys with Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e52512.	1.1	99
80	Magnetic resonance imaging of muscle in nemaline myopathy. Neuromuscular Disorders, 2004, 14, 779-784.	0.3	98
81	Cognitive Outcome at Early School Age in Term-Born Children With Perinatally Acquired Middle Cerebral Artery Territory Infarction. Stroke, 2008, 39, 403-410.	1.0	98
82	The everâ€expanding spectrum of congenital muscular dystrophies. Annals of Neurology, 2012, 72, 9-17.	2.8	98
83	Long Term Natural History Data in Ambulant Boys with Duchenne Muscular Dystrophy: 36-Month Changes. PLoS ONE, 2014, 9, e108205.	1.1	98
84	Ischaemic and haemorrhagic brain lesions in newborns with seizures and normal Apgar scores.. Archives of Disease in Childhood: Fetal and Neonatal Edition, 1995, 73, F67-F74.	1.4	97
85	Correlation between visual function, neurodevelopmental outcome, and magnetic resonance imaging findings in infants with periventricular leucomalacia. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2000, 82, 134F-140.	1.4	97
86	Spectrum of Brain Changes in Patients With Congenital Muscular Dystrophy and FKRP Gene Mutations. Archives of Neurology, 2006, 63, 251.	4.9	97
87	Daily salbutamol in young patients with SMA type II. Neuromuscular Disorders, 2008, 18, 536-540.	0.3	97
88	Follow-up to Age 4 Years of Treatment of Type 1 Retinopathy of Prematurity Intravitreal Bevacizumab Injection versus Laser: Fluorescein Angiographic Findings. Ophthalmology, 2018, 125, 218-226.	2.5	97
89	Developmental milestones in type I spinal muscular atrophy. Neuromuscular Disorders, 2016, 26, 754-759.	0.3	96
90	The Dubowitz neurological examination of the full-term newborn. Mental Retardation and Developmental Disabilities Research Reviews, 2005, 11, 52-60.	3.5	95

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91	Application of the Sleep Disturbance Scale for Children (SDSC) in preschool age. European Journal of Paediatric Neurology, 2013, 17, 374-382.	0.7	95
92	Safety and efficacy of olesoxime in patients with type 2 or non-ambulatory type 3 spinal muscular atrophy: a randomised, double-blind, placebo-controlled phase 2 trial. Lancet Neurology, The, 2017, 16, 513-522.	4.9	95
93	Neurologic examination in infants with hypoxic-ischemic encephalopathy at age 9 to 14 months: Use of optimality scores and correlation with magnetic resonance imaging findings. Journal of Pediatrics, 2001, 138, 332-337.	0.9	94
94	Muscle imaging in clinical practice: diagnostic value of muscle magnetic resonance imaging in inherited neuromuscular disorders. Current Opinion in Neurology, 2005, 18, 526-537.	1.8	94
95	Feeding problems and weight gain in Duchenne muscular dystrophy. European Journal of Paediatric Neurology, 2006, 10, 231-236.	0.7	94
96	TREAT-NMD workshop: Pattern recognition in genetic muscle diseases using muscle MRI. Neuromuscular Disorders, 2012, 22, S42-S53.	0.3	93
97	Mild phenotype of nemaline myopathy with sleep hypoventilation due to a mutation in the skeletal muscle Î±-actin (ACTA1) gene. Neuromuscular Disorders, 2001, 11, 35-40.	0.3	92
98	Feeding problems and malnutrition in spinal muscular atrophy type II. Neuromuscular Disorders, 2008, 18, 389-393.	0.3	92
99	Mutation Update and Genotype-Phenotype Correlations of Novel and Previously Described Mutations inTPM2andTPM3Causing Congenital Myopathies. Human Mutation, 2014, 35, 779-790.	1.1	92
100	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.5	92
101	Fracture prevalence in Duchenne muscular dystrophy. Developmental Medicine and Child Neurology, 2002, 44, 695-8.	1.1	89
102	Muscle Magnetic Resonance Imaging in Congenital Myopathies Due to Ryanodine Receptor Type 1 Gene Mutations. Archives of Neurology, 2011, 68, 1171.	4.9	89
103	Spinal muscular atrophy â€" insights and challenges in the treatment era. Nature Reviews Neurology, 2020, 16, 706-715.	4.9	89
104	Safety and efficacy of once-daily risdiplam in type 2 and non-ambulant type 3 spinal muscular atrophy (SUNFISH part 2): a phase 3, double-blind, randomised, placebo-controlled trial. Lancet Neurology, The, 2022, 21, 42-52.	4.9	89
105	Neonatal Neurological Examination in Infants with Hypoxic Ischaemic Encephalopathy: Correlation with MRI Findings. Neuropediatrics, 1999, 30, 83-89.	0.3	86
106	Visual function in term infants with hypoxic-ischaemic insults: correlation with neurodevelopment at 2Âyears of age. Archives of Disease in Childhood: Fetal and Neonatal Edition, 1999, 80, F99-F104.	1.4	85
107	Natural history of pulmonary function in collagen VI-related myopathies. Brain, 2013, 136, 3625-3633.	3.7	85
108	Neurologic examination of preterm infants at term age: Comparison with term infants. Journal of Pediatrics, 2003, 142, 647-655.	0.9	83

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109	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. Neuromuscular Disorders, 2014, 24, 201-206.	0.3	83
110	Nusinersen in type 1 spinal muscular atrophy: Twelveâ€month realâ€world data. Annals of Neurology, 2019, 86, 443-451.	2.8	83
111	Cognitive profile of disorders associated with dysregulation of the RAS/MAPK signaling cascade. American Journal of Medical Genetics, Part A, 2009, 149A, 140-146.	0.7	82
112	Safety and effectiveness of ataluren: comparison of results from the STRIDE Registry and CINRG DMD Natural History Study. Journal of Comparative Effectiveness Research, 2020, 9, 341-360.	0.6	82
113	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. Neurology, 2012, 79, 159-162.	1.5	81
114	Risk Factors for Neonatal Arterial Ischemic Stroke: The Importance of the Intrapartum Period. Journal of Pediatrics, 2016, 173, 62-68.e1.	0.9	81
115	A short protocol for muscle MRI in children with muscular dystrophies. European Journal of Paediatric Neurology, 2002, 6, 305-307.	0.7	79
116	Cerebellar infarction and atrophy in infants and children with a history of premature birth. Pediatric Radiology, 1997, 27, 139-143.	1.1	77
117	Neurodevelopmental outcome at 12 and 18 months in late preterm infants. European Journal of Paediatric Neurology, 2010, 14, 503-507.	0.7	77
118	SEPN1 -related myopathies. Neurology, 2011, 76, 2073-2078.	1.5	77
119	Stakeholder cooperation to overcome challenges in orphan medicine development: the example of Duchenne muscular dystrophy. Lancet Neurology, The, 2016, 15, 882-890.	4.9	77
120	Mapping the differences in care for 5,000 Spinal Muscular Atrophy patients, a survey of 24 national registries in North America, Australasia and Europe. Journal of Neurology, 2014, 261, 152-163.	1.8	76
121	Thalamic atrophy in infants with PVL and cerebral visual impairment. Early Human Development, 2006, 82, 591-595.	0.8	75
122	Prevalence of congenital muscular dystrophy in Italy. Neurology, 2015, 84, 904-911.	1.5	75
123	A current approach to heart failure in Duchenne muscular dystrophy. Heart, 2017, 103, 1770-1779.	1.2	75
124	European ad-hoc consensus statement on gene replacement therapy for spinal muscular atrophy. European Journal of Paediatric Neurology, 2020, 28, 38-43.	0.7	74
125	Visual Function in Full-Term Infants with Hypoxic-Ischaemic Encephalopathy. Neuropediatrics, 1997, 28, 155-161.	0.3	72
126	Muscle magnetic resonance imaging in patients with congenital muscular dystrophy and Ullrich phenotype. Neuromuscular Disorders, 2003, 13, 554-558.	0.3	72

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127	Sleep disorders in children with cerebral palsy: neurodevelopmental and behavioral correlates. Sleep Medicine, 2014, 15, 213-218.	0.8	72
128	Assessing upper limb function in nonambulant SMA patients: Development of a new module. Neuromuscular Disorders, 2011, 21, 406-412.	0.3	71
129	Association Study of Exon Variants in the NF-Î°B and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	2.6	71
130	Upper Girdle Imaging in Facioscapulohumeral Muscular Dystrophy. PLoS ONE, 2014, 9, e100292.	1.1	71
131	Visual disorders in children with brain lesions:. European Journal of Paediatric Neurology, 2001, 5, 115-119.	0.7	70
132	MYH7-related myopathies: clinical, histopathological and imaging findings in a cohort of Italian patients. Orphanet Journal of Rare Diseases, 2016, 11, 91.	1.2	70
133	Visual function of prematurely born children with and without perceptual-motor difficulties. Early Human Development, 1996, 45, 73-82.	0.8	69
134	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. JAMA Neurology, 2018, 75, 557.	4.5	69
135	An Integrated Safety Analysis of Infants and Children with Symptomatic Spinal Muscular Atrophy (SMA) Treated with Nusinersen in Seven Clinical Trials. CNS Drugs, 2019, 33, 919-932.	2.7	69
136	Extreme variability of skeletal and cardiac muscle involvement in patients with mutations in exon 11 of the lamin A/C gene. Muscle and Nerve, 2005, 31, 602-609.	1.0	68
137	Swallowing difficulties in Duchenne muscular dystrophy: Indications for feeding assessment and outcome of videofluroscopic swallow studies. European Journal of Paediatric Neurology, 2008, 12, 239-245.	0.7	68
138	Predictive factors for the development of scoliosis in Duchenne muscular dystrophy. European Journal of Paediatric Neurology, 2007, 11, 160-166.	0.7	67
139	Muscle MRI in Becker muscular dystrophy. Neuromuscular Disorders, 2012, 22, S100-S106.	0.3	67
140	A critical review of functional assessment tools for upper limbs in Duchenne muscular dystrophy. Developmental Medicine and Child Neurology, 2012, 54, 879-885.	1.1	67
141	Revised Hammersmith Scale for spinal muscular atrophy: A SMA specific clinical outcome assessment tool. PLoS ONE, 2017, 12, e0172346.	1.1	67
142	Nusinersen in type 1 SMA infants, children and young adults: Preliminary results on motor function. Neuromuscular Disorders, 2018, 28, 582-585.	0.3	67
143	6 Minute Walk Test in Duchenne MD Patients with Different Mutations: 12 Month Changes. PLoS ONE, 2014, 9, e83400.	1.1	65
144	Ambulatory function in spinal muscular atrophy: Age-related patterns of progression. PLoS ONE, 2018, 13, e0199657.	1.1	65

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145	Somatosensory and Visual Evoked Potentials in Congenital Muscular Dystrophy: Correlation with MRI Changes and Muscle Merosin Status. Neuropediatrics, 1995, 26, 3-7.	0.3	64
146	Reliability of the Hammersmith functional motor scale for spinal muscular atrophy in a multicentric study. Neuromuscular Disorders, 2006, 16, 93-98.	0.3	64
147	Neurological examination of preterm infants at term equivalent age. Early Human Development, 2008, 84, 751-761.	0.8	64
148	Autosomal recessive Bethlem myopathy. Neurology, 2009, 73, 1883-1891.	1.5	64
149	Behavioral Profile in RASopathies. American Journal of Medical Genetics, Part A, 2014, 164, 934-942.	0.7	64
150	Evolution of Unilateral Perinatal Arterial Ischemic Stroke on Conventional and Diffusion-Weighted MR Imaging. American Journal of Neuroradiology, 2009, 30, 998-1004.	1.2	63
151	Neuropsychological development in children with Dravet syndrome. Epilepsy Research, 2011, 95, 86-93.	0.8	63
152	Genetic characterization in symptomatic female DMD carriers: lack of relationship between X-inactivation, transcriptional DMD allele balancing and phenotype. BMC Medical Genetics, 2012, 13, 73.	2.1	63
153	Early neurodevelopmental assessment in Duchenne muscular dystrophy. Neuromuscular Disorders, 2013, 23, 451-455.	0.3	63
154	Muscular dystrophy. Current Opinion in Pediatrics, 2013, 25, 701-707.	1.0	63
155	Management of scoliosis in Duchenne muscular dystrophy: a large 10-year retrospective study. Developmental Medicine and Child Neurology, 2006, 48, 513.	1.1	63
156	Hand movements at 3â€ƒmonths predict later hemiplegia in term infants with neonatal cerebral infarction. Developmental Medicine and Child Neurology, 2010, 52, 767-772.	1.1	62
157	Nusinersen improves walking distance and reduces fatigue in laterâ€onset spinal muscular atrophy. Muscle and Nerve, 2019, 60, 409-414.	1.0	62
158	Clinical Trial and Postmarketing Safety of Onasemnogene Abeparvovec Therapy. Drug Safety, 2021, 44, 1109-1119.	1.4	62
159	Salbutamol increases survival motor neuron (SMN) transcript levels in leucocytes of spinal muscular atrophy (SMA) patients: relevance for clinical trial design. Journal of Medical Genetics, 2010, 47, 856-858.	1.5	61
160	Revised upper limb module for spinal muscular atrophy: 12â€‰month changes. Muscle and Nerve, 2019, 59, 426-430.	1.0	61
161	Minicore myopathy in children: a clinical and histopathological study of 19 cases. Neuromuscular Disorders, 2000, 10, 264-273.	0.3	60
162	Duchenne muscular dystrophy and epilepsy. Neuromuscular Disorders, 2013, 23, 313-315.	0.3	60

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163	Rasch analysis of clinical outcome measures in spinal muscular atrophy. Muscle and Nerve, 2014, 49, 422-430.	1.0	60
164	Prognostic value of the neurologic optimality score at 9 and 18 months in preterm infants born before 31 weeks' gestation. Journal of Pediatrics, 2002, 140, 57-60.	0.9	59
165	Expanding the phenotype of potassium channelopathy: severe neuromyotonia and skeletal deformities without prominent Episodic Ataxia. Neuromuscular Disorders, 2004, 14, 689-693.	0.3	58
166	Motor function in type 2 and 3 SMA patients treated with Nusinersen: a critical review and meta-analysis. Orphanet Journal of Rare Diseases, 2021, 16, 430.	1.2	58
167	Long-Term Safety and Efficacy Data of Golodirsen in Ambulatory Patients with Duchenne Muscular Dystrophy Amenable to Exon 53 Skipping: A First-in-human, Multicenter, Two-Part, Open-Label, Phase 1/2 Trial. Nucleic Acid Therapeutics, 2022, 32, 29-39.	2.0	58
168	LMNA -associated myopathies. Neurology, 2014, 83, 1634-1644.	1.5	57
169	Categorizing natural history trajectories of ambulatory function measured by the 6-minute walk distance in patients with Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, 576-583.	0.3	57
170	Therapeutic approaches for spinal muscular atrophy (SMA). Gene Therapy, 2017, 24, 514-519.	2.3	56
171	Pilot Trial of Salbutamol in Central Core and Multi-Minicore Diseases. Neuropediatrics, 2004, 35, 262-266.	0.3	55
172	Maturation of cerebral electrical activity and development of cortical folding in young very preterm infants. Clinical Neurophysiology, 2007, 118, 53-59.	0.7	55
173	Visual Function at 35 and 40 Weeks' Postmenstrual Age in Low-Risk Preterm Infants. Pediatrics, 2008, 122, e1193-e1198.	1.0	55
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