Eugenio Mercuri

List of Publications by Citations

526 papers	23,599 citations	79 h-index	129 g-index
621 ext. papers	28,905 ext. citations	4.1 avg, IF	6.56 L-index

#	Paper	IF	Citations
526	Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. *New England Journal of Medicine*, **2017**, 377, 1723-1732	59.2	957
525	Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. *New England Journal of Medicine*, **2018**, 378, 625-635	59.2	617
524	Mutations in the fukutin-related protein gene (FKRP) cause a form of congenital muscular dystrophy with secondary laminin alpha2 deficiency and abnormal glycosylation of alpha-dystroglycan. *American Journal of Human Genetics*, **2001**, 69, 1198-209	11	498
523	Origin and timing of brain lesions in term infants with neonatal encephalopathy. *Lancet, The*, **2003**, 361, 736-42	40	467
522	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	9.4	367
521	Longitudinal effect of eteplirsen versus historical control on ambulation in Duchenne muscular dystrophy. *Annals of Neurology*, **2016**, 79, 257-71	9.4	329
520	Refining genotype phenotype correlations in muscular dystrophies with defective glycosylation of dystroglycan. *Brain*, **2007**, 130, 2725-35	11.2	322
519	Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. *Neuromuscular Disorders*, **2018**, 28, 103-115	2.9	319
518	Abnormal magnetic resonance signal in the internal capsule predicts poor neurodevelopmental outcome in infants with hypoxic-ischemic encephalopathy. *Pediatrics*, **1998**, 102, 323-8	7.4	305
517	Ataluren treatment of patients with nonsense mutation dystrophinopathy. *Muscle and Nerve*, **2014**, 50, 477-87	3.4	281
516	Muscular dystrophies. *Lancet, The*, **2013**, 381, 845-60	40	280
515	Muscle MRI in inherited neuromuscular disorders: past, present, and future. *Journal of Magnetic Resonance Imaging*, **2007**, 25, 433-40	5.6	277
514	Early prognostic indicators of outcome in infants with neonatal cerebral infarction: a clinical, electroencephalogram, and magnetic resonance imaging study. *Pediatrics*, **1999**, 103, 39-46	7.4	252
513	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	40	237
512	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	2.9	236
511	Spinal muscular atrophy. *Orphanet Journal of Rare Diseases*, **2011**, 6, 71	4.2	223
510	Childhood spinal muscular atrophy: controversies and challenges. *Lancet Neurology, The*, **2012**, 11, 443-52	24.1	200

509	Phenotypic spectrum associated with mutations in the fukutin-related protein gene. *Annals of Neurology*, **2003**, 53, 537-42	9.4	193
508	Optimality score for the neurologic examination of the infant at 12 and 18 months of age. *Journal of Pediatrics*, **1999**, 135, 153-61	3.6	193
507	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, 1210-26	0.9	168
506	The Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP INTEND): test development and reliability. *Neuromuscular Disorders*, **2010**, 20, 155-61	2.9	152
505	Probabilistic diffusion tractography of the optic radiations and visual function in preterm infants at term equivalent age. *Brain*, **2008**, 131, 573-82	11.2	150
504	Neonatal cerebral infarction and neuromotor outcome at school age. *Pediatrics*, **2004**, 113, 95-100	7.4	148
503	Brain involvement in muscular dystrophies with defective dystroglycan glycosylation. *Annals of Neurology*, **2008**, 64, 573-82	9.4	144
502	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, 155-9	3.8	142
501	Prothrombotic disorders and abnormal neurodevelopmental outcome in infants with neonatal cerebral infarction. *Pediatrics*, **2001**, 107, 1400-4	7.4	142
500	An optimality score for the neurologic examination of the term newborn. *Journal of Pediatrics*, **1998**, 133, 406-16	3.6	141
499	Randomized, double-blind, placebo-controlled trial of phenylbutyrate in spinal muscular atrophy. *Neurology*, **2007**, 68, 51-5	6.5	141
498	Development of the Performance of the Upper Limb module for Duchenne muscular dystrophy. *Developmental Medicine and Child Neurology*, **2013**, 55, 1038-45	3.3	139
497	Autosomal recessive inheritance of RYR1 mutations in a congenital myopathy with cores. *Neurology* , **2002**, 59, 284-7	6.5	139
496	Phenylbutyrate increases SMN gene expression in spinal muscular atrophy patients. *European Journal of Human Genetics*, **2005**, 13, 256-9	5.3	137
495	Magnetic resonance image correlates of hemiparesis after neonatal and childhood middle cerebral artery stroke. *Pediatrics*, **2005**, 115, 321-6	7.4	135
494	North Star Ambulatory Assessment, 6-minute walk test and timed items in ambulant boys with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2010**, 20, 712-6	2.9	134
493	Congenital muscular dystrophies with defective glycosylation of dystroglycan: a population study. *Neurology*, **2009**, 72, 1802-9	6.5	134
492	Neurological and perceptual-motor outcome at 5 - 6 years of age in children with neonatal encephalopathy: relationship with neonatal brain MRI. *Neuropediatrics*, **2002**, 33, 242-8	1.6	132

491	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	3.3	132
490	Clinical and imaging findings in six cases of congenital muscular dystrophy with rigid spine syndrome linked to chromosome 1p (RSMD1). *Neuromuscular Disorders*, **2002**, 12, 631-8	2.9	130
489	Functional changes in Duchenne muscular dystrophy: a 12-month longitudinal cohort study. *Neurology*, **2011**, 77, 250-6	6.5	125
488	Reliability of the North Star Ambulatory Assessment in a multicentric setting. *Neuromuscular Disorders*, **2009**, 19, 458-61	2.9	125
487	Muscle MRI in Ullrich congenital muscular dystrophy and Bethlem myopathy. *Neuromuscular Disorders*, **2005**, 15, 303-10	2.9	122
486	Natural history of Ullrich congenital muscular dystrophy. *Neurology*, **2009**, 73, 25-31	6.5	118
485	Muscle magnetic resonance imaging involvement in muscular dystrophies with rigidity of the spine. *Annals of Neurology*, **2010**, 67, 201-8	9.4	118
484	Mutations in KANSL1 cause the 17q21.31 microdeletion syndrome phenotype. *Nature Genetics*, **2012**, 44, 636-8	36.3	110
483	General movements detect early signs of hemiplegia in term infants with neonatal cerebral infarction. *Neuropediatrics*, **2003**, 34, 61-6	1.6	109
482	Pilot trial of phenylbutyrate in spinal muscular atrophy. *Neuromuscular Disorders*, **2004**, 14, 130-5	2.9	109
481	Muscle MRI findings in patients with limb girdle muscular dystrophy with calpain 3 deficiency (LGMD2A) and early contractures. *Neuromuscular Disorders*, **2005**, 15, 164-71	2.9	108
480	Increased dystrophin production with golodirsen in patients with Duchenne muscular dystrophy. *Neurology*, **2020**, 94, e2270-e2282	6.5	106
479	Magnetic resonance imaging of muscle in congenital myopathies associated with RYR1 mutations. *Neuromuscular Disorders*, **2004**, 14, 785-90	2.9	106
478	Patterns of disease progression in type 2 and 3 SMA: Implications for clinical trials. *Neuromuscular Disorders*, **2016**, 26, 126-31	2.9	105
477	Fracture prevalence in Duchenne muscular dystrophy. *Developmental Medicine and Child Neurology*, **2007**, 44, 695-698	3.3	105
476	Disease severity in dominant Emery Dreifuss is increased by mutations in both emerin and desmin proteins. *Brain*, **2006**, 129, 1260-8	11.2	105
475	DPM2-CDG: a muscular dystrophy-dystroglycanopathy syndrome with severe epilepsy. *Annals of Neurology*, **2012**, 72, 550-8	9.4	103
474	Combined use of electroencephalogram and magnetic resonance imaging in full-term neonates with acute encephalopathy. *Pediatrics*, **2001**, 107, 461-8	7.4	102

473	Head growth in infants with hypoxic-ischemic encephalopathy: correlation with neonatal magnetic resonance imaging. *Pediatrics*, **2000**, 106, 235-43	7.4	101
472	Antepartum and intrapartum factors preceding neonatal hypoxic-ischemic encephalopathy. *Pediatrics*, **2013**, 132, e952-9	7.4	100
471	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	4.8	100
470	Muscular dystrophies. *Lancet, The*, **2019**, 394, 2025-2038	40	99
469	Extreme variability of phenotype in patients with an identical missense mutation in the lamin A/C gene: from congenital onset with severe phenotype to milder classic Emery-Dreifuss variant. *Archives of Neurology*, **2004**, 61, 690-4		96
468	Pilot trial of albuterol in spinal muscular atrophy. *Neurology*, **2002**, 59, 609-10	6.5	96
467	Histological effects of givinostat in boys with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2016**, 26, 643-649	2.9	96
466	Does cranial ultrasound imaging identify arterial cerebral infarction in term neonates?. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **2005**, 90, F252-6	4.7	95
465	Spectrum of brain changes in patients with congenital muscular dystrophy and FKRP gene mutations. *Archives of Neurology*, **2006**, 63, 251-7		94
464	Minor neurological signs and perceptual-motor difficulties in prematurely born children. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **1997**, 76, F9-14	4.7	93
463	Collagen VI involvement in Ullrich syndrome: a clinical, genetic, and immunohistochemical study. *Neurology*, **2002**, 58, 1354-9	6.5	89
462	Skeletal muscle pathology in autosomal dominant Emery-Dreifuss muscular dystrophy with lamin A/C mutations. *Neuropathology and Applied Neurobiology*, **2001**, 27, 281-90	5.2	88
461	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, 332-7	3.6	87
460	Ischaemic and haemorrhagic brain lesions in newborns with seizures and normal Apgar scores. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **1995**, 73, F67-74	4.7	87
459	Duchenne muscular dystrophy. *Nature Reviews Disease Primers*, **2021**, 7, 13	51.1	87
458	Selective muscle involvement on magnetic resonance imaging in autosomal dominant Emery-Dreifuss muscular dystrophy. *Neuropediatrics*, **2002**, 33, 10-4	1.6	85
457	A short protocol for muscle MRI in children with muscular dystrophies. *European Journal of Paediatric Neurology*, **2002**, 6, 305-7	3.8	85
456	Cognitive outcome at early school age in term-born children with perinatally acquired middle cerebral artery territory infarction. *Stroke*, **2008**, 39, 403-10	6.7	84

455	Motor and perceptual-motor competence in children with Down syndrome: variation in performance with age. *European Journal of Paediatric Neurology*, **1999**, 3, 7-13	3.8	84
454	Daily salbutamol in young patients with SMA type II. *Neuromuscular Disorders*, **2008**, 18, 536-40	2.9	83
453	Long term natural history data in ambulant boys with Duchenne muscular dystrophy: 36-month changes. *PLoS ONE*, **2014**, 9, e108205	3.7	82
452	Attention deficit hyperactivity disorder and cognitive function in Duchenne muscular dystrophy: phenotype-genotype correlation. *Journal of Pediatrics*, **2012**, 161, 705-9.e1	3.6	82
451	The ever-expanding spectrum of congenital muscular dystrophies. *Annals of Neurology*, **2012**, 72, 9-17	9.4	82
450	Novel mutations expand the clinical spectrum of DYNC1H1-associated spinal muscular atrophy. *Neurology*, **2015**, 84, 668-79	6.5	81
449	Revised upper limb module for spinal muscular atrophy: Development of a new module. *Muscle and Nerve*, **2017**, 55, 869-874	3.4	81
448	Neonatal neurological examination in infants with hypoxic ischaemic encephalopathy: correlation with MRI findings. *Neuropediatrics*, **1999**, 30, 83-9	1.6	78
447	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, 240-5	3.3	78
446	Magnetic resonance imaging of muscle in nemaline myopathy. *Neuromuscular Disorders*, **2004**, 14, 779-84	2.9	77
445	The Dubowitz neurological examination of the full-term newborn. *Mental Retardation and Developmental Disabilities Research Reviews*, **2005**, 11, 52-60		77
444	Mild phenotype of nemaline myopathy with sleep hypoventilation due to a mutation in the skeletal muscle alpha-actin (ACTA1) gene. *Neuromuscular Disorders*, **2001**, 11, 35-40	2.9	77
443	Neurofilament as a potential biomarker for spinal muscular atrophy. *Annals of Clinical and Translational Neurology*, **2019**, 6, 932-944	5.3	76
442	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, F134-40	4.7	76
441	24 month longitudinal data in ambulant boys with Duchenne muscular dystrophy. *PLoS ONE*, **2013**, 8, e52512	3.7	75
440	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	24.1	74
439	TREAT-NMD workshop: pattern recognition in genetic muscle diseases using muscle MRI: 25-26 February 2011, Rome, Italy. *Neuromuscular Disorders*, **2012**, 22 Suppl 2, S42-53	2.9	73
438	Muscle magnetic resonance imaging in congenital myopathies due to ryanodine receptor type 1 gene mutations. *Archives of Neurology*, **2011**, 68, 1171-9		72

437	Feeding problems and weight gain in Duchenne muscular dystrophy. *European Journal of Paediatric Neurology*, **2006**, 10, 231-6	3.8	72
436	Cerebellar infarction and atrophy in infants and children with a history of premature birth. *Pediatric Radiology*, **1997**, 27, 139-43	2.8	71
435	The genetic basis of undiagnosed muscular dystrophies and myopathies: Results from 504 patients. *Neurology*, **2016**, 87, 71-6	6.5	70
434	Natural history of pulmonary function in collagen VI-related myopathies. *Brain*, **2013**, 136, 3625-33	11.2	69
433	Feeding problems and malnutrition in spinal muscular atrophy type II. *Neuromuscular Disorders*, **2008**, 18, 389-93	2.9	69
432	Risdiplam in Type 1 Spinal Muscular Atrophy. *New England Journal of Medicine*, **2021**, 384, 915-923	59.2	69
431	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2014**, 24, 201-6	2.9	68
430	Thalamic atrophy in infants with PVL and cerebral visual impairment. *Early Human Development*, **2006**, 82, 591-5	2.2	68
429	Muscle imaging in clinical practice: diagnostic value of muscle magnetic resonance imaging in inherited neuromuscular disorders. *Current Opinion in Neurology*, **2005**, 18, 526-37	7.1	68
428	A randomized placebo-controlled phase 3 trial of an antisense oligonucleotide, drisapersen, in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2018**, 28, 4-15	2.9	68
427	Mutation update and genotype-phenotype correlations of novel and previously described mutations in TPM2 and TPM3 causing congenital myopathies. *Human Mutation*, **2014**, 35, 779-90	4.7	67
426	Neurologic examination of preterm infants at term age: comparison with term infants. *Journal of Pediatrics*, **2003**, 142, 647-55	3.6	67
425	Visual function in term infants with hypoxic-ischaemic insults: correlation with neurodevelopment at 2 years of age. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **1999**, 80, F99-104	4.7	64
424	The NorthStar Ambulatory Assessment in Duchenne muscular dystrophy: considerations for the design of clinical trials. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 149-55	5.5	63
423	Visual function in full-term infants with hypoxic-ischaemic encephalopathy. *Neuropediatrics*, **1997**, 28, 155-61	1.6	63
422	A short protocol for muscle MRI in children with muscular dystrophies. *European Journal of Paediatric Neurology*, **2002**, 6, 305-307	3.8	63
421	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. *Neurology*, **2012**, 79, 159-62	6.5	62
420	Developmental milestones in type I spinal muscular atrophy. *Neuromuscular Disorders*, **2016**, 26, 754-759	2.9	62

419	Cognitive profile of disorders associated with dysregulation of the RAS/MAPK signaling cascade. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 140-6	2.5	61
418	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, 152-63	5.5	60
417	SEPN1-related myopathies: clinical course in a large cohort of patients. *Neurology*, **2011**, 76, 2073-8	6.5	60
416	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	7.3	59
415	Autosomal recessive Bethlem myopathy. *Neurology*, **2009**, 73, 1883-91	6.5	59
414	Neurodevelopmental outcome at 12 and 18 months in late preterm infants. *European Journal of Paediatric Neurology*, **2010**, 14, 503-7	3.8	59
413	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, 602-9	3.4	59
412	Stakeholder cooperation to overcome challenges in orphan medicine development: the example of Duchenne muscular dystrophy. *Lancet Neurology, The*, **2016**, 15, 882-890	24.1	58
411	Application of the sleep disturbance scale for children (SDSC) in preschool age. *European Journal of Paediatric Neurology*, **2013**, 17, 374-82	3.8	58
410	Assessing upper limb function in nonambulant SMA patients: development of a new module. *Neuromuscular Disorders*, **2011**, 21, 406-12	2.9	58
409	Upper girdle imaging in facioscapulohumeral muscular dystrophy. *PLoS ONE*, **2014**, 9, e100292	3.7	58
408	Content validity and clinical meaningfulness of the HFMSE in spinal muscular atrophy. *BMC Neurology*, **2017**, 17, 39	3.1	56
407	6 Minute walk test in Duchenne MD patients with different mutations: 12 month changes. *PLoS ONE*, **2014**, 9, e83400	3.7	56
406	Somatosensory and visual evoked potentials in congenital muscular dystrophy: correlation with MRI changes and muscle merosin status. *Neuropediatrics*, **1995**, 26, 3-7	1.6	56
405	Fracture prevalence in Duchenne muscular dystrophy. *Developmental Medicine and Child Neurology*, **2002**, 44, 695-8	3.3	55
404	Visual function of prematurely born children with and without perceptual-motor difficulties. *Early Human Development*, **1996**, 45, 73-82	2.2	55
403	Risk Factors for Neonatal Arterial Ischemic Stroke: The Importance of the Intrapartum Period. *Journal of Pediatrics*, **2016**, 173, 62-68.e1	3.6	55
402	A critical review of functional assessment tools for upper limbs in Duchenne muscular dystrophy. *Developmental Medicine and Child Neurology*, **2012**, 54, 879-85	3.3	54

401	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, 856-8	5.8	54
400	Neuropsychological development in children with Dravet syndrome. *Epilepsy Research*, **2011**, 95, 86-93	3	54
399	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	3.6	54
398	Management of scoliosis in Duchenne muscular dystrophy: a large 10-year retrospective study. *Developmental Medicine and Child Neurology*, **2006**, 48, 513-8	3.3	54
397	Nusinersen in type 1 SMA infants, children and young adults: Preliminary results on motor function. *Neuromuscular Disorders*, **2018**, 28, 582-585	2.9	53
396	Sleep disorders in children with cerebral palsy: neurodevelopmental and behavioral correlates. *Sleep Medicine*, **2014**, 15, 213-8	4.6	53
395	Visual disorders in children with brain lesions: 2. Visual impairment associated with cerebral palsy. *European Journal of Paediatric Neurology*, **2001**, 5, 115-9	3.8	53
394	Evolution of unilateral perinatal arterial ischemic stroke on conventional and diffusion-weighted MR imaging. *American Journal of Neuroradiology*, **2009**, 30, 998-1004	4.4	52
393	Hand movements at 3 months predict later hemiplegia in term infants with neonatal cerebral infarction. *Developmental Medicine and Child Neurology*, **2010**, 52, 767-72	3.3	52
392	Swallowing difficulties in Duchenne muscular dystrophy: indications for feeding assessment and outcome of videofluroscopic swallow studies. *European Journal of Paediatric Neurology*, **2008**, 12, 239-45	3.8	51
391	Reliability of the Hammersmith functional motor scale for spinal muscular atrophy in a multicentric study. *Neuromuscular Disorders*, **2006**, 16, 93-8	2.9	51
390	Expanding the phenotype of potassium channelopathy: severe neuromyotonia and skeletal deformities without prominent Episodic Ataxia. *Neuromuscular Disorders*, **2004**, 14, 689-93	2.9	51
389	Predictive factors for the development of scoliosis in Duchenne muscular dystrophy. *European Journal of Paediatric Neurology*, **2007**, 11, 160-6	3.8	50
388	Muscle magnetic resonance imaging in patients with congenital muscular dystrophy and Ullrich phenotype. *Neuromuscular Disorders*, **2003**, 13, 554-8	2.9	50
387	Prevalence of congenital muscular dystrophy in Italy: a population study. *Neurology*, **2015**, 84, 904-11	6.5	49
386	Muscular dystrophy: new challenges and review of the current clinical trials. *Current Opinion in Pediatrics*, **2013**, 25, 701-7	3.2	49
385	Maturation of cerebral electrical activity and development of cortical folding in young very preterm infants. *Clinical Neurophysiology*, **2007**, 118, 53-9	4.3	49
384	Muscle MRI in Becker muscular dystrophy. *Neuromuscular Disorders*, **2012**, 22 Suppl 2, S100-6	2.9	48

383	Neurological examination of preterm infants at term equivalent age. *Early Human Development*, **2008**, 84, 751-61	2.2	48
382	A current approach to heart failure in Duchenne muscular dystrophy. *Heart*, **2017**, 103, 1770-1779	5.1	47
381	Relative frequency of congenital muscular dystrophy subtypes: analysis of the UK diagnostic service 2001-2008. *Neuromuscular Disorders*, **2012**, 22, 522-7	2.9	47
380	Pilot trial of salbutamol in central core and multi-minicore diseases. *Neuropediatrics*, **2004**, 35, 262-6	1.6	47
379	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	2.1	46
378	Rasch analysis of clinical outcome measures in spinal muscular atrophy. *Muscle and Nerve*, **2014**, 49, 422-30	3.4	46
377	Centronuclear myopathy related to dynamin 2 mutations: clinical, morphological, muscle imaging and genetic features of an Italian cohort. *Neuromuscular Disorders*, **2013**, 23, 229-38	2.9	45
376	Early neurodevelopmental assessment in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2013**, 23, 451-5	2.9	45
375	LMNA-associated myopathies: the Italian experience in a large cohort of patients. *Neurology*, **2014**, 83, 1634-44	6.5	45
374	Minicore myopathy in children: a clinical and histopathological study of 19 cases. *Neuromuscular Disorders*, **2000**, 10, 264-73	2.9	45
373	Ambulatory function in spinal muscular atrophy: Age-related patterns of progression. *PLoS ONE*, **2018**, 13, e0199657	3.7	45
372	Cortical vision, MRI and developmental outcome in preterm infants. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **2008**, 93, F292-7	4.7	44
371	Can the Griffiths scales predict neuromotor and perceptual-motor impairment in term infants with neonatal encephalopathy?. *Archives of Disease in Childhood*, **2004**, 89, 637-43	2.2	44
370	Therapeutic approaches for spinal muscular atrophy (SMA). *Gene Therapy*, **2017**, 24, 514-519	4	42
369	Nusinersen in type 1 spinal muscular atrophy: Twelve-month real-world data. *Annals of Neurology*, **2019**, 86, 443-451	9.4	42
368	Centronuclear myopathies: genotype-phenotype correlation and frequency of defined genetic forms in an Italian cohort. *Journal of Neurology*, **2015**, 262, 1728-40	5.5	42
367	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. *JAMA Neurology*, **2018**, 75, 557-565	17.2	42
366	Association Study of Exon Variants in the NF-B and TGF Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. *American Journal of Human Genetics*, **2016**, 99, 1163-1171	11	42

365	Nusinersen improves walking distance and reduces fatigue in later-onset spinal muscular atrophy. *Muscle and Nerve*, **2019**, 60, 409-414	3.4	42
364	Behavioral profile in RASopathies. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 934-42	2.5	42
363	Duchenne muscular dystrophy and epilepsy. *Neuromuscular Disorders*, **2013**, 23, 313-5	2.9	42
362	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	42
361	Visual function at 35 and 40 weeks' postmenstrual age in low-risk preterm infants. *Pediatrics*, **2008**, 122, e1193-8	7.4	42
360	Neonatal cerebral infarction and visual function at school age. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **2003**, 88, F487-91	4.7	42
359	MRI lesions and infants with neonatal encephalopathy. Is the Apgar score predictive?. *Neuropediatrics*, **2002**, 33, 150-6	1.6	42
358	MYH7-related myopathies: clinical, histopathological and imaging findings in a cohort of Italian patients. *Orphanet Journal of Rare Diseases*, **2016**, 11, 91	4.2	42
357	Visual function and perinatal focal cerebral infarction. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **1996**, 75, F76-81	4.7	41
356	Remission of clinical signs in early duchenne muscular dystrophy on intermittent low-dosage prednisolone therapy. *European Journal of Paediatric Neurology*, **2002**, 6, 153-9	3.8	40
355	Revised upper limb module for spinal muscular atrophy: 12 month changes. *Muscle and Nerve*, **2019**, 59, 426-430	3.4	39
354	The Hammersmith functional score correlates with the SMN2 copy number: a multicentric study. *Neuromuscular Disorders*, **2007**, 17, 400-3	2.9	39
353	Sequential study of central and peripheral nervous system involvement in an infant with merosin-deficient congenital muscular dystrophy. *Neuromuscular Disorders*, **1996**, 6, 425-9	2.9	39
352	Muscular dystrophies due to defective glycosylation of dystroglycan. *Acta Myologica*, **2007**, 26, 129-35	1.6	39
351	Adiposity is increased among high-functioning, non-ambulatory patients with spinal muscular atrophy. *Neuromuscular Disorders*, **2010**, 20, 448-52	2.9	38
350	Revised Hammersmith Scale for spinal muscular atrophy: A SMA specific clinical outcome assessment tool. *PLoS ONE*, **2017**, 12, e0172346	3.7	37
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