Eugenio Mercuri

List of Publications by Year in Descending Order

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	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	24.1	9
525	Restoration of Nusinersen Levels Following Treatment Interruption in People With Spinal Muscular Atrophy: Simulations Based on a Population Pharmacokinetic Model.. *CNS Drugs*, **2022**, 1	6.7	0
524	Body mass index in type 2 spinal muscular atrophy: a longitudinal study.. *European Journal of Pediatrics*, **2022**, 1	4.1	0
523	Hammersmith Infant Neurological Examination in infants born at term: Predicting outcomes other than cerebral palsy.. *Developmental Medicine and Child Neurology*, **2022**,	3.3	1
522	Hammersmith Infant Neurological Examination in low-risk infants born very preterm: a longitudinal prospective study.. *Developmental Medicine and Child Neurology*, **2022**,	3.3	1
521	Quantitative magnetic resonance imaging measures as biomarkers of disease progression in boys with Duchenne muscular dystrophy: a phase 2 trial of domagrozumab.. *Journal of Neurology*, **2022**, 1	5.5	0
520	Expanding the clinical-pathological and genetic spectrum of RYR1-related congenital myopathies with cores and minicores: an Italian population study.. *Acta Neuropathologica Communications*, **2022** , 10, 54	7.3	1
519	Assessing floppy infants: a new module.. *European Journal of Pediatrics*, **2022**, 1	4.1	1
518	Neurological assessment of newborns with spinal muscular atrophy identified through neonatal screening.. *European Journal of Pediatrics*, **2022**, 1	4.1	2
517	Genetic modifiers of upper limb function in Duchenne muscular dystrophy.. *Journal of Neurology*, **2022**, 1	5.5	1
516	Predictive models in SMA II natural history trajectories using machine learning: A proof of concept study.. *PLoS ONE*, **2022**, 17, e0267930	3.7	0
515	Cortical Thickness and Clinical Findings in Prescholar Children With Autism Spectrum Disorder.. *Frontiers in Neuroscience*, **2021**, 15, 776860	5.1	0
514	Spinal muscular atrophy: from rags to riches. *Neuromuscular Disorders*, **2021**, 31, 998-1003	2.9	1
513	Ataluren delays loss of ambulation and respiratory decline in nonsense mutation Duchenne muscular dystrophy patients. *Journal of Comparative Effectiveness Research*, **2021**,	2.1	6
512	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*, **2021**,	4.8	8
511	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	4.2	3
510	Hammersmith Infant Neurological Examination for infants born preterm: predicting outcomes other than cerebral palsy. *Developmental Medicine and Child Neurology*, **2021**, 63, 939-946	3.3	13

509	Risdiplam in Type 1 Spinal Muscular Atrophy. *New England Journal of Medicine*, **2021**, 384, 915-923	59.2	69
508	Multicenter prospective longitudinal study in 34 patients with Dravet syndrome: Neuropsychological development in the first six years of life. *Brain and Development*, **2021**, 43, 419-430	2.2	3
507	International retrospective natural history study of -related congenital muscular dystrophy. *Brain Communications*, **2021**, 3, fcab075	4.5	2
506	Longitudinal data of neuropsychological profile in a cohort of Duchenne muscular dystrophy boys without cognitive impairment. *Neuromuscular Disorders*, **2021**, 31, 319-327	2.9	5
505	Population pharmacokinetics-based recommendations for a single delayed or missed dose of nusinersen. *Neuromuscular Disorders*, **2021**, 31, 310-318	2.9	1
504	Spinal muscular atrophy: state of the art and new therapeutic strategies. *Neurological Sciences*, **2021**, 1	3.5	5
503	Reader Response: Nusinersen in Adult Patients With Spinal Muscular Atrophy: Observations From a Single Center. *Neurology*, **2021**, 96, 1061.2-1062	6.5	
502	Application of the Sleep Disturbance Scale for Children (SDSC) in infants and toddlers (6-36 months). *Sleep Medicine*, **2021**, 81, 62-68	4.6	2
501	Neural substrates of neuropsychological profiles in dystrophynopathies: A pilot study of diffusion tractography imaging. *PLoS ONE*, **2021**, 16, e0250420	3.7	0
500	Visual Function and Ophthalmological Findings in CHARGE Syndrome: Revision of Literature, Definition of a New Clinical Spectrum and Genotype Phenotype Correlation. *Genes*, **2021**, 12,	4.2	3
499	Nusinersen in pediatric and adult patients with type III spinal muscular atrophy. *Annals of Clinical and Translational Neurology*, **2021**, 8, 1622-1634	5.3	6
498	North Star Ambulatory Assessment changes in ambulant Duchenne boys amenable to skip exons 44, 45, 51, and 53: A 3 year follow up. *PLoS ONE*, **2021**, 16, e0253882	3.7	1
497	The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2021**, 31, 479-488	2.9	
496	Open-Label Evaluation of Eteplirsen in Patients with Duchenne Muscular Dystrophy Amenable to Exon 51 Skipping: PROMOVI Trial. *Journal of Neuromuscular Diseases*, **2021**, 8, 989-1001	5	13
495	Adeno-associated virus serotype 9 antibodies in patients screened for treatment with onasemnogene abeparvovec. *Molecular Therapy - Methods and Clinical Development*, **2021**, 21, 76-82	6.4	9
494	Circadian Genes as Exploratory Biomarkers in DMD: Results From Both the Mouse Model and Patients. *Frontiers in Physiology*, **2021**, 12, 678974	4.6	0
493	Oral and Swallowing Abilities Tool (OrSAT) for Type 1 SMA Patients: Development of a New Module. *Journal of Neuromuscular Diseases*, **2021**, 8, 589-601	5	2
492	Sometimes they come back: New and old spinal muscular atrophy adults in the era of nusinersen. *European Journal of Neurology*, **2021**, 28, 602-608	6	3

491	Respiratory Trajectories in Type 2 and 3 Spinal Muscular Atrophy in the iSMAC Cohort Study. *Neurology*, **2021**, 96, e587-e599	6.5	12
490	Risdiplam treatment has not led to retinal toxicity in patients with spinal muscular atrophy. *Annals of Clinical and Translational Neurology*, **2021**, 8, 54-65	5.3	5
489	Response to letter: A decision for life - Treatment decisions in newly diagnosed families with spinal muscular atrophy. *European Journal of Paediatric Neurology*, **2021**, 30, 103-104	3.8	
488	Expanding the spectrum of congenital myopathies: prenatal onset with extreme hyperextension of the neck. *Neurological Sciences*, **2021**, 42, 1549-1553	3.5	0
487	The administration of antisense oligonucleotide golodirsen reduces pathological regeneration in patients with Duchenne muscular dystrophy. *Acta Neuropathologica Communications*, **2021**, 9, 7	7.3	8
486	Type I SMA "new natural history": long-term data in nusinersen-treated patients. *Annals of Clinical and Translational Neurology*, **2021**, 8, 548-557	5.3	10
485	Contactless: a new personalised telehealth model in chronic pediatric diseases and disability during the COVID-19 era. *Italian Journal of Pediatrics*, **2021**, 47, 29	3.2	4
484	Duchenne muscular dystrophy. *Nature Reviews Disease Primers*, **2021**, 7, 13	51.1	87
483	The Spinal Muscular Atrophy Health Index: Italian validation of a disease-specific outcome measure. *Neuromuscular Disorders*, **2021**, 31, 409-418	2.9	2
482	Comparison of Long-term Ambulatory Function in Patients with Duchenne Muscular Dystrophy Treated with Eteplirsen and Matched Natural History Controls. *Journal of Neuromuscular Diseases*, **2021**, 8, 469-479	5	8
481	Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. *Neuromuscular Disorders*, **2021**, 31, 596-602	2.9	9
480	Extra-uterine growth restriction in preterm infants: Neurodevelopmental outcomes according to different definitions. *European Journal of Paediatric Neurology*, **2021**, 33, 135-145	3.8	4
479	Early Gross Motor Milestones in Duchenne Muscular Dystrophy. *Journal of Neuromuscular Diseases*, **2021**, 8, 453-456	5	0
478	Ultrasound assisted lumbar intrathecal administration of nusinersen in adult patients with spinal muscular atrophy: A case series. *Muscle and Nerve*, **2021**, 64, 594-599	3.4	0
477	A Combined Prospective and Retrospective Comparison of Long-Term Functional Outcomes Suggests Delayed Loss of Ambulation and Pulmonary Decline with Long-Term Eteplirsen Treatment. *Journal of Neuromuscular Diseases*, **2021**,	5	1
476	Clinical Trial and Postmarketing Safety of Onasemnogene Abeparvovec Therapy. *Drug Safety*, **2021**, 44, 1109-1119	5.1	5
475	Different trajectories in upper limb and gross motor function in spinal muscular atrophy. *Muscle and Nerve*, **2021**, 64, 552-559	3.4	4
474	Reply to: The need for evidence-based treatment decisions in spinal muscular atrophy type 0. *Annals of Clinical and Translational Neurology*, **2021**, 8, 2093	5.3	

473	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	24.1	24
472	Sleep Disorders in Autism Spectrum Disorder Pre-School Children: An Evaluation Using the Sleep Disturbance Scale for Children. *Medicina (Lithuania)*, **2021**, 57,	3.1	3
471	The social smile in infants during the COVID-19 pandemia.. *Heliyon*, **2021**, 7, e08648	3.6	
470	Nusinersen in type 0 spinal muscular atrophy: should we treat?. *Annals of Clinical and Translational Neurology*, **2020**, 7, 2481-2483	5.3	4
469	Long-term follow-up of patients with type 2 and non-ambulant type 3 spinal muscular atrophy (SMA) treated with olesoxime in the OLEOS trial. *Neuromuscular Disorders*, **2020**, 30, 959-969	2.9	4
468	Ultrasound assessment of diaphragmatic function in type 1 spinal muscular atrophy. *Pediatric Pulmonology*, **2020**, 55, 1781-1788	3.5	6
467	Spinal cord demyelination in children: A diagnostic challenge in neuropaediatrics for a good outcome. *Brain and Development*, **2020**, 42, 457-461	2.2	2
466	Randomized phase 2 trial and open-label extension of domagrozumab in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2020**, 30, 492-502	2.9	20
465	Patient and parent oriented tools to assess health-related quality of life, activity of daily living and caregiver burden in SMA. Rome, 13 July 2019. *Neuromuscular Disorders*, **2020**, 30, 431-436	2.9	5
464	Measuring Outcomes in Adults with Spinal Muscular Atrophy - Challenges and Future Directions - Meeting Report. *Journal of Neuromuscular Diseases*, **2020**, 7, 523-534	5	16
463	The Genetic Landscape of Dystrophin Mutations in Italy: A Nationwide Study. *Frontiers in Genetics*, **2020**, 11, 131	4.5	25
462	Early Childhood Attention Battery: Italian adaptation and new expanded normative data. *Early Human Development*, **2020**, 144, 105013	2.2	1
461	Nusinersen in adults with spinal muscular atrophy: new challenges. *Lancet Neurology, The*, **2020**, 19, 283-284	24.1	10
460	Diagnostic journey in Spinal Muscular Atrophy: Is it still an odyssey?. *PLoS ONE*, **2020**, 15, e0230677	3.7	15
459	Increased dystrophin production with golodirsen in patients with Duchenne muscular dystrophy. *Neurology*, **2020**, 94, e2270-e2282	6.5	106
458	Pediatric Motor Inflammatory Neuropathy: The Role of Antiphospholipid Antibodies. *Brain Sciences*, **2020**, 10,	3.4	
457	RESTORE: A Prospective Multinational Registry of Patients with Genetically Confirmed Spinal Muscular Atrophy - Rationale and Study Design. *Journal of Neuromuscular Diseases*, **2020**, 7, 145-152	5	7
456	Suitability of external controls for drug evaluation in Duchenne muscular dystrophy. *Neurology*, **2020**, 95, e1381-e1391	6.5	12

455	European ad-hoc consensus statement on gene replacement therapy for spinal muscular atrophy. *European Journal of Paediatric Neurology*, **2020**, 28, 38-43	3.8	35
454	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. *Frontiers in Genetics*, **2020**, 11, 605	4.5	3
453	Respiratory Needs in Patients with Type 1 Spinal Muscular Atrophy Treated with Nusinersen. *Journal of Pediatrics*, **2020**, 219, 223-228.e4	3.6	25
452	Cranial ultrasound evaluation in term neonates. *Early Human Development*, **2020**, 143, 104983	2.2	3
451	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
450	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. *Annals of Clinical and Translational Neurology*, **2020**, 7, 786-798	5.3	14
449	Spinal muscular atrophy care in the COVID-19 pandemic era. *Muscle and Nerve*, **2020**, 62, 46-49	3.4	22
448	Hypoglycaemia in patients with type 1 SMA: an underdiagnosed problem?. *Archives of Disease in Childhood*, **2020**, 105, 707	2.2	2
447	Respiratory function and therapeutic expectations in DMD: families experience and perspective. *Acta Myologica*, **2020**, 39, 121-129	1.6	
446	Estimating the impact of COVID-19 pandemic on services provided by Italian Neuromuscular Centers: an Italian Association of Myology survey of the acute phase. *Acta Myologica*, **2020**, 39, 57-66	1.6	15
445	Functional and Morphologic Findings at Four Years After Intravitreal Bevacizumab or Laser for Type 1 ROP. *Ophthalmic Surgery Lasers and Imaging Retina*, **2020**, 51, 180-186	1.4	1
444	De Novo Partial 13q22-q34 Trisomy with Typical Neurological and Immunological Findings: A Case Report with New Genetic Insights. *Brain Sciences*, **2020**, 11,	3.4	0
443	Performance of Upper Limb module for Duchenne muscular dystrophy. *Developmental Medicine and Child Neurology*, **2020**, 62, 633-639	3.3	17
442	Long-term data with idebenone on respiratory function outcomes in patients with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2020**, 30, 5-16	2.9	18
441	MRI patterns of muscle involvement in type 2 and 3 spinal muscular atrophy patients. *Journal of Neurology*, **2020**, 267, 898-912	5.5	17
440	SMA THERAPY. *Neuromuscular Disorders*, **2020**, 30, S121	2.9	2
439	SMA THERAPY. *Neuromuscular Disorders*, **2020**, 30, S123-S124	2.9	2
438	A Recurrent Pathogenic Variant of Underlies Autosomal Recessive Congenital Muscular Dystrophy With Cataracts and Intellectual Disability: Evidence for a Founder Effect in Southern Italy. *Frontiers in Genetics*, **2020**, 11, 565868	4.5	2

437	Spinal muscular atrophy - insights and challenges in the treatment era. *Nature Reviews Neurology*, **2020**, 16, 706-715	15	22
436	Children's Healthcare During Corona Virus Disease 19 Pandemic: the Italian Experience. *Pediatric Infectious Disease Journal*, **2020**, 39, e137-e140	3.4	10
435	Genotype-phenotype correlations in recessive titinopathies. *Genetics in Medicine*, **2020**, 22, 2029-2040	8.1	12
434	Gain and loss of abilities in type II SMA: A 12-month natural history study. *Neuromuscular Disorders*, **2020**, 30, 765-771	2.9	8
433	Early visual and neuro-development in preterm infants with and without retinopathy. *Early Human Development*, **2020**, 148, 105134	2.2	2
432	Age and baseline values predict 12 and 24-month functional changes in type 2 SMA. *Neuromuscular Disorders*, **2020**, 30, 756-764	2.9	13
431	Early Neurological Assessment and Long-Term Neuromotor Outcomes in Late Preterm Infants: A Critical Review. *Medicina (Lithuania)*, **2020**, 56,	3.1	2
430	Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy. *Journal of Comparative Effectiveness Research*, **2020**, 9, 973-984	2.1	19
429	Clinical Variability in Spinal Muscular Atrophy Type III. *Annals of Neurology*, **2020**, 88, 1109-1117	9.4	14
428	Visual Function Classification System for children with cerebral palsy: development and validation. *Developmental Medicine and Child Neurology*, **2020**, 62, 104-110	3.3	19
427	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). *Journal of Neurology*, **2020**, 267, 45-56	5.5	19
426	Longitudinal natural history of type I spinal muscular atrophy: a critical review. *Orphanet Journal of Rare Diseases*, **2020**, 15, 84	4.2	18
425	Early Neurological Assessment in Infants with Hypoxic Ischemic Encephalopathy Treated with Therapeutic Hypothermia. *Journal of Clinical Medicine*, **2019**, 8,	5.1	4
424	Long-term progression in type II spinal muscular atrophy: A retrospective observational study. *Neurology*, **2019**, 93, e1241-e1247	6.5	19
423	Predominant distal muscle involvement in spinal muscular atrophy. *Neuromuscular Disorders*, **2019**, 29, 910-911	2.9	2
422	MYO-MRI diagnostic protocols in genetic myopathies. *Neuromuscular Disorders*, **2019**, 29, 827-841	2.9	23
421	Revised upper limb module for spinal muscular atrophy: 12 month changes. *Muscle and Nerve*, **2019**, 59, 426-430	3.4	39
420	Clinical features and genetic analysis of two siblings with startle disease in an Italian family: a case report. *BMC Medical Genetics*, **2019**, 20, 40	2.1	2

419	Nusinersen in type 1 spinal muscular atrophy: Twelve-month real-world data. *Annals of Neurology*, **2019**, 86, 443-451	9.4	42
418	Long-term natural history data in Duchenne muscular dystrophy ambulant patients with mutations amenable to skip exons 44, 45, 51 and 53. *PLoS ONE*, **2019**, 14, e0218683	3.7	21
417	Sleep disorders in low-risk preschool very preterm children. *Sleep Medicine*, **2019**, 63, 137-141	4.6	4
416	Neurofilament as a potential biomarker for spinal muscular atrophy. *Annals of Clinical and Translational Neurology*, **2019**, 6, 932-944	5.3	76
415	An unusual ryanodine receptor 1 (RYR1) phenotype: Mild calf-predominant myopathy. *Neurology*, **2019**, 92, e1600-e1609	6.5	10
414	Value of structured reporting in neuromuscular disorders. *Radiologia Medica*, **2019**, 124, 628-635	6.5	6
413	Brain morphometry of preschool age children affected by autism spectrum disorder: Correlation with clinical findings. *Clinical Anatomy*, **2019**, 32, 143-150	2.5	6
412	Ataluren use in patients with nonsense mutation Duchenne muscular dystrophy: patient demographics and characteristics from the STRIDE Registry. *Journal of Comparative Effectiveness Research*, **2019**, 8, 1187-1200	2.1	21
411	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	6.7	32
410	Neonatal hypotonia and neuromuscular conditions. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2019**, 162, 435-448	3	3
409	Nusinersen improves walking distance and reduces fatigue in later-onset spinal muscular atrophy. *Muscle and Nerve*, **2019**, 60, 409-414	3.4	42
408	P.063 SUNFISH Part 1 results and Part 2 trial design in patients with type 2/3 spinal muscular atrophy (SMA) receiving risdiplam (RG7916). *Canadian Journal of Neurological Sciences*, **2019**, 46, S31	1	
407	P.223Respiratory function in SMA type 2 and non-ambulant SMA type 3, longitudinal data from the international SMA consortium (iSMAc). *Neuromuscular Disorders*, **2019**, 29, S131-S132	2.9	2
406	P.353FIREFISH Part 1: 16-month safety and exploratory outcomes of risdiplam (RG7916) treatment in infants with type 1 spinal muscular atrophy. *Neuromuscular Disorders*, **2019**, 29, S184	2.9	8
405	O.41Sunfish part 1: 18-month safety and exploratory outcomes of risdiplam (RG7916) treatment in patients with type 2 or 3 spinal muscular atrophy. *Neuromuscular Disorders*, **2019**, 29, S208	2.9	4
404	P.363JEWELFISH: safety and pharmacodynamic data in patients with spinal muscular atrophy (SMA) receiving treatment with risdiplam (RG7916) that have previously been treated with nusinersen. *Neuromuscular Disorders*, **2019**, 29, S187	2.9	3
403	Development of an academic disease registry for spinal muscular atrophy. *Neuromuscular Disorders*, **2019**, 29, 794-799	2.9	17
402	Longitudinal natural history in young boys with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2019**, 29, 857-862	2.9	12

401	Il profilo dei deficit neuropsicologici cognitivi della malattia di Duchenne: uno studio empirico controllato e le sue ricadute psicoter apiche. *Quaderni Di Psicoterapia Cognitiva*, **2019**, 40-55	0.5	
400	Resolution of skin necrosis after nusinersen treatment in an infant with spinal muscular atrophy. *Muscle and Nerve*, **2019**, 59, E42-E44	3.4	4
399	A critical review of patient and parent caregiver oriented tools to assess health-related quality of life, activity of daily living and caregiver burden in spinal muscular atrophy. *Neuromuscular Disorders* , **2019**, 29, 940-950	2.9	13
398	Muscular dystrophies. *Lancet, The*, **2019**, 394, 2025-2038	40	99
397	Next-generation sequencing approach to hyperCKemia: A 2-year cohort study. *Neurology: Genetics*, **2019**, 5, e352	3.8	12
396	Longitudinal evaluation of SMN levels as biomarker for spinal muscular atrophy: results of a phase IIb double-blind study of salbutamol. *Journal of Medical Genetics*, **2019**, 56, 293-300	5.8	16
395	Intrathecal nusinersen treatment for SMA in a dedicated neuromuscular clinic: an example of multidisciplinary and integrated care. *Neurological Sciences*, **2019**, 40, 327-332	3.5	12
394	The oral splicing modifier RG7800 increases full length survival of motor neuron 2 mRNA and survival of motor neuron protein: Results from trials in healthy adults and patients with spinal muscular atrophy. *Neuromuscular Disorders*, **2019**, 29, 21-29	2.9	15
393	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
392	Joint Laxity in Preschool Children Born Preterm. *Journal of Pediatrics*, **2018**, 197, 104-108	3.6	4
391	Evidence-based care in Duchenne muscular dystrophy. *Lancet Neurology, The*, **2018**, 17, 389-391	24.1	9
390	A 5-center experience with intrathecal administration of nusinersen in SMA1 in Italy letter to the editor of european journal of pediatric neurology regarding the manuscript "single-center experience with intrathecal administration of nusinersen in children with spinal muscular atrophy type 1" written by pechmann and colleagues". *European Journal of Paediatric Neurology*, **2018**, 22, 729-731	3.8	3
389	Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. *New England Journal of Medicine*, **2018**, 378, 625-635	59.2	617
388	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. *JAMA Neurology*, **2018**, 75, 557-565	17.2	42
387	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
386	MRI in sarcoglycanopathies: a large international cohort study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 72-77	5.5	34
385	Early involvement of the supinator muscle in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2018**, 28, 62-63	2.9	3
384	Clinical phenotypes and trajectories of disease progression in type 1 spinal muscular atrophy. *Neuromuscular Disorders*, **2018**, 28, 24-28	2.9	30

383	Upper limb function in Duchenne muscular dystrophy: 24 month longitudinal data. *PLoS ONE*, **2018**, 13, e0199223	3.7	28
382	Does albuterol have an effect on neuromuscular junction dysfunction in spinal muscular atrophy?. *Neuromuscular Disorders*, **2018**, 28, 863-864	2.9	6
381	An observational study of functional abilities in infants, children, and adults with type 1 SMA. *Neurology*, **2018**, 91, e696-e703	6.5	12
380	Quantitative Evaluation of Lower Extremity Joint Contractures in Spinal Muscular Atrophy: Implications for Motor Function. *Pediatric Physical Therapy*, **2018**, 30, 209-215	0.9	10
379	Effects of Lycra suits in children with cerebral palsy. *European Journal of Paediatric Neurology*, **2018**, 22, 831-836	3.8	3
378	Nusinersen in type 1 SMA infants, children and young adults: Preliminary results on motor function. *Neuromuscular Disorders*, **2018**, 28, 582-585	2.9	53
377	Implicit learning deficit in children with Duchenne muscular dystrophy: Evidence for a cerebellar cognitive impairment?. *PLoS ONE*, **2018**, 13, e0191164	3.7	13
376	Evaluator Training and Reliability for SMA Global Nusinersen Trials1. *Journal of Neuromuscular Diseases*, **2018**, 5, 159-166	5	17
375	Functional levels and MRI patterns of muscle involvement in upper limbs in Duchenne muscular dystrophy. *PLoS ONE*, **2018**, 13, e0199222	3.7	12
374	Early Diagnosis and Treatment The Use of Ataluren in the Effective Management of Duchenne Muscular Dystrophy. *European Neurological Review*, **2018**, 13, 31	0.5	1
373	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
372	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
371	Cognitive profile in Duchenne muscular dystrophy boys without intellectual disability: The role of executive functions. *Neuromuscular Disorders*, **2018**, 28, 122-128	2.9	20
370	Neuromuscular disorders: 2017, a year to remember. *Lancet Neurology, The*, **2018**, 17, 12-13	24.1	0
369	Broad phenotypic spectrum and genotype-phenotype correlations in GMPPB-related dystroglycanopathies: an Italian cross-sectional study. *Orphanet Journal of Rare Diseases*, **2018**, 13, 170	4.2	21
368	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. *Genes*, **2018**, 9,	4.2	4
367	Ambulatory function in spinal muscular atrophy: Age-related patterns of progression. *PLoS ONE*, **2018**, 13, e0199657	3.7	45
366	Genetic therapies for inherited neuromuscular disorders. *The Lancet Child and Adolescent Health*, **2018**, 2, 600-609	14.5	19

365	Sleep disorders in spinal muscular atrophy. *Sleep Medicine*, **2017**, 30, 160-163	4.6	12
364	Diagnosis of Duchenne Muscular Dystrophy in Italy in the last decade: Critical issues and areas for improvements. *Neuromuscular Disorders*, **2017**, 27, 447-451	2.9	24
363	218th ENMC International Workshop:: Revisiting the consensus on standards of care in SMA Naarden, The Netherlands, 19-21 February 2016. *Neuromuscular Disorders*, **2017**, 27, 596-605	2.9	32
362	Content validity and clinical meaningfulness of the HFMSE in spinal muscular atrophy. *BMC Neurology*, **2017**, 17, 39	3.1	56
361	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
360	Recessive mutations in MSTO1 cause mitochondrial dynamics impairment, leading to myopathy and ataxia. *Human Mutation*, **2017**, 38, 970-977	4.7	33
359	Therapeutic approaches for spinal muscular atrophy (SMA). *Gene Therapy*, **2017**, 24, 514-519	4	42
358	Neonatal neurological examination during the first 6h after birth. *Early Human Development*, **2017**, 108, 41-44	2.2	7
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