Eugenio Mercuri

List of Publications by Year
in descending order

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602 papers	33,331 citations	3531 90 h-index	7518 151 g-index
621 all docs	621 docs citations	621 times ranked	18996 citing authors

#	Article	IF	Citations
1	Risdiplam in types 2 and 3 spinal muscular atrophy: A randomised, placeboâ€controlled, doseâ€finding trial followed by 24â€‰months of treatment. European Journal of Neurology, 2023, 30, 1945-1956.	3.3	23
2	Spinal muscular atrophy: state of the art and new therapeutic strategies. Neurological Sciences, 2022, 43, 615-624.	1.9	13
3	Focus on the road to modelling cardiomyopathy in muscular dystrophy. Cardiovascular Research, 2022, 118, 1872-1884.	3.8	1
4	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, 2022, 9, 39-52.	2.6	24
5	Revised upper limb module in type II and III spinal muscular atrophy: 24-month changes. Neuromuscular Disorders, 2022, 32, 36-42.	0.6	13
6	Ataluren delays loss of ambulation and respiratory decline in nonsense mutation Duchenne muscular dystrophy patients. Journal of Comparative Effectiveness Research, 2022, 11, 139-155.	1.4	29
7	Muscle â€œislandsâ€: An MRI signature distinguishing neurogenic from myopathic causes of early onset distal weakness. Neuromuscular Disorders, 2022, 32, 142-149.	0.6	6
8	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.	3.6	58
9	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.	10.2	89
10	Restoration of Nusinersen Levels Following Treatment Interruption in People With Spinal Muscular Atrophy: Simulations Based on a Population Pharmacokinetic Model. CNS Drugs, 2022, 36, 181-190.	5.9	6
11	Longitudinal Cognitive Assessment in Low-Risk Very Preterm Infants. Medicina (Lithuania), 2022, 58, 133.	2.0	4
12	Body mass index in type 2 spinal muscular atrophy: a longitudinal study. European Journal of Pediatrics, 2022, , 1.	2.7	2
13	Real-world and natural history data for drug evaluation in Duchenne muscular dystrophy: suitability of the North Star Ambulatory Assessment for comparisons with external controls. Neuromuscular Disorders, 2022, 32, 271-283.	0.6	13
14	Hammersmith Infant Neurological Examination in infants born at term: Predicting outcomes other than cerebral palsy. Developmental Medicine and Child Neurology, 2022, 64, 871-880.	2.1	12
15	Nusinersen efficacy data for 24â€month in type 2 and 3 spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2022, 9, 404-409.	3.7	22
16	Hammersmith Infant Neurological Examination in lowâ€risk infants born very preterm: a longitudinal prospective study. Developmental Medicine and Child Neurology, 2022, 64, 863-870.	2.1	11
17	Muscle-MRI and Functional Levels for the Evaluation of Upper Limbs in Duchenne Muscular Dystrophy: A Critical Review of the Literature. Medicina (Lithuania), 2022, 58, 440.	2.0	1
18	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, 269, 4421-4435.	3.6	6

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19	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.	5.2	3
20	Assessing floppy infants: a new module. European Journal of Pediatrics, 2022, 181, 2771-2778.	2.7	5
21	Neurological assessment of newborns with spinal muscular atrophy identified through neonatal screening. European Journal of Pediatrics, 2022, 181, 2821-2829.	2.7	17
22	Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894.	3.6	2
23	Predictive models in SMA II natural history trajectories using machine learning: A proof of concept study. PLoS ONE, 2022, 17, e0267930.	2.5	2
24	Scientific rationale for a higher dose of nusinersen. Annals of Clinical and Translational Neurology, 2022, 9, 819-829.	3.7	9
25	Oral and Swallowing Abilities Tool (OrSAT) in nusinersen treated patients. Archives of Disease in Childhood, 2022, 107, 912-916.	1.9	3
26	Pregnancy-related psychopathology: A comparison between pre-COVID-19 and COVID-19â€“related social restriction periods. World Journal of Clinical Cases, 2022, 10, 6370-6384.	0.8	3
27	Developmental Coordination Disorder and Joint Hypermobility in Childhood: A Narrative Review. Children, 2022, 9, 1011.	1.5	3
28	Hammersmith Infant Neurological Examination for infants born preterm: predicting outcomes other than cerebral palsy. Developmental Medicine and Child Neurology, 2021, 63, 939-946.	2.1	32
29	Sometimes they come back: New and old spinal muscular atrophy adults in the era of nusinersen. European Journal of Neurology, 2021, 28, 602-608.	3.3	9
30	Respiratory Trajectories in Type 2 and 3 Spinal Muscular Atrophy in the iSMAC Cohort Study. Neurology, 2021, 96, e587-e599.	1.1	36
31	Risdiplam treatment has not led to retinal toxicity in patients with spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2021, 8, 54-65.	3.7	28
32	Response to letter: A decision for life â€“ Treatment decisions in newly diagnosed families with spinal muscular atrophy. European Journal of Paediatric Neurology, 2021, 30, 103-104.	1.6	1
33	Expanding the spectrum of congenital myopathies: prenatal onset with extreme hyperextension of the neck. Neurological Sciences, 2021, 42, 1549-1553.	1.9	2
34	The administration of antisense oligonucleotide golodirsen reduces pathological regeneration in patients with Duchenne muscular dystrophy. Acta Neuropathologica Communications, 2021, 9, 7.	5.2	24
35	Type I SMA â€œnew natural historyâ€: longâ€term data in nusinersenâ€treated patients. Annals of Clinical and Translational Neurology, 2021, 8, 548-557.	3.7	35
36	Contactless: a new personalised telehealth model in chronic pediatric diseases and disability during the COVID-19 era. Italian Journal of Pediatrics, 2021, 47, 29.	2.6	13

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37	Duchenne muscular dystrophy. Nature Reviews Disease Primers, 2021, 7, 13.	30.5	448
38	The Spinal Muscular Atrophy Health Index: Italian validation of a disease-specific outcome measure. Neuromuscular Disorders, 2021, 31, 409-418.	0.6	7
39	Risdiplam in Type 1 Spinal Muscular Atrophy. New England Journal of Medicine, 2021, 384, 915-923.	27.0	229
40	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.	1.1	7
41	International retrospective natural history study of LMNA-related congenital muscular dystrophy. Brain Communications, 2021, 3, fcab075.	3.3	17
42	Longitudinal data of neuropsychological profile in a cohort of Duchenne muscular dystrophy boys without cognitive impairment. Neuromuscular Disorders, 2021, 31, 319-327.	0.6	13
43	Population pharmacokinetics-based recommendations for a single delayed or missed dose of nusinersen. Neuromuscular Disorders, 2021, 31, 310-318.	0.6	10
44	Reader Response: Nusinersen in Adult Patients With Spinal Muscular Atrophy: Observations From a Single Center. Neurology, 2021, 96, 1061-1062.	1.1	0
45	Application of the Sleep Disturbance Scale for Children (SDSC) in infants and toddlers (6â€“36 months). Sleep Medicine, 2021, 81, 62-68.	1.6	16
46	Neural substrates of neuropsychological profiles in dystrophynopathies: A pilot study of diffusion tractography imaging. PLoS ONE, 2021, 16, e0250420.	2.5	4
47	Visual Function and Ophthalmological Findings in CHARGE Syndrome: Revision of Literature, Definition of a New Clinical Spectrum and Genotype Phenotype Correlation. Genes, 2021, 12, 972.	2.4	12
48	Nusinersen in pediatric and adult patients with type III spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2021, 8, 1622-1634.	3.7	27
49	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.	2.5	6
50	The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 479-488.	0.6	0
51	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.	2.6	50
52	Adeno-associated virus serotype 9 antibodies in patients screened for treatment with onasemnogene abeparvovec. Molecular Therapy - Methods and Clinical Development, 2021, 21, 76-82.	4.1	24
53	Circadian Genes as Exploratory Biomarkers in DMD: Results From Both the mdx Mouse Model and Patients. Frontiers in Physiology, 2021, 12, 678974.	2.8	1
54	Oral and Swallowing Abilities Tool (OrSAT) for Type 1 SMA Patients: Development of a New Module. Journal of Neuromuscular Diseases, 2021, 8, 589-601.	2.6	16

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55	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.	2.6	22
56	Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. Neuromuscular Disorders, 2021, 31, 596-602.	0.6	29
57	Extra-uterine growth restriction in preterm infants: Neurodevelopmental outcomes according to different definitions. European Journal of Paediatric Neurology, 2021, 33, 135-145.	1.6	19
58	Early Gross Motor Milestones in Duchenne Muscular Dystrophy. Journal of Neuromuscular Diseases, 2021, 8, 453-456.	2.6	2
59	Ultrasound assisted lumbar intrathecal administration of nusinersen in adult patients with spinal muscular atrophy: A case series. Muscle and Nerve, 2021, 64, 594-599.	2.2	3
60	Clinical Trial and Postmarketing Safety of Onasemnogene Abeparvovec Therapy. Drug Safety, 2021, 44, 1109-1119.	3.2	62
61	Different trajectories in upper limb and gross motor function in spinal muscular atrophy. Muscle and Nerve, 2021, 64, 552-559.	2.2	18
62	Reply to: The need for evidenceâ€based treatment decisions in spinal muscular atrophy type 0. Annals of Clinical and Translational Neurology, 2021, 8, 2093.	3.7	0
63	SMA-miRs (miR-181a-5p, -324-5p, and -451a) are overexpressed in spinal muscular atrophy skeletal muscle and serum samples. ELife, 2021, 10, .	6.0	13
64	SMA - TREATMENT. Neuromuscular Disorders, 2021, 31, S138-S139.	0.6	0
65	SMA - TREATMENT. Neuromuscular Disorders, 2021, 31, S136.	0.6	0
66	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.	10.2	112
67	DMD - TREATMENT. Neuromuscular Disorders, 2021, 31, S94-S95.	0.6	1
68	DMD/BMD â€“ OUTCOME MEASURES. Neuromuscular Disorders, 2021, 31, S87.	0.6	1
69	Sleep Disorders in Autism Spectrum Disorder Pre-School Children: An Evaluation Using the Sleep Disturbance Scale for Children. Medicina (Lithuania), 2021, 57, 95.	2.0	12
70	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.	2.7	58
71	De Novo Partial 13q22-q34 Trisomy with Typical Neurological and Immunological Findings: A Case Report with New Genetic Insights. Brain Sciences, 2021, 11, 21.	2.3	1
72	Spinal muscular atrophy: from rags to riches. Neuromuscular Disorders, 2021, 31, 998-1003.	0.6	14

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73	Pulmonary Function in Nonambulatory Patients with nmDMD from the STRIDE Registry and CINRG Duchenne Natural History Study: A Matched Cohort Analysis. , 2021, 52, .		0
74	Longitudinal Motor Functional Outcomes and Magnetic Resonance Imaging Patterns of Muscle Involvement in Upper Limbs in Duchenne Muscular Dystrophy. Medicina (Lithuania), 2021, 57, 1267.	2.0	4
75	Cortical Thickness and Clinical Findings in Prescholar Children With Autism Spectrum Disorder. Frontiers in Neuroscience, 2021, 15, 776860.	2.8	2
76	The social smile in infants during the COVID-19 pandemia. Heliyon, 2021, 7, e08648.	3.2	1
77	Visual Function Classification System for children with cerebral palsy: development and validation. Developmental Medicine and Child Neurology, 2020, 62, 104-110.	2.1	46
78	European muscle MRI study in limb girdle muscular dystrophy type R1/2A (LGMDR1/LGMD2A). Journal of Neurology, 2020, 267, 45-56.	3.6	43
79	Performance of Upper Limb module for Duchenne muscular dystrophy. Developmental Medicine and Child Neurology, 2020, 62, 633-639.	2.1	35
80	Long-term data with idebenone on respiratory function outcomes in patients with Duchenne muscular dystrophy. Neuromuscular Disorders, 2020, 30, 5-16.	0.6	33
81	MRI patterns of muscle involvement in type 2 and 3 spinal muscular atrophy patients. Journal of Neurology, 2020, 267, 898-912.	3.6	32
82	DMD & BMD â€“ CLINICAL. Neuromuscular Disorders, 2020, 30, S63-S64.	0.6	3
83	MUSCLE IMAGING â€“ MRI. Neuromuscular Disorders, 2020, 30, S95-S96.	0.6	0
84	SMA: REGISTRIES, BIOMARKERS & OUTCOME MEASURES. Neuromuscular Disorders, 2020, 30, S96-S97.	0.6	0
85	SMA â€“ THERAPY. Neuromuscular Disorders, 2020, 30, S120-S121.	0.6	1
86	SMA â€“ THERAPY. Neuromuscular Disorders, 2020, 30, S121.	0.6	4
87	SMA â€“ THERAPY. Neuromuscular Disorders, 2020, 30, S123.	0.6	1
88	SMA â€“ THERAPY. Neuromuscular Disorders, 2020, 30, S123-S124.	0.6	6
89	SMA â€“ THERAPY. Neuromuscular Disorders, 2020, 30, S126-S127.	0.6	0
90	DMD â€“ THERAPY. Neuromuscular Disorders, 2020, 30, S129-S130.	0.6	1

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91	A Recurrent Pathogenic Variant of INPP5K 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.	2.3	8
92	Spinal muscular atrophy â€" insights and challenges in the treatment era. Nature Reviews Neurology, 2020, 16, 706-715.	10.1	89
93	Childrenâ€™s Healthcare During Corona Virus Disease 19 Pandemic. Pediatric Infectious Disease Journal, 2020, 39, e137-e140.	2.0	14
94	Genotypeâ€“phenotype correlations in recessive titinopathies. Genetics in Medicine, 2020, 22, 2029-2040.	2.4	35
95	Gain and loss of abilities in type II SMA: A 12-month natural history study. Neuromuscular Disorders, 2020, 30, 765-771.	0.6	22
96	Early visual and neuro-development in preterm infants with and without retinopathy. Early Human Development, 2020, 148, 105134.	1.8	9
97	Age and baseline values predict 12 and 24-month functional changes in type 2 SMA. Neuromuscular Disorders, 2020, 30, 756-764.	0.6	25
98	Early Neurological Assessment and Long-Term Neuromotor Outcomes in Late Preterm Infants: A Critical Review. Medicina (Lithuania), 2020, 56, 475.	2.0	10
99	Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy. Journal of Comparative Effectiveness Research, 2020, 9, 973-984.	1.4	41
100	Clinical Variability in Spinal Muscular Atrophy Type <scp>III</scp>. Annals of Neurology, 2020, 88, 1109-1117.	5.3	34
101	NEW GENES AND DISEASES / NGS & RELATED TECHNIQUES. Neuromuscular Disorders, 2020, 30, S141-S142.	0.6	0
102	Nusinersen in type 0 spinal muscular atrophy: should we treat?. Annals of Clinical and Translational Neurology, 2020, 7, 2481-2483.	3.7	11
103	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.	0.6	15
104	Ultrasound assessment of diaphragmatic function in type 1 spinal muscular atrophy. Pediatric Pulmonology, 2020, 55, 1781-1788.	2.0	18
105	Spinal cord demyelination in children: A diagnostic challenge in neuropaediatrics for a good outcome. Brain and Development, 2020, 42, 457-461.	1.1	3
106	Randomized phase 2 trial and open-label extension of domagrozumab in Duchenne muscular dystrophy. Neuromuscular Disorders, 2020, 30, 492-502.	0.6	40
107	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.	0.6	11
108	Measuring Outcomes in Adults with Spinal Muscular Atrophy â€" Challenges and Future Directions â€" Meeting Report. Journal of Neuromuscular Diseases, 2020, 7, 523-534.	2.6	39

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109	The Genetic Landscape of Dystrophin Mutations in Italy: A Nationwide Study. Frontiers in Genetics, 2020, 11, 131.	2.3	49
110	Early Childhood Attention Battery: Italian adaptation and new expanded normative data. Early Human Development, 2020, 144, 105013.	1.8	2
111	Nusinersen in adults with spinal muscular atrophy: new challenges. Lancet Neurology, The, 2020, 19, 283-284.	10.2	15
112	Diagnostic journey in Spinal Muscular Atrophy: Is it still an odyssey?. PLoS ONE, 2020, 15, e0230677.	2.5	38
113	Increased dystrophin production with golodirsen in patients with Duchenne muscular dystrophy. Neurology, 2020, 94, e2270-e2282.	1.1	207
114	Pediatric Motor Inflammatory Neuropathy: The Role of Antiphospholipid Antibodies. Brain Sciences, 2020, 10, 156.	2.3	0
115	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.	2.6	17
116	Suitability of external controls for drug evaluation in Duchenne muscular dystrophy. Neurology, 2020, 95, e1381-e1391.	1.1	27
117	European ad-hoc consensus statement on gene replacement therapy for spinal muscular atrophy. European Journal of Paediatric Neurology, 2020, 28, 38-43.	1.6	74
118	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. Frontiers in Genetics, 2020, 11, 605.	2.3	9
119	Respiratory Needs in Patients with Type 1 Spinal Muscular Atrophy TreatedÂwith Nusinersen. Journal of Pediatrics, 2020, 219, 223-228.e4.	1.8	51
120	Cranial ultrasound evaluation in term neonates. Early Human Development, 2020, 143, 104983.	1.8	5
121	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.	1.4	82
122	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	3.7	36
123	Spinal muscular atrophy care in the COVIDâ€19 pandemic era. Muscle and Nerve, 2020, 62, 46-49.	2.2	31
124	Longitudinal natural history of type I spinal muscular atrophy: a critical review. Orphanet Journal of Rare Diseases, 2020, 15, 84.	2.7	45
125	Hypoglycaemia in patients with type 1 SMA: an underdiagnosed problem?. Archives of Disease in Childhood, 2020, 105, 707-707.	1.9	6
126	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.	1.5	24

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127	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.	0.7	9
128	Respiratory function and therapeutic expectations in DMD: families experience and perspective. Acta Myologica, 2020, 39, 121-129.	1.5	0
129	Brain morphometry of preschool age children affected by autism spectrum disorder: Correlation with clinical findings. Clinical Anatomy, 2019, 32, 143-150.	2.7	8
130	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.	1.4	29
131	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.	5.9	69
132	Neonatal hypotonia and neuromuscular conditions. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2019, 162, 435-448.	1.8	13
133	Nusinersen improves walking distance and reduces fatigue in laterâ€onset spinal muscular atrophy. Muscle and Nerve, 2019, 60, 409-414.	2.2	62
134	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.	0.5	0
135	P.151Motor performances in exon-2 duplication of the dystrophin gene. Neuromuscular Disorders, 2019, 29, S92-S93.	0.6	0
136	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.	0.6	3
137	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.	0.6	13
138	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.	0.6	4
139	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.	0.6	4
140	P.267Modifiers of respiratory and cardiac function in the Italian Duchenne muscular dystrophy network and CINRG Duchenne natural history study. Neuromuscular Disorders, 2019, 29, S145.	0.6	1
141	Development of an academic disease registry for spinal muscular atrophy. Neuromuscular Disorders, 2019, 29, 794-799.	0.6	29
142	Longitudinal natural history in young boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2019, 29, 857-862.	0.6	23
143	Early Neurological Assessment in Infants with Hypoxic Ischemic Encephalopathy Treated with Therapeutic Hypothermia. Journal of Clinical Medicine, 2019, 8, 1247.	2.4	7
144	Long-term progression in type II spinal muscular atrophy. Neurology, 2019, 93, e1241-e1247.	1.1	34

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145	Predominant distal muscle involvement in spinal muscular atrophy. Neuromuscular Disorders, 2019, 29, 910-911.	0.6	2
146	MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscular Disorders, 2019, 29, 827-841.	0.6	46
147	Revised upper limb module for spinal muscular atrophy: 12â€‰month changes. Muscle and Nerve, 2019, 59, 426-430.	2.2	61
148	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	4
149	Nusinersen in type 1 spinal muscular atrophy: Twelveâ€month realâ€world data. Annals of Neurology, 2019, 86, 443-451.	5.3	83
150	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.	2.5	47
151	Sleep disorders in low-risk preschool very preterm children. Sleep Medicine, 2019, 63, 137-141.	1.6	7
152	Neurofilament as a potential biomarker for spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 932-944.	3.7	137
153	An unusual ryanodine receptor 1 (RYR1) phenotype. Neurology, 2019, 92, e1600-e1609.	1.1	16
154	Value of structured reporting in neuromuscular disorders. Radiologia Medica, 2019, 124, 628-635.	7.7	7
155	Resolution of skin necrosis after nusinersen treatment in an infant with spinal muscular atrophy. Muscle and Nerve, 2019, 59, E42-E44.	2.2	4
156	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.	0.6	26
157	Muscular dystrophies. Lancet, The, 2019, 394, 2025-2038.	13.7	276
158	Next-generation sequencing approach to hyperCKemia. Neurology: Genetics, 2019, 5, e352.	1.9	31
159	Onasemnogene abeparvovec gene-replacement therapy (GRT) for spinal muscular atrophy type 1 (SMA1): Global pivotal phase 3 study program (STR1VE-US, STR1VE-EU, STR1VE-AP). Journal of the Neurological Sciences, 2019, 405, 277-278.	0.6	2
160	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.	3.2	30
161	Intrathecal nusinersen treatment for SMA in a dedicated neuromuscular clinic: an example of multidisciplinary and integrated care. Neurological Sciences, 2019, 40, 327-332.	1.9	18
162	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.	0.6	30

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163	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.	0.1	0
164	Study Design of STR1VE-EU, a Phase 3 Trial of AVXS-101 Gene-Replacement Therapy (GRT) in Patients With Spinal Muscular Atrophy Type 1 (SMA1) in Europe. , 2019, 50, .		0
165	Effect of Ataluren on Age at Loss of Ambulation in Nonsense Mutation Duchenne Muscular Dystrophy: Observational Data from the STRIDE Registry. , 2019, 50, .		0
166	Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. Neuromuscular Disorders, 2018, 28, 103-115.	0.6	584
167	Joint Laxity in Preschool Children Born Preterm. Journal of Pediatrics, 2018, 197, 104-108.	1.8	5
168	Evidence-based care in Duchenne muscular dystrophy. Lancet Neurology, The, 2018, 17, 389-391.	10.2	13
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170	Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2018, 378, 625-635.	27.0	977
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