Richard S Finkel

List of Publications by Year
in descending order

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140	13,141	36271	24232
papers	citations	51 h-index	110 g-index

145	145	145	6949
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Preventing amyotrophic lateral sclerosis: insights from pre-symptomatic neurodegenerative diseases. Brain, 2022, 145, 27-44.	3.7	38
2	Revised upper limb module in type II and III spinal muscular atrophy: 24-month changes. Neuromuscular Disorders, 2022, 32, 36-42.	0.3	13
3	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	1.1	7
4	Efficacy and Safety of Vamorolone in Duchenne Muscular Dystrophy. JAMA Network Open, 2022, 5, e2144178.	2.8	31
5	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.	2.7	6
6	Clinical practice guideline for the management of paediatric Charcot-Marie-Tooth disease. Journal of Neurology, Neurosurgery and Psychiatry, 2022, 93, 530-538.	0.9	10
7	Pre-symptomatic spinal muscular atrophy: a proposed nosology. Brain, 2022, 145, 2247-2249.	3.7	11
8	Intron mutations and early transcription termination in Duchenne and Becker muscular dystrophy. Human Mutation, 2022, 43, 511-528.	1.1	16
9	Friend or Foe(tal): challenges in development of a large animal model for pre-clinical fetal gene therapy. Gene Therapy, 2022, , .	2.3	1
10	Repeated intravenous cardiosphere-derived cell therapy in late-stage Duchenne muscular dystrophy (HOPE-2): a multicentre, randomised, double-blind, placebo-controlled, phase 2 trial. Lancet, The, 2022, 399, 1049-1058.	6.3	36
11	Distribution of Weight, Stature and Growth Status in Children and Adolescents with Spinal Muscular Atrophy: An Observational Retrospective Study in the United States. Muscle and Nerve, 2022, , .	1.0	2
12	Effect of Different Corticosteroid Dosing Regimens on Clinical Outcomes in Boys With Duchenne Muscular Dystrophy. JAMA - Journal of the American Medical Association, 2022, 327, 1456.	3.8	43
13	Step Activity Monitoring in Boys with Duchenne Muscular Dystrophy and its Correlation with Magnetic Resonance Measures and Functional Performance. Journal of Neuromuscular Diseases, 2022, , 1-14.	1.1	2
14	Scientific rationale for a higher dose of nusinersen. Annals of Clinical and Translational Neurology, 2022, 9, 819-829.	1.7	9
15	Oral and Swallowing Abilities Tool (OrSAT) in nusinersen treated patients. Archives of Disease in Childhood, 2022, 107, 912-916.	1.0	3
16	Onasemnogene abeparvovec for presymptomatic infants with two copies of SMN2 at risk for spinal muscular atrophy type 1: the Phase III SPR1NT trial. Nature Medicine, 2022, 28, 1381-1389.	15.2	99
17	Onasemnogene abeparvovec for presymptomatic infants with three copies of SMN2 at risk for spinal muscular atrophy: the Phase III SPR1NT trial. Nature Medicine, 2022, 28, 1390-1397.	15.2	93
18	Respiratory Trajectories in Type 2 and 3 Spinal Muscular Atrophy in the iSMAC Cohort Study. Neurology, 2021, 96, e587-e599.	1.5	36

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19	Development and Validation of the Pediatric Charcotâ€“Marieâ€“Tooth Disease Quality of Life Outcome Measure. Annals of Neurology, 2021, 89, 369-379.	2.8	13
20	Reldesemtiv in Patients with Spinal Muscular Atrophy: a Phase 2 Hypothesis-Generating Study. Neurotherapeutics, 2021, 18, 1127-1136.	2.1	28
21	Disease-modifying effects of edasalonexent, an NF-ÎºB inhibitor, in young boys with Duchenne muscular dystrophy: Results of the MoveDMD phase 2 and open label extension trial. Neuromuscular Disorders, 2021, 31, 385-396.	0.3	20
22	Onasemnogene abeparvovec gene therapy for symptomatic infantile-onset spinal muscular atrophy in patients with two copies of SMN2 (STR1VE): an open-label, single-arm, multicentre, phase 3 trial. Lancet Neurology, The, 2021, 20, 284-293.	4.9	227
23	Population pharmacokinetics-based recommendations for a single delayed or missed dose of nusinersen. Neuromuscular Disorders, 2021, 31, 310-318.	0.3	10
24	Maybe too much of a good thing in gene therapy. Nature Neuroscience, 2021, 24, 901-902.	7.1	4
25	Nusinersen in pediatric and adult patients with type III spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2021, 8, 1622-1634.	1.7	27
26	Adeno-associated virus serotype 9 antibodies in patients screened for treatment with onasemnogene abeparvovec. Molecular Therapy - Methods and Clinical Development, 2021, 21, 76-82.	1.8	24
27	Treatment of infantile-onset spinal muscular atrophy with nusinersen: final report of a phase 2, open-label, multicentre, dose-escalation study. The Lancet Child and Adolescent Health, 2021, 5, 491-500.	2.7	47
28	Oral and Swallowing Abilities Tool (OrSAT) for Type 1 SMA Patients: Development of a New Module. Journal of Neuromuscular Diseases, 2021, 8, 589-601.	1.1	16
29	Assessing the ability of boys with Duchenne muscular dystrophy age 4â€“7 years to swallow softgel capsules: Clinical trial experience with edasalonexent. Journal of Clinical Pharmacy and Therapeutics, 2021, , .	0.7	0
30	Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. Neuromuscular Disorders, 2021, 31, 596-602.	0.3	29
31	Clinical Trial and Postmarketing Safety of Onasemnogene Abeparvovec Therapy. Drug Safety, 2021, 44, 1109-1119.	1.4	62
32	Different trajectories in upper limb and gross motor function in spinal muscular atrophy. Muscle and Nerve, 2021, 64, 552-559.	1.0	18
33	Association Between Body Mass Index and Disability in Children With Charcot-Marie-Tooth Disease. Neurology, 2021, 97, e1727-e1736.	1.5	2
34	Psychometric properties of the PEDI-CAT for children and youth with spinal muscular atrophy. Journal of Pediatric Rehabilitation Medicine, 2021, 14, 451-461.	0.3	2
35	A Randomized, Double-Blind, Placebo-Controlled, Global Phase 3 Study of Edasalonexent in Pediatric Patients with Duchenne Muscular Dystrophy: Results of the PolarisDMD Trial. Journal of Neuromuscular Diseases, 2021, 8, 769-784.	1.1	13
36	Nusinersen Treatment in Adults With Spinal Muscular Atrophy. Neurology: Clinical Practice, 2021, 11, e317-e327.	0.8	35

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37	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
38	Spinal muscular atrophy â€" insights and challenges in the treatment era. Nature Reviews Neurology, 2020, 16, 706-715.	4.9	89
39	Refining clinical trial inclusion criteria to optimize the standardized response mean of the CMTPedS. Annals of Clinical and Translational Neurology, 2020, 7, 1713-1715.	1.7	5
40	Gain and loss of abilities in type II SMA: A 12-month natural history study. Neuromuscular Disorders, 2020, 30, 765-771.	0.3	22
41	Editorial: Spinal Muscular Atrophy: Evolutions and Revolutions of Modern Therapy. Frontiers in Neurology, 2020, 11, 783.	1.1	5
42	Age and baseline values predict 12 and 24-month functional changes in type 2 SMA. Neuromuscular Disorders, 2020, 30, 756-764.	0.3	25
43	Efficacy and safety of vamorolone in Duchenne muscular dystrophy:ÂAn 18-month interim analysis of a non-randomized open-label extension study. PLoS Medicine, 2020, 17, e1003222.	3.9	41
44	Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy. Journal of Comparative Effectiveness Research, 2020, 9, 973-984.	0.6	41
45	Clinical Variability in Spinal Muscular Atrophy Type <scp>III</scp>. Annals of Neurology, 2020, 88, 1109-1117.	2.8	34
46	<scp>GGPS1</scp> Mutations Cause Muscular Dystrophy/Hearing Loss/Ovarian Insufficiency Syndrome. Annals of Neurology, 2020, 88, 332-347.	2.8	22
47	Scoliosis Surgery Significantly Impacts Motor Abilities in Higher-functioning Individuals with Spinal Muscular Atrophy1. Journal of Neuromuscular Diseases, 2020, 7, 183-192.	1.1	7
48	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.	1.1	17
49	A longitudinal study of CMT1A using Rasch analysis based CMT neuropathy and examination scores. Neurology, 2020, 94, e884-e896.	1.5	29
50	GATAD2B-associatedneurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-relateddisorder. Genetics in Medicine, 2020, 22, 878-888.	1.1	22
51	Spinal muscular atrophy care in the COVIDâ€19 pandemic era. Muscle and Nerve, 2020, 62, 46-49.	1.0	31
52	Revised Recommendations for the Treatment of Infants Diagnosed with Spinal Muscular Atrophy Via Newborn Screening Who Have 4 Copies of SMN2. Journal of Neuromuscular Diseases, 2020, 7, 97-100.	1.1	89
53	Longitudinal natural history of type I spinal muscular atrophy: a critical review. Orphanet Journal of Rare Diseases, 2020, 15, 84.	1.2	45
54	Natural history of Charcot-Marie-Tooth disease type 2A: a large international multicentre study. Brain, 2020, 143, 3589-3602.	3.7	39

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55	Modeling disease trajectory in Duchenne muscular dystrophy. Neurology, 2020, 94, e1622-e1633.	1.5	49
56	Title is missing!. , 2020, 17, e1003222.		0
57	Title is missing!. , 2020, 17, e1003222.		0
58	Title is missing!. , 2020, 17, e1003222.		0
59	Title is missing!. , 2020, 17, e1003222.		0
60	Title is missing!. , 2020, 17, e1003222.		0
61	Title is missing!. , 2020, 17, e1003222.		0
62	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
63	Nusinersen improves walking distance and reduces fatigue in laterâ€onset spinal muscular atrophy. Muscle and Nerve, 2019, 60, 409-414.	1.0	62
64	Development of an academic disease registry for spinal muscular atrophy. Neuromuscular Disorders, 2019, 29, 794-799.	0.3	29
65	Evolution of Next Generation Therapeutics: Past, Present, and Future of Precision Medicines. Clinical and Translational Science, 2019, 12, 560-563.	1.5	1
66	Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. Neuromuscular Disorders, 2019, 29, 842-856.	0.3	401
67	Twiceâ€weekly glucocorticosteroids in infants and young boys with Duchenne muscular dystrophy. Muscle and Nerve, 2019, 59, 650-657.	1.0	32
68	Revised upper limb module for spinal muscular atrophy: 12â€‰month changes. Muscle and Nerve, 2019, 59, 426-430.	1.0	61
69	Maximizing the Benefit of Life-Saving Treatments for Pompe Disease, Spinal Muscular Atrophy, and Duchenne Muscular Dystrophy Through Newborn Screening. JAMA Neurology, 2019, 76, 978.	4.5	14
70	Neurofilament as a potential biomarker for spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2019, 6, 932-944.	1.7	137
71	Balance impairment in pediatric charcotâ€“marieâ€“tooth disease. Muscle and Nerve, 2019, 60, 242-249.	1.0	22
72	Selective serotonin reuptake inhibitors ameliorate MEGF10 myopathy. Human Molecular Genetics, 2019, 28, 2365-2377.	1.4	7

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73	Vamorolone trial in Duchenne muscular dystrophy shows dose-related improvement of muscle function. Neurology, 2019, 93, e1312-e1323.	1.5	64
74	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.	0.3	26
75	Phase 1 Study of Edasalonexent (CAT-1004), an Oral NF-Î¹B Inhibitor, in Pediatric Patients with Duchenne Muscular Dystrophy. Journal of Neuromuscular Diseases, 2019, 6, 43-54.	1.1	38
76	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
77	Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy through Newborn Screening. Journal of Neuromuscular Diseases, 2018, 5, 145-158.	1.1	148
78	Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2018, 378, 625-635.	13.9	977
79	Motor milestone assessment of infants with spinal muscular atrophy using the hammersmith infant neurological Examâ€"Part 2: Experience from a nusinersen clinical study. Muscle and Nerve, 2018, 57, 142-146.	1.0	47
80	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
81	Phase IIa trial in Duchenne muscular dystrophy shows vamorolone is a first-in-class dissociative steroidal anti-inflammatory drug. Pharmacological Research, 2018, 136, 140-150.	3.1	69
82	Ambulatory function in spinal muscular atrophy: Age-related patterns of progression. PLoS ONE, 2018, 13, e0199657.	1.1	65
83	Genetic therapies for inherited neuromuscular disorders. The Lancet Child and Adolescent Health, 2018, 2, 600-609.	2.7	40
84	Position Statement: Sharing of Clinical Research Data in Spinal Muscular Atrophy to Accelerate Research and Improve Outcomes for Patients. Journal of Neuromuscular Diseases, 2018, 5, 131-133.	1.1	10
85	Quantitative Evaluation of Lower Extremity Joint Contractures in Spinal Muscular Atrophy: Implications for Motor Function. Pediatric Physical Therapy, 2018, 30, 209-215.	0.3	18
86	A checklist for clinical trials in rare disease: obstacles and anticipatory actionsâ€"lessons learned from the FOR-DMD trial. Trials, 2018, 19, 291.	0.7	26
87	Longitudinal timed function tests in Duchenne muscular dystrophy: ImagingDMD cohort natural history. Muscle and Nerve, 2018, 58, 631-638.	1.0	41
88	Evaluator Training and Reliability for SMA Global Nusinersen Trials1. Journal of Neuromuscular Diseases, 2018, 5, 159-166.	1.1	36
89	Skeletal muscle magnetic resonance biomarkers correlate with function and sentinel events in Duchenne muscular dystrophy. PLoS ONE, 2018, 13, e0194283.	1.1	52
90	218th ENMC International Workshop:. Neuromuscular Disorders, 2017, 27, 596-605.	0.3	49

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91	Content validity and clinical meaningfulness of the HFMSE in spinal muscular atrophy. BMC Neurology, 2017, 17, 39.	0.8	102
92	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. Contemporary Clinical Trials, 2017, 58, 34-39.	0.8	56
93	Spinal muscular atrophy: A changing phenotype beyond the clinical trials. Neuromuscular Disorders, 2017, 27, 883-889.	0.3	89
94	Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2017, 377, 1723-1732.	13.9	1,533
95	A phase 3 randomized placebo-controlled trial of tadalafil for Duchenne muscular dystrophy. Neurology, 2017, 89, 1811-1820.	1.5	58
96	X-linked myotubular myopathy. Neurology, 2017, 89, 1316-1317.	1.5	1
97	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
98	Delayed onset of ambulation in boys with Duchenne muscular dystrophy: Potential use as an endpoint in clinical trials. Neuromuscular Disorders, 2017, 27, 905-910.	0.3	9
99	Natural history of Charcotâ€Marieâ€Tooth disease during childhood. Annals of Neurology, 2017, 82, 353-359.	2.8	50
100	Natural history of infantileâ€onset spinal muscular atrophy. Annals of Neurology, 2017, 82, 883-891.	2.8	276
101	Spinal Muscular Atrophy Type I. Journal of Child Neurology, 2017, 32, 155-160.	0.7	18
102	Treatment effect of idebenone on inspiratory function in patients with Duchenne muscular dystrophy. Pediatric Pulmonology, 2017, 52, 508-515.	1.0	32
103	Revised upper limb module for spinal muscular atrophy: Development of a new module. Muscle and Nerve, 2017, 55, 869-874.	1.0	166
104	Revised Hammersmith Scale for spinal muscular atrophy: A SMA specific clinical outcome assessment tool. PLoS ONE, 2017, 12, e0172346.	1.1	67
105	Rasch analysis of the Pediatric Evaluation of Disability Inventoryâ€“computer adaptive test (PEDIâ€CAT) item bank for children and young adults with spinal muscular atrophy. Muscle and Nerve, 2016, 54, 1097-1107.	1.0	17
106	Physical therapy services received by individuals with spinal muscular atrophy (SMA). Journal of Pediatric Rehabilitation Medicine, 2016, 9, 35-44.	0.3	13
107	Treatment of infantile-onset spinal muscular atrophy with nusinersen: a phase 2, open-label, dose-escalation study. Lancet, The, 2016, 388, 3017-3026.	6.3	801
108	Idebenone reduces respiratory complications in patients with Duchenne muscular dystrophy. Neuromuscular Disorders, 2016, 26, 473-480.	0.3	55

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109	Phenotypic Variability of Childhood Charcot-Marie-Tooth Disease. JAMA Neurology, 2016, 73, 645.	4.5	71
110	Developmental milestones in type I spinal muscular atrophy. Neuromuscular Disorders, 2016, 26, 754-759.	0.3	96
111	Baseline results of the Neuro<scp>NEXT</scp> spinal muscular atrophy infant biomarker study. Annals of Clinical and Translational Neurology, 2016, 3, 132-145.	1.7	106
112	Multicenter prospective longitudinal study of magnetic resonance biomarkers in a large duchenne muscular dystrophy cohort. Annals of Neurology, 2016, 79, 535-547.	2.8	131
113	Patterns of disease progression in type 2 and 3 SMA: Implications for clinical trials. Neuromuscular Disorders, 2016, 26, 126-131.	0.3	142
114	Old measures and new scores in spinal muscular atrophy patients. Muscle and Nerve, 2015, 52, 435-437.	1.0	6
115	Spinal muscular atrophy functional composite score: A functional measure in spinal muscular atrophy. Muscle and Nerve, 2015, 52, 942-947.	1.0	26
116	Spectrum of Neuropathophysiology in Spinal Muscular Atrophy Type I. Journal of Neuropathology and Experimental Neurology, 2015, 74, 15-24.	0.9	80
117	Efficacy of idebenone on respiratory function in patients with Duchenne muscular dystrophy not using glucocorticoids (DELOS): a double-blind randomised placebo-controlled phase 3 trial. Lancet, The, 2015, 385, 1748-1757.	6.3	160
118	Association of a NovelACTA1Mutation With a Dominant Progressive Scapuloperoneal Myopathy in an Extended Family. JAMA Neurology, 2015, 72, 689.	4.5	35
119	Magnetic Resonance Imaging and Spectroscopy Assessment of Lower Extremity Skeletal Muscles in Boys with Duchenne Muscular Dystrophy: A Multicenter Cross Sectional Study. PLoS ONE, 2014, 9, e106435.	1.1	94
120	Examination of effects of corticosteroids on skeletal muscles of boys with DMD using MRI and MRS. Neurology, 2014, 83, 974-980.	1.5	131
121	Respiratory muscle function in infants with spinal muscular atrophy type I. Pediatric Pulmonology, 2014, 49, 1234-1242.	1.0	25
122	The motor neuron response to SMN1 deficiency in spinal muscular atrophy. Muscle and Nerve, 2014, 49, 636-644.	1.0	34
123	Observational study of spinal muscular atrophy type I and implications for clinical trials. Neurology, 2014, 83, 810-817.	1.5	367
124	Electrophysiological and motor function scale association in a pre-symptomatic infant with spinal muscular atrophy type I. Neuromuscular Disorders, 2013, 23, 112-115.	0.3	35
125	SMA-MAP: A Plasma Protein Panel for Spinal Muscular Atrophy. PLoS ONE, 2013, 8, e60113.	1.1	40
126	Phase 2a Study of Ataluren-Mediated Dystrophin Production in Patients with Nonsense Mutation Duchenne Muscular Dystrophy. PLoS ONE, 2013, 8, e81302.	1.1	201

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127	Prospective cohort study of spinal muscular atrophy types 2 and 3. Neurology, 2012, 79, 1889-1897.	1.5	207
128	Candidate Proteins, Metabolites and Transcripts in the Biomarkers for Spinal Muscular Atrophy (BforSMA) Clinical Study. PLoS ONE, 2012, 7, e35462.	1.1	71
129	Validation of the Charcotâ€“Marieâ€“Tooth disease pediatric scale as an outcome measure of disability. Annals of Neurology, 2012, 71, 642-652.	2.8	137
130	Evaluation of SMN Protein, Transcript, and Copy Number in the Biomarkers for Spinal Muscular Atrophy (BforSMA) Clinical Study. PLoS ONE, 2012, 7, e33572.	1.1	130
131	Assessing upper limb function in nonambulant SMA patients: Development of a new module. Neuromuscular Disorders, 2011, 21, 406-412.	0.3	71
132	Validation of the Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	0.3	106
133	Thigh Muscle Volume Measured by Magnetic Resonance Imaging Is Stable Over a 6-Month Interval in Spinal Muscular Atrophy. Journal of Child Neurology, 2011, 26, 1252-1259.	0.7	25
134	Validation of the Expanded Hammersmith Functional Motor Scale in Spinal Muscular Atrophy Type II and III. Journal of Child Neurology, 2011, 26, 1499-1507.	0.7	143
135	Observational Study of Spinal Muscular Atrophy Type 2 and 3. Archives of Neurology, 2011, 68, 779.	4.9	142
136	Read-Through Strategies for Suppression of Nonsense Mutations in Duchenne/ Becker Muscular Dystrophy: Aminoglycosides and Ataluren (PTC124). Journal of Child Neurology, 2010, 25, 1158-1164.	0.7	135
137	Adiposity is increased among high-functioning, non-ambulatory patients with spinal muscular atrophy. Neuromuscular Disorders, 2010, 20, 448-452.	0.3	47
138	The Test of Infant Motor Performance: Reliability in Spinal Muscular Atrophy Type I. Pediatric Physical Therapy, 2008, 20, 242-246.	0.3	26
139	An expanded version of the Hammersmith Functional Motor Scale for SMA II and III patients. Neuromuscular Disorders, 2007, 17, 693-697.	0.3	245
140	Consensus Statement for Standard of Care in Spinal Muscular Atrophy. Journal of Child Neurology, 2007, 22, 1027-1049.	0.7	754