Janbernd Kirschner

List of Publications by Year
in descending order

152 papers	11,613 citations	50276 46 h-index	30922 102 g-index
162 all docs	162 docs citations	162 times ranked	12259 citing authors

#	Article	IF	Citations
1	Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2017, 377, 1723-1732.	27.0	1,533
2	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. Nature, 2013, 495, 467-473.	27.8	1,249
3	Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. New England Journal of Medicine, 2018, 378, 625-635.	27.0	977
4	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
5	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.	0.6	421
6	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.	0.6	401
7	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.	13.7	365
8	Use of Whole-Exome Sequencing to Determine the Genetic Basis of Multiple Mitochondrial Respiratory Chain Complex Deficiencies. JAMA - Journal of the American Medical Association, 2014, 312, 68.	7.4	304
9	ORAI1 deficiency and lack of store-operated Ca2+ entry cause immunodeficiency, myopathy, and ectodermal dysplasia. Journal of Allergy and Clinical Immunology, 2009, 124, 1311-1318.e7.	2.9	289
10	Nusinersen in adults with 5q spinal muscular atrophy: a non-interventional, multicentre, observational cohort study. Lancet Neurology, The, 2020, 19, 317-325.	10.2	196
11	Lack of the Mitochondrial Protein Acylglycerol Kinase Causes Sengers Syndrome. American Journal of Human Genetics, 2012, 90, 314-320.	6.2	192
12	Intravenously Administered Immunoglobulin in the Treatment of Childhood Guillain-Barre Syndrome: A Randomized Trial. Pediatrics, 2005, 116, 8-14.	2.1	160
13	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. American Journal of Human Genetics, 2011, 88, 162-172.	6.2	153
14	Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. Human Mutation, 2012, 33, 981-988.	2.5	145
15	Clinical and morphological phenotype of the filamin myopathy: a study of 31 German patients. Brain, 2007, 130, 3250-3264.	7.6	132
16	Safety and Treatment Effects of Nusinersen in Longstanding Adult 5q-SMA Type 3 â€“ A Prospective Observational Study. Journal of Neuromuscular Diseases, 2019, 6, 453-465.	2.6	132
17	Life expectancy at birth in Duchenne muscular dystrophy: a systematic review and meta-analysis. European Journal of Epidemiology, 2020, 35, 643-653.	5.7	132
18	Peripheral Nerve Demyelination Caused by a Mutant Rho GTPase Guanine Nucleotide Exchange Factor, Frabin/FGD4. American Journal of Human Genetics, 2007, 81, 158-164.	6.2	128

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19	Proteomic identification of FHL1 as the protein mutated in human reducing body myopathy. Journal of Clinical Investigation, 2008, 118, 904-12.	8.2	126
20	Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. Journal of Neuromuscular Diseases, 2017, 4, 293-306.	2.6	125
21	Advances in Treatment of Spinal Muscular Atrophy â€“ New Phenotypes, New Challenges, New Implications for Care. Journal of Neuromuscular Diseases, 2020, 7, 1-13.	2.6	124
22	A randomized placebo-controlled phase 3 trial of an antisense oligonucleotide, drisapersen, in Duchenne muscular dystrophy. Neuromuscular Disorders, 2018, 28, 4-15.	0.6	102
23	Adult-onset cerebellar ataxia due to mutations inCABC1/ADCK3. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 174-178.	1.9	99
24	A multi-source approach to determine SMA incidence and research ready population. Journal of Neurology, 2017, 264, 1465-1473.	3.6	98
25	Evaluation of Children with SMA Type 1 Under Treatment with Nusinersen within the Expanded Access Program in Germany. Journal of Neuromuscular Diseases, 2018, 5, 135-143.	2.6	97
26	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.	10.2	95
27	Mitochondrial Tubulopathy in Tenofovir Disoproxil Fumarate-Treated Rats. Journal of Acquired Immune Deficiency Syndromes (1999), 2009, 51, 258-263.	2.1	90
28	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
29	Ullrich congenital muscular dystrophy: Connective tissue abnormalities in the skin support overlap with Ehlers-Danlos syndromes. , 2005, 132A, 296-301.		87
30	Pontocerebellar hypoplasia type 1. Neurology, 2013, 80, 438-446.	1.1	84
31	Elevated satellite cell number in Duchenne muscular dystrophy. Cell and Tissue Research, 2010, 340, 541-548.	2.9	83
32	A Combined Laser Microdissection and Mass Spectrometry Approach Reveals New Disease Relevant Proteins Accumulating in Aggregates of Filaminopathy Patients. Molecular and Cellular Proteomics, 2013, 12, 215-227.	3.8	74
33	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
34	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850.	3.6	72
35	SMArtCAREÂ-ÂA platform to collect real-life outcome data of patients with spinal muscular atrophy. Orphanet Journal of Rare Diseases, 2019, 14, 18.	2.7	67
36	SIL1 mutations and clinical spectrum in patients with Marinesco-SjÃ¶gren syndrome. Brain, 2013, 136, 3634-3644.	7.6	65

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37	p.S143F mutation in lamin A/C: A new phenotype combining myopathy and progeria. Annals of Neurology, 2005, 57, 148-151.	5.3	63
38	Evaluation of Botulinum Toxin A Therapy in Children With Adductor Spasm by Gross Motor Function Measure. Journal of Child Neurology, 2000, 15, 214-217.	1.4	60
39	The Congenital and Limb-Girdle Muscular Dystrophies. Archives of Neurology, 2004, 61, 189.	4.5	60
40	A homozygous splice-site mutation in CARS2 is associated with progressive myoclonic epilepsy. Neurology, 2014, 83, 2183-2187.	1.1	59
41	Duchenne muscular dystrophy and caregiver burden: a systematic review. Developmental Medicine and Child Neurology, 2018, 60, 987-996.	2.1	59
42	Mutations in Subunits of the Activating Signal Cointegrator 1 Complex Are Associated with Prenatal Spinal Muscular Atrophy and Congenital Bone Fractures. American Journal of Human Genetics, 2016, 98, 473-489.	6.2	56
43	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. Contemporary Clinical Trials, 2017, 58, 34-39.	1.8	56
44	European Cross-Sectional Survey ofÂCurrent Care Practices for Duchenne Muscular Dystrophy Reveals Regional andÂAge-Dependent Differences. Journal of Neuromuscular Diseases, 2016, 3, 517-527.	2.6	55
45	Treatment of Duchenne muscular dystrophy with ciclosporin A: a randomised, double-blind, placebo-controlled multicentre trial. Lancet Neurology, The, 2010, 9, 1053-1059.	10.2	54
46	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. Human Molecular Genetics, 2018, 27, 1186-1195.	2.9	52
47	Myopathy in Marinescoâ€“SjÃ¶gren syndrome links endoplasmic reticulum chaperone dysfunction to nuclear envelope pathology. Acta Neuropathologica, 2014, 127, 761-777.	7.7	51
48	Whole-body vibration training in children with Duchenne muscular dystrophy and spinal muscular atrophy. European Journal of Paediatric Neurology, 2014, 18, 140-149.	1.6	48
49	Quality of life of patients with spinal muscular atrophy: A systematic review. European Journal of Paediatric Neurology, 2019, 23, 347-356.	1.6	48
50	Sarcoglycanopathies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2011, 101, 41-46.	1.8	46
51	Uridine supplementation antagonizes zalcitabine-induced microvesicular steatohepatitis in mice. Hepatology, 2007, 45, 72-79.	7.3	45
52	Discrepancy in redetermination of SMN2 copy numbers in children with SMA. Neurology, 2019, 93, 267-269.	1.1	43
53	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	3.6	43
54	Drug treatment of Duchenne muscular dystrophy: available evidence and perspectives. Acta Myologica, 2012, 31, 4-8.	1.5	43

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55	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.	7.4	43
56	Targeted sequencing with expanded gene profile enables high diagnostic yield in non-5q-spinal muscular atrophies. Human Mutation, 2018, 39, 1284-1298.	2.5	42
57	Adult care for Duchenne muscular dystrophy in the UK. Journal of Neurology, 2015, 262, 629-641.	3.6	41
58	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
59	Congenital Muscular Dystrophies and Myopathies: An Overview and Update. Neuropediatrics, 2017, 48, 247-261.	0.6	40
60	Safety Monitoring of Gene Therapy for Spinal Muscular Atrophy with Onasemnogene Abeparvovec â€“A Single Centre Experience. Journal of Neuromuscular Diseases, 2021, 8, 209-216.	2.6	39
61	Decompressive Craniectomy after Severe Traumatic Brain Injury in Children: Complications and Outcome. Neuropediatrics, 2015, 46, 005-012.	0.6	38
62	Infections and vaccinations preceding childhood Guillain-BarrÃ© syndrome: a prospective study. European Journal of Pediatrics, 2006, 165, 605-612.	2.7	37
63	<scp>TPM</scp>3 deletions cause a hypercontractile congenital muscle stiffness phenotype. Annals of Neurology, 2015, 78, 982-994.	5.3	36
64	Characterization of pulmonary function in 10â€“18 year old patients with Duchenne muscular dystrophy. Neuromuscular Disorders, 2017, 27, 307-314.	0.6	36
65	Variable impairment of platelet functions in patients with severe, genetically linked immune deficiencies. Haematologica, 2018, 103, 540-549.	3.5	36
66	Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. Lancet Neurology, The, 2020, 19, 522-532.	10.2	36
67	Expanding Phenotype of De Novo Mutations in GNAO1: Four New Cases and Review of Literature. Neuropediatrics, 2017, 48, 371-377.	0.6	33
68	Absence of transcallosal inhibition in adolescents with diplegic cerebral palsy. , 1999, 22, 255-257.		32
69	Single-center experience with intrathecal administration of Nusinersen in children with spinal muscular atrophy type 1. European Journal of Paediatric Neurology, 2018, 22, 122-127.	1.6	31
70	Predominant fiber atrophy and fiber type disproportion in early ullrich disease. Muscle and Nerve, 2008, 38, 1184-1191.	2.2	30
71	Uridine supplementation antagonizes zidovudineâ€induced mitochondrial myopathy and hyperlactatemia in mice. Arthritis and Rheumatism, 2008, 58, 318-326.	6.7	29
72	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

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73	Botulinum toxin treatment in cerebral palsy: evidence for a new treatment option. Journal of Neurology, 2001, 248, I28-I30.	3.6	28
74	Respiratory chain deficiency precedes the disrupted calcium homeostasis in chronic doxorubicin cardiomyopathy. Cardiovascular Pathology, 2010, 19, e167-e174.	1.6	28
75	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	12.8	28
76	Muscle-fiber transdifferentiation in an experimental model of respiratory chain myopathy. Arthritis Research and Therapy, 2012, 14, R233.	3.5	27
77	Congenital muscular dystrophies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1377-1385.	1.8	27
78	The TREAT-NMD care and trial site registry: an online registry to facilitate clinical research for neuromuscular diseases. Orphanet Journal of Rare Diseases, 2013, 8, 171.	2.7	26
79	High creatine kinase levels and white matter changes: Clinical and genetic spectrum of congenital muscular dystrophies with laminin alpha-2 deficiency. Molecular and Cellular Probes, 2014, 28, 118-122.	2.1	26
80	A checklist for clinical trials in rare disease: obstacles and anticipatory actionsâ€"lessons learned from the FOR-DMD trial. Trials, 2018, 19, 291.	1.6	26
81	Predictors of Health-Related Quality of Life in boys with Duchenne muscular dystrophy from six European countries. Journal of Neurology, 2017, 264, 709-723.	3.6	25
82	Respiratory chain deficiency in nonmitochondrial disease. Neurology: Genetics, 2015, 1, e6.	1.9	23
83	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
84	Adductor spasticity in children with cerebral palsy and treatment with botulinum toxin type A: the parents' view of functional outcome. European Journal of Neurology, 1999, 6, s47-s50.	3.3	22
85	Novel homozygous RARS2 mutation in two siblings without pontocerebellar hypoplasia â€“ further expansion of the phenotypic spectrum. Orphanet Journal of Rare Diseases, 2016, 11, 140.	2.7	22
86	De-duplicating patient records from three independent data sources reveals the incidence of rare neuromuscular disorders in Germany. Orphanet Journal of Rare Diseases, 2019, 14, 152.	2.7	22
87	Effect and safety of treatment with ACE-inhibitor Enalapril and Î²-blocker metoprolol on the onset of left ventricular dysfunction in Duchenne muscular dystrophy - a randomized, double-blind, placebo-controlled trial. Orphanet Journal of Rare Diseases, 2019, 14, 105.	2.7	21
88	Oral uridine supplementation antagonizes the peripheral neuropathy and encephalopathy induced by antiretroviral nucleoside analogues. Aids, 2010, 24, 345-352.	2.2	20
89	Somatropin treatment of spinal muscular atrophy: A placebo-controlled, double-blind crossover pilot study. Neuromuscular Disorders, 2014, 24, 134-142.	0.6	20
90	Congenital myasthenic syndrome with mild intellectual disability caused by a recurrent SLC25A1 variant. European Journal of Human Genetics, 2020, 28, 373-377.	2.8	20

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91	A case of childhood Pompe disease demonstrating phenotypic variability of p.Asp645Asn. Neuromuscular Disorders, 2004, 14, 371-374.	0.6	18
92	â€˜Why do children with cerebral palsy discontinue therapy with botulinum toxin A?â€™. Developmental Medicine and Child Neurology, 2006, 48, 319-320.	2.1	18
93	Diagnosis and New Treatment Avenues in Spinal Muscular Atrophy. Neuropediatrics, 2017, 48, 273-281.	0.6	18
94	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
95	Severe nemaline myopathy associated with consecutive mutations E74D and H75Y on a single ACTA1 allele. Neuromuscular Disorders, 2009, 19, 481-484.	0.6	16
96	A mild case of molybdenum cofactor deficiency defines an alternative route of MOCS1 protein maturation. Journal of Inherited Metabolic Disease, 2018, 41, 187-196.	3.6	16
97	Late-onset autosomal dominant limb girdle muscular dystrophy and Paget's disease of bone unlinked to the VCP gene locus. Journal of the Neurological Sciences, 2010, 291, 79-85.	0.6	15
98	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
99	Guidance in Social and Ethical Issues Related to Clinical, Diagnostic Care and Novel Therapies for Hereditary Neuromuscular Rare Diseases: â€œTranslatingâ€œ the Translational. PLOS Currents, 2013, 5, .	1.4	15
100	Mitochondrial fatty acid biosynthesis and muscle fiber plasticity in very longâ€chain acylâ€CoA dehydrogenaseâ€deficient mice. FEBS Letters, 2018, 592, 219-232.	2.8	14
101	MLIP causes recessive myopathy with rhabdomyolysis, myalgia and baseline elevatedÂ serum creatine kinase. Brain, 2021, 144, 2722-2731.	7.6	14
102	Treatment with Nusinersen â€“ Challenges Regarding the Indication for Children with SMA Type 1. Journal of Neuromuscular Diseases, 2020, 7, 41-46.	2.6	13
103	Cathepsin D as biomarker in cerebrospinal fluid of nusinersenâ€treated patients with spinal muscular atrophy. European Journal of Neurology, 2022, 29, 2084-2096.	3.3	13
104	Long-Term Follow-Up on Health-Related Quality of Life After Mechanical Circulatory Support in Children. Pediatric Critical Care Medicine, 2017, 18, 176-182.	0.5	12
105	Experiences of caregivers of children with spinal muscular atrophy participating in the expanded access program for nusinersen: a longitudinal qualitative study. Orphanet Journal of Rare Diseases, 2020, 15, 194.	2.7	12
106	Role of Pyrimidine Depletion in the Mitochondrial Cardiotoxicity of Nucleoside Analogue Reverse Transcriptase Inhibitors. Journal of Acquired Immune Deficiency Syndromes (1999), 2010, 55, 550-557.	2.1	11
107	Hereditary neuralgic amyotrophy in childhood caused by duplication within the SEPT9 gene: A family study. Cytoskeleton, 2019, 76, 131-136.	2.0	11
108	Health-Related Quality of Life in Patients with Adult-Onset Myotonic Dystrophy Type 1: A Systematic Review. Patient, 2019, 12, 365-373.	2.7	11

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109	CD59 deficiency presenting as polyneuropathy and Moyamoya syndrome with endothelial abnormalities of small brain vessels. European Journal of Paediatric Neurology, 2018, 22, 870-877.	1.6	10
110	Population pharmacokinetics-based recommendations for a single delayed or missed dose of nusinersen. Neuromuscular Disorders, 2021, 31, 310-318.	0.6	10
111	Development and Pilot Test of the Registry Evaluation and Quality Standards Tool: An Information Technologyâ€“Based Tool to Support and Review Registries. Value in Health, 2022, 25, 1390-1398.	0.3	10
112	Mitochondrial DNA mutation 14487T>C manifesting as Leberâ€™s hereditary optic neuropathy. Journal of Neurology, 2015, 262, 2776-2779.	3.6	9
113	Tasks and interfaces in primary and specialized palliative care for Duchenne muscular dystrophy â€“ A patientsâ€™ perspective. Neuromuscular Disorders, 2020, 30, 975-985.	0.6	8
114	Gene Therapy for Monogenic Inherited Disorders: Opportunities and Challenges. Deutsches Ärzteblatt International, 2020, 117, 878-885.	0.9	8
115	Zidovudine Induces Visceral Mitochondrial Toxicity and Intra-Abdominal Fat Gain in a Rodent Model of Lipodystrophy. Antiviral Therapy, 2014, 19, 783-792.	1.0	7
116	A comparative study of care practices for young boys with Duchenne muscular dystrophy between Japan and European countries: Implications of early diagnosis. Neuromuscular Disorders, 2017, 27, 894-904.	0.6	7
117	Muscle hypertrophy of the lower leg caused by L5 radiculopathy. Joint Bone Spine, 2009, 76, 562-564.	1.6	6
118	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
119	De novo variant in SCN4A causes neonatal sodium channel myotonia with general muscle stiffness and respiratory failure. Neuromuscular Disorders, 2019, 29, 907-909.	0.6	5
120	Randomisation versus prioritisation in a managed access programme: Lessons from spinal muscular atrophy. Neuromuscular Disorders, 2020, 30, 267-269.	0.6	5
121	Coagulation disorders in Duchenne muscular dystrophy? Results of a registry-based online survey. Acta Myologica, 2020, 39, 2-12.	1.5	5
122	Two novel nebulin variants in an adult patient with congenital nemaline myopathy. Neuromuscular Disorders, 2015, 25, 392-396.	0.6	4
123	Decision-Making Regarding Ventilator Support in Children with SMA Type 1â€”A Cross-Sectional Survey among Physicians. Neuropediatrics, 2019, 50, 359-366.	0.6	4
124	Health related quality of life in young, steroid-naÃ¯ve boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 1161-1168.	0.6	4
125	Postnatal gene therapy for neuromuscular diseasesÂâ€“ opportunities and limitations. Journal of Perinatal Medicine, 2021, 49, 1011-1015.	1.4	4
126	Clinical and Genetic Aspects of Juvenile Onset Pompe Disease. Neuropediatrics, 2021, , .	0.6	4

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127	Measuring muscle strength in clinical trials â€“ Authors' reply. Lancet Neurology, The, 2010, 9, 1146-1147.	10.2	3
128	Differential Analysis of Bone Density in Children and Adolescents with Neuromuscular Disorders and Cerebral Palsy. Neuropediatrics, 2015, 46, 385-391.	0.6	3
129	Parents' Perspectives on Diagnosis and Decision-Making regarding Ventilator Support in Children with SMA Type 1. Neuropediatrics, 2022, 53, 122-128.	0.6	3
130	Botulinum toxin A in children with cerebral palsy: evaluation of therapy using the Pediatric Evaluation of Disability Inventory (PEDI). Journal of Pediatric Neurology, 2015, 01, 029-034.	0.2	2
131	Outcomes in Duchenne muscular dystrophy: nature, nurture, cultureâ€“or all three?. Developmental Medicine and Child Neurology, 2017, 59, 780-781.	2.1	2
132	Author response: Discrepancy in redetermination of SMN2 copy numbers in children with SMA. Neurology, 2020, 95, 145-145.	1.1	2
133	New Perspectives in the Management of Duchenne Muscular Dystrophy. European Neurological Review, 2014, 9, 78.	0.5	2
134	The Congenital And Limb-girdle Muscular Dystrophies. Neurological Disease and Therapy, 2005, , 153-174.	0.0	2
135	Deep dynamic modeling with just two time points: Can we still allow for individual trajectories?. Biometrical Journal, 2022, , .	1.0	2
136	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
137	Facilitation of drug-resistant epilepsy and catastrophic status epilepticus in children with combined pituitary hormone deficiency. European Journal of Paediatric Neurology, 2021, 33, 99-105.	1.6	1
138	Alpers- and MNGIE-like disease with disturbed CSF folate transport and an unusual mode of genetic transmission of POLG mutations: a case report. Journal of International Child Neurology Association, 2021, 1, .	0.0	1
139	Freiburg Neuropathology Case Conference: a Ring-Enhancing Brain Lesion in an Adolescent. Klinische Neuroradiologie, 2009, 19, 238-241.	0.9	0
140	Letter to the editor: In reply to Sansone etÂal.. European Journal of Paediatric Neurology, 2018, 22, 732.	1.6	0
141	CONGENITAL MYASTHENIC SYNDROMES AND MYASTHENIA. Neuromuscular Disorders, 2018, 28, S52.	0.6	0
142	Postâ€dural puncture headacheâ€”a singleâ€centre analysis in paediatric patients with and without SMA. Acta Paediatrica, International Journal of Paediatrics, 2021, 110, 1895-1901.	1.5	0
143	Progressive Muskeldystrophien und FSHD. , 2014, , 1805-1808.		0
144	Spinale Muskelatrophien. , 2014, , 1783-1784.		0

#	Article	IF	Citations
145	Spinale Muskelatrophien. , 2015, , 1-4.		0
146	P 898. SMArtCARE: Longitudinal Data Collection of Patients with Spinal Muscular Atrophy in German-Speaking Countries. Neuropediatrics, 2018, 49, .	0.6	0
147	FV 953. Parentsâ€™ Experiences during the Compassionate Use Program (Nusinersen) for Patients with Spinal Muscular Atrophy Type 1â€”A Qualitative Interview Study. , 2018, 49, .		0
148	P 900. Decision Making Regarding Ventilator Support in Children with SMA type 1â€”A Cross-sectional Survey among German Physicians. , 2018, 49, .		0
149	FV 880. Disorders of Coagulation in Duchenne Muscular Dystrophy?â€”Results of a Registry-Based Online Questionnaire. Neuropediatrics, 2018, 49, .	0.6	0
150	Spinale Muskelatrophien. Springer Reference Medizin, 2019, , 1-3.	0.0	0
151	Spinale Muskelatrophien. Springer Reference Medizin, 2020, , 2647-2649.	0.0	0
152	SMArtCARE Real-World Data on Drug Treatment for Spinal Muscular Atrophy. , 2021, 52, .		0