Janbernd Kirschner

List of Publications by Year in Descending Order

142 papers	7,682 citations	40 h-index	86 g-index
162 ext. papers	9,895 ext. citations	6.4 avg, IF	5.49 L-index

#	Paper	IF	Citations
142	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
141	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
140	Effect of Different Corticosteroid Dosing Regimens on Clinical Outcomes in Boys With Duchenne Muscular Dystrophy: A Randomized Clinical Trial.. *JAMA - Journal of the American Medical Association*, **2022**,	27.4	6
139	SMArtCARE Real-World Data on Drug Treatment for Spinal Muscular Atrophy **2021**, 52,		
138	Safety Monitoring of Gene Therapy for Spinal Muscular Atrophy with Onasemnogene Abeparvovec -A Single Centre Experience. *Journal of Neuromuscular Diseases*, **2021**, 8, 209-216	5	9
137	Population pharmacokinetics-based recommendations for a single delayed or missed dose of nusinersen. *Neuromuscular Disorders*, **2021**, 31, 310-318	2.9	1
136	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. *Nature Communications*, **2021**, 12, 2558	17.4	4
135	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	3.8	0
134	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	
133	Post-dural puncture headache-a single-centre analysis in paediatric patients with and without SMA. *Acta Paediatrica, International Journal of Paediatrics*, **2021**, 110, 1895-1901	3.1	
132	MLIP causes recessive myopathy with rhabdomyolysis, myalgia and baseline elevated´serum creatine kinase. *Brain*, **2021**, 144, 2722-2731	11.2	5
131	Postnatal gene therapy for neuromuscular diseases´- opportunities and limitations. *Journal of Perinatal Medicine*, **2021**, 49, 1011-1015	2.7	2
130	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
129	Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. *Lancet Neurology, The*, **2020**, 19, 522-532	24.1	21
128	Zur Gentherapie der Spinalen Muskelatrophie mit Onasemnogene Abeparvovec. Stellungnahme der Gesellschaft f□ Neurop□diatrie. *Monatsschrift Fur Kinderheilkunde*, **2020**, 168, 938-941	0.2	1
127	Nusinersen in adults with 5q spinal muscular atrophy: a non-interventional, multicentre, observational cohort study. *Lancet Neurology, The*, **2020**, 19, 317-325	24.1	98
126	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

125	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
124	Life expectancy at birth in Duchenne muscular dystrophy: a systematic review and meta-analysis. *European Journal of Epidemiology*, **2020**, 35, 643-653	12.1	44
123	Author response: Discrepancy in redetermination of copy numbers in children with SMA. *Neurology*, **2020**, 95, 145	6.5	1
122	Spinale Muskelatrophien. *Springer Reference Medizin*, **2020**, 2647-2649	0	
121	Coagulation disorders in Duchenne muscular dystrophy? Results of a registry-based online survey. *Acta Myologica*, **2020**, 39, 2-12	1.6	2
120	Gene Therapy for Monogenic Inherited Disorders. *Deutsches Ärzteblatt International*, **2020**, 117, 878-885	2.5	2
119	Advances in Treatment of Spinal Muscular Atrophy - New Phenotypes, New Challenges, New Implications for Care. *Journal of Neuromuscular Diseases*, **2020**, 7, 1-13	5	47
118	Treatment with Nusinersen - Challenges Regarding the Indication for Children with SMA Type 1. *Journal of Neuromuscular Diseases*, **2020**, 7, 41-46	5	5
117	Tasks and interfaces in primary and specialized palliative care for Duchenne muscular dystrophy - A patientsQperspective. *Neuromuscular Disorders*, **2020**, 30, 975-985	2.9	0
116	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	4.2	2
115	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
114	Congenital myasthenic syndrome with mild intellectual disability caused by a recurrent SLC25A1 variant. *European Journal of Human Genetics*, **2020**, 28, 373-377	5.3	6
113	Clinical presentation and proteomic signature of patients with TANGO2 mutations. *Journal of Inherited Metabolic Disease*, **2020**, 43, 297-308	5.4	17
112	Neue Therapieoptionen und deren Implikationen f□die Transition. *Neurotransmitter*, **2019**, 30, 36-41	0.1	
111	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	2.9	188
110	De novo variant in SCN4A causes neonatal sodium channel myotonia with general muscle stiffness and respiratory failure. *Neuromuscular Disorders*, **2019**, 29, 907-909	2.9	3
109	Safety and Treatment Effects of Nusinersen in Longstanding Adult 5q-SMA Type 3 - A Prospective Observational Study. *Journal of Neuromuscular Diseases*, **2019**, 6, 453-465	5	75
108	SMArtCARE´´A platform to collect real-life outcome data of patients with spinal muscular atrophy. *Orphanet Journal of Rare Diseases*, **2019**, 14, 18	4.2	33

107	Effect and safety of treatment with ACE-inhibitor Enalapril and β-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	4.2	13
106	Quality of life of patients with spinal muscular atrophy: A systematic review. *European Journal of Paediatric Neurology*, **2019**, 23, 347-356	3.8	27
105	Health-Related Quality of Life in Patients with Adult-Onset Myotonic Dystrophy Type 1: A Systematic Review. *Patient*, **2019**, 12, 365-373	3.7	6
104	Hereditary neuralgic amyotrophy in childhood caused by duplication within the SEPT9 gene: A family study. *Cytoskeleton*, **2019**, 76, 131-136	2.4	8
103	Decision-Making Regarding Ventilator Support in Children with SMA Type 1-A Cross-Sectional Survey among Physicians. *Neuropediatrics*, **2019**, 50, 359-366	1.6	2
102	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
101	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	4.2	12
100	Spinale Muskelatrophien. *Springer Reference Medizin*, **2019**, 1-3	0	
99	Discrepancy in redetermination of copy numbers in children with SMA. *Neurology*, **2019**, 93, 267-269	6.5	23
98	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
97	Nusinersen versus Sham Control in Later-Onset Spinal Muscular Atrophy. *New England Journal of Medicine*, **2018**, 378, 625-635	59.2	617
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	5.4	9
95	A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. *Human Molecular Genetics*, **2018**, 27, 1186-1195	5.6	40
94	Variable impairment of platelet functions in patients with severe, genetically linked immune deficiencies. *Haematologica*, **2018**, 103, 540-549	6.6	25
93	Letter to the editor: In reply to Sansone et´al. *European Journal of Paediatric Neurology*, **2018**, 22, 732	3.8	
92	A checklist for clinical trials in rare disease: obstacles and anticipatory actions-lessons learned from the FOR-DMD trial. *Trials*, **2018**, 19, 291	2.8	17
91	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	5	57
90	Targeted sequencing with expanded gene profile enables high diagnostic yield in non-5q-spinal muscular atrophies. *Human Mutation*, **2018**, 39, 1284-1298	4.7	22

89	Duchenne muscular dystrophy and caregiver burden: a systematic review. *Developmental Medicine and Child Neurology*, **2018**, 60, 987-996	3.3	31
88	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
87	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
86	Mitochondrial fatty acid biosynthesis and muscle fiber plasticity in very long-chain acyl-CoA dehydrogenase-deficient mice. *FEBS Letters*, **2018**, 592, 219-232	3.8	8
85	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	3.8	24
84	Von der isolierten Optikusatrophie zur Multisystemerkrankung. *Monatsschrift Fur Kinderheilkunde*, **2018**, 166, 994-997	0.2	
83	CD59 deficiency presenting as polyneuropathy and Moyamoya syndrome with endothelial abnormalities of small brain vessels. *European Journal of Paediatric Neurology*, **2018**, 22, 870-877	3.8	6
82	Predictors of Health-Related Quality of Life in boys with Duchenne muscular dystrophy from six European countries. *Journal of Neurology*, **2017**, 264, 709-723	5.5	15
81	Characterization of pulmonary function in 10-18 year old patients with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2017**, 27, 307-314	2.9	27
80	Developing standardized corticosteroid treatment for Duchenne muscular dystrophy. *Contemporary Clinical Trials*, **2017**, 58, 34-39	2.3	39
79	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
78	Expanding Phenotype of De Novo Mutations in GNAO1: Four New Cases and Review of Literature. *Neuropediatrics*, **2017**, 48, 371-377	1.6	21
77	Outcomes in Duchenne muscular dystrophy: nature, nurture, culture-or all three?. *Developmental Medicine and Child Neurology*, **2017**, 59, 780-781	3.3	2
76	Diagnosis and New Treatment Avenues in Spinal Muscular Atrophy. *Neuropediatrics*, **2017**, 48, 273-281	1.6	13
75	Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. *New England Journal of Medicine*, **2017**, 377, 1723-1732	59.2	957
74	Long-Term Follow-Up on Health-Related Quality of Life After Mechanical Circulatory Support in Children. *Pediatric Critical Care Medicine*, **2017**, 18, 176-182	3	10
73	Ataluren in patients with nonsense mutation Duchenne muscular dystrophy (ACT DMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. *Lancet, The*, **2017**, 390, 1489-1498	40	237
72	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	2.9	4

71	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	5	84
70	A multi-source approach to determine SMA incidence and research ready population. *Journal of Neurology*, **2017**, 264, 1465-1473	5.5	56
69	Congenital Muscular Dystrophies and Myopathies: An Overview and Update. *Neuropediatrics*, **2017**, 48, 247-261	1.6	31
68	Novel homozygous RARS2 mutation in two siblings without pontocerebellar hypoplasia - further expansion of the phenotypic spectrum. *Orphanet Journal of Rare Diseases*, **2016**, 11, 140	4.2	18
67	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	11	42
66	European Cross-Sectional Survey of´Current Care Practices for Duchenne Muscular Dystrophy Reveals Regional and´Age-Dependent Differences. *Journal of Neuromuscular Diseases*, **2016**, 3, 517-527	5	36
65	Adult care for Duchenne muscular dystrophy in the UK. *Journal of Neurology*, **2015**, 262, 629-41	5.5	33
64	Decompressive craniectomy after severe traumatic brain injury in children: complications and outcome. *Neuropediatrics*, **2015**, 46, 5-12	1.6	34
63	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	
62	TPM3 deletions cause a hypercontractile congenital muscle stiffness phenotype. *Annals of Neurology*, **2015**, 78, 982-994	9.4	21
61	Respiratory chain deficiency in nonmitochondrial disease. *Neurology: Genetics*, **2015**, 1, e6	3.8	19
60	Mitochondrial DNA mutation 14487T>C manifesting as Leber© hereditary optic neuropathy. *Journal of Neurology*, **2015**, 262, 2776-9	5.5	7
59	Differential Analysis of Bone Density in Children and Adolescents with Neuromuscular Disorders and Cerebral Palsy. *Neuropediatrics*, **2015**, 46, 385-91	1.6	2
58	Two novel nebulin variants in an adult patient with congenital nemaline myopathy. *Neuromuscular Disorders*, **2015**, 25, 392-6	2.9	3
57	Spinale Muskelatrophien **2015**, 1-4		
56	Somatropin treatment of spinal muscular atrophy: a placebo-controlled, double-blind crossover pilot study. *Neuromuscular Disorders*, **2014**, 24, 134-42	2.9	18
55	Whole-body vibration training in children with Duchenne muscular dystrophy and spinal muscular atrophy. *European Journal of Paediatric Neurology*, **2014**, 18, 140-9	3.8	32
54	Zidovudine induces visceral mitochondrial toxicity and intra-abdominal fat gain in a rodent model of lipodystrophy. *Antiviral Therapy*, **2014**, 19, 783-92	1.6	7

53	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, 68-77	27.4	244
52	A homozygous splice-site mutation in CARS2 is associated with progressive myoclonic epilepsy. *Neurology*, **2014**, 83, 2183-7	6.5	42
51	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, 118-22	3.3	17
50	Myopathy in Marinesco-Sjögren syndrome links endoplasmic reticulum chaperone dysfunction to nuclear envelope pathology. *Acta Neuropathologica*, **2014**, 127, 761-77	14.3	46
49	Progressive Muskeldystrophien und FSHD **2014**, 1805-1808		
48	Spinale Muskelatrophien **2014**, 1783-1784		
47	Congenital muscular dystrophies. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2013**, 113, 1377-85	3	18
46	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	4.2	22
45	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, 215-27	7.6	59
44	Mutations in prion-like domains in hnRNPA2B1 and hnRNPA1 cause multisystem proteinopathy and ALS. *Nature*, **2013**, 495, 467-73	50.4	965
43	SIL1 mutations and clinical spectrum in patients with Marinesco-Sjogren syndrome. *Brain*, **2013**, 136, 3634-44	11.2	58
42	Pontocerebellar hypoplasia type 1: clinical spectrum and relevance of EXOSC3 mutations. *Neurology*, **2013**, 80, 438-46	6.5	65
41	Guidance in social and ethical issues related to clinical, diagnostic care and novel therapies for hereditary neuromuscular rare diseases: "translating" the translational. *PLOS Currents*, **2013**, 5,		12
40	Diagnose und Therapie der Muskeldystrophie Duchenne. *Monatsschrift Fur Kinderheilkunde*, **2012**, 160, 177-186	0.2	6
39	Muscle-fiber transdifferentiation in an experimental model of respiratory chain myopathy. *Arthritis Research and Therapy*, **2012**, 14, R233	5.7	19
38	Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. *Human Mutation*, **2012**, 33, 981-8	4.7	119
37	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. *Journal of Neurology*, **2012**, 259, 838-50	5.5	58
36	Lack of the mitochondrial protein acylglycerol kinase causes Sengers syndrome. *American Journal of Human Genetics*, **2012**, 90, 314-20	11	160

35	Adult-onset cerebellar ataxia due to mutations in CABC1/ADCK3. *Journal of Neurology, Neurosurgery and Psychiatry*, **2012**, 83, 174-8	5.5	82
34	Drug treatment of Duchenne muscular dystrophy: available evidence and perspectives. *Acta Myologica*, **2012**, 31, 4-8	1.6	35
33	Sarcoglycanopathies. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2011**, 101, 41-6	3	37
32	Hexosamine biosynthetic pathway mutations cause neuromuscular transmission defect. *American Journal of Human Genetics*, **2011**, 88, 162-72	11	124
31	Role of pyrimidine depletion in the mitochondrial cardiotoxicity of nucleoside analogue reverse transcriptase inhibitors. *Journal of Acquired Immune Deficiency Syndromes (1999)*, **2010**, 55, 550-7	3.1	10
30	Late-onset autosomal dominant limb girdle muscular dystrophy and Paget© disease of bone unlinked to the VCP gene locus. *Journal of the Neurological Sciences*, **2010**, 291, 79-85	3.2	11
29	Respiratory chain deficiency precedes the disrupted calcium homeostasis in chronic doxorubicin cardiomyopathy. *Cardiovascular Pathology*, **2010**, 19, e167-74	3.8	24
28	Oral uridine supplementation antagonizes the peripheral neuropathy and encephalopathy induced by antiretroviral nucleoside analogues. *Aids*, **2010**, 24, 345-52	3.5	17
27	Elevated satellite cell number in Duchenne muscular dystrophy. *Cell and Tissue Research*, **2010**, 340, 541-8	4.2	63
26	Treatment of Duchenne muscular dystrophy with ciclosporin A: a randomised, double-blind, placebo-controlled multicentre trial. *Lancet Neurology, The*, **2010**, 9, 1053-9	24.1	46
25	Measuring muscle strength in clinical trials □AuthorsOreply. *Lancet Neurology, The*, **2010**, 9, 1146-1147	24.1	3
24	Muscle hypertrophy of the lower leg caused by L5 radiculopathy. *Joint Bone Spine*, **2009**, 76, 562-4	2.9	5
23	Diagnose und Therapie der Muskeldystrophie Duchenne und Becker. *Medizinische Genetik*, **2009**, 21, 322-326	0.5	1
22	Freiburg neuropathology case conference: a ring-enhancing brain lesion in an adolescent. *Klinische Neuroradiologie*, **2009**, 19, 238-41		
21	Severe nemaline myopathy associated with consecutive mutations E74D and H75Y on a single ACTA1 allele. *Neuromuscular Disorders*, **2009**, 19, 481-4	2.9	13
20	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	11.5	238
19	Mitochondrial tubulopathy in tenofovir disoproxil fumarate-treated rats. *Journal of Acquired Immune Deficiency Syndromes (1999)*, **2009**, 51, 258-63	3.1	75
18	Predominant fiber atrophy and fiber type disproportion in early ullrich disease. *Muscle and Nerve*, **2008**, 38, 1184-91	3.4	25

17	Uridine supplementation antagonizes zidovudine-induced mitochondrial myopathy and hyperlactatemia in mice. *Arthritis and Rheumatism*, **2008**, 58, 318-26		26
16	Proteomic identification of FHL1 as the protein mutated in human reducing body myopathy. *Journal of Clinical Investigation*, **2008**, 118, 904-12	15.9	108
15	Uridine supplementation antagonizes zalcitabine-induced microvesicular steatohepatitis in mice. *Hepatology*, **2007**, 45, 72-9	11.2	32
14	Clinical and morphological phenotype of the filamin myopathy: a study of 31 German patients. *Brain*, **2007**, 130, 3250-64	11.2	120
13	Peripheral nerve demyelination caused by a mutant Rho GTPase guanine nucleotide exchange factor, frabin/FGD4. *American Journal of Human Genetics*, **2007**, 81, 158-64	11	107
12	Why do children with cerebral palsy discontinue therapy with botulinum toxin A? *Developmental Medicine and Child Neurology*, **2006**, 48, 319-20	3.3	14
11	Infections and vaccinations preceding childhood Guillain-Barré syndrome: a prospective study. *European Journal of Pediatrics*, **2006**, 165, 605-12	4.1	28
10	Ullrich congenital muscular dystrophy: connective tissue abnormalities in the skin support overlap with Ehlers-Danlos syndromes **2005**, 132A, 296-301		74
9	p.S143F mutation in lamin A/C: a new phenotype combining myopathy and progeria. *Annals of Neurology*, **2005**, 57, 148-51	9.4	51
8	Intravenously administered immunoglobulin in the treatment of childhood Guillain-Barré syndrome: a randomized trial. *Pediatrics*, **2005**, 116, 8-14	7.4	122
7	The Congenital And Limb-girdle Muscular Dystrophies. *Neurological Disease and Therapy*, **2005**, 153-174		
6	The congenital and limb-girdle muscular dystrophies: sharpening the focus, blurring the boundaries. *Archives of Neurology*, **2004**, 61, 189-99		52
5	A case of childhood Pompe disease demonstrating phenotypic variability of p.Asp645Asn. *Neuromuscular Disorders*, **2004**, 14, 371-4	2.9	16
4	Botulinum toxin treatment in cerebral palsy: evidence for a new treatment option. *Journal of Neurology*, **2001**, 248 Suppl 1, 28-30	5.5	18
3	Evaluation of botulinum toxin A therapy in children with adductor spasm by gross motor function measure. *Journal of Child Neurology*, **2000**, 15, 214-7	2.5	49
2	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	6	14
1	Absence of transcallosal inhibition in adolescents with diplegic cerebral palsy. *Muscle and Nerve*, **1999**, 22, 255-7	3.4	29