Brunhilde Wirth

List of Publications by Year
in descending order

164 papers	15,159 citations	20759 60 h-index	18606 119 g-index
171 all docs	171 docs citations	171 times ranked	9200 citing authors

#	Article	IF	Citations
1	A single nucleotide in the SMN gene regulates splicing and is responsible for spinal muscular atrophy. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 6307-6311.	3.3	1,320
2	Quantitative Analyses of SMN1 and SMN2 Based on Real-Time LightCycler PCR: Fast and Highly Reliable Carrier Testing and Prediction of Severity of Spinal Muscular Atrophy. American Journal of Human Genetics, 2002, 70, 358-368.	2.6	852
3	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
4	An update of the mutation spectrum of the survival motor neuron gene (SMN1) in autosomal recessive spinal muscular atrophy (SMA). Human Mutation, 2000, 15, 228-237.	1.1	554
5	SMN oligomerization defect correlates with spinal muscular atrophy severity. Nature Genetics, 1998, 19, 63-66.	9.4	470
6	Plastin 3 Is a Protective Modifier of Autosomal Recessive Spinal Muscular Atrophy. Science, 2008, 320, 524-527.	6.0	434
7	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
8	Quantitative Analysis of Survival Motor Neuron Copies: Identification of Subtle SMN1 Mutations in Patients with Spinal Muscular Atrophy, Genotype-Phenotype Correlation, and Implications for Genetic Counseling. American Journal of Human Genetics, 1999, 64, 1340-1356.	2.6	335
9	Valproic acid increases the SMN2 protein level: a well-known drug as a potential therapy for spinal muscular atrophy. Human Molecular Genetics, 2003, 12, 2481-2489.	1.4	333
10	Exome Sequencing Identifies Truncating Mutations in Human SERPINF1 in Autosomal-Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2011, 88, 362-371.	2.6	316
11	Htra2-beta 1 stimulates an exonic splicing enhancer and can restore full-length SMN expression to survival motor neuron 2 (SMN2). Proceedings of the National Academy of Sciences of the United States of America, 2000, 97, 9618-9623.	3.3	299
12	Molecular analysis of spinal muscular atrophy and modification of the phenotype by SMN2. Genetics in Medicine, 2002, 4, 20-26.	1.1	296
13	Mildly affected patients with spinal muscular atrophy are partially protected by an increased SMN2 copy number. Human Genetics, 2006, 119, 422-428.	1.8	292
14	Localizing multiple X chromosome-linked retinitis pigmentosa loci using multilocus homogeneity tests.. Proceedings of the National Academy of Sciences of the United States of America, 1990, 87, 701-704.	3.3	283
15	Molecular analysis of candidate genes on chromosome 5q13 in autosomal recessive spinal muscular atrophy: evidence of homozygous deletions of the SMN gene in unaffected individuals. Human Molecular Genetics, 1995, 4, 1927-1933.	1.4	267
16	Mapping of the gene for autosomal recessive polycystic kidney disease (ARPKD) to chromosome 6p21â€“cen. Nature Genetics, 1994, 7, 429-432.	9.4	253
17	A Mutation in the 5â€²-UTR of IFITM5 Creates an In-Frame Start Codon and Causes Autosomal-Dominant Osteogenesis Imperfecta Type V with Hyperplastic Callus. American Journal of Human Genetics, 2012, 91, 349-357.	2.6	205
18	SAHA ameliorates the SMA phenotype in two mouse models for spinal muscular atrophy. Human Molecular Genetics, 2010, 19, 1492-1506.	1.4	195

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19	Mitochondrial dysfunction, peroxidation damage and changes in glutathione metabolism in PARK6. Neurobiology of Disease, 2007, 25, 401-411.	2.1	180
20	PLS3 Mutations in X-Linked Osteoporosis with Fractures. New England Journal of Medicine, 2013, 369, 1529-1536.	13.9	171
21	hnRNP-G promotes exon 7 inclusion of survival motor neuron (SMN) via direct interaction with Htra2-beta1. Human Molecular Genetics, 2002, 11, 2037-2049.	1.4	167
22	Congenital heart disease is a feature of severe infantile spinal muscular atrophy. Journal of Medical Genetics, 2008, 45, 635-638.	1.5	158
23	Neurocalcin Delta Suppression Protects against Spinal Muscular Atrophy in Humans and across Species by Restoring Impaired Endocytosis. American Journal of Human Genetics, 2017, 100, 297-315.	2.6	156
24	De Novo Rearrangements Found in 2% of Index Patients with Spinal Muscular Atrophy: Mutational Mechanisms, Parental Origin, Mutation Rate, and Implications for Genetic Counseling. American Journal of Human Genetics, 1997, 61, 1102-1111.	2.6	155
25	Histone deacetylase inhibitors: possible implications for neurodegenerative disorders. Expert Opinion on Investigational Drugs, 2008, 17, 169-184.	1.9	154
26	The Power of Human Protective Modifiers: PLS3 and CORO1C Unravel Impaired Endocytosis in Spinal Muscular Atrophy and Rescue SMA Phenotype. American Journal of Human Genetics, 2016, 99, 647-665.	2.6	154
27	Dysregulation of ubiquitin homeostasis and Î²-catenin signaling promote spinal muscular atrophy. Journal of Clinical Investigation, 2014, 124, 1821-1834.	3.9	151
28	Mutations in BICD2, which Encodes a Golgin and Important Motor Adaptor, Cause Congenital Autosomal-Dominant Spinal Muscular Atrophy. American Journal of Human Genetics, 2013, 92, 946-954.	2.6	150
29	In vitro and ex vivo evaluation of second-generation histone deacetylase inhibitors for the treatment of spinal muscular atrophy. Journal of Neurochemistry, 2006, 98, 193-202.	2.1	140
30	Twenty-Five Years of Spinal Muscular Atrophy Research: From Phenotype to Genotype to Therapy, and What Comes Next. Annual Review of Genomics and Human Genetics, 2020, 21, 231-261.	2.5	134
31	In vivo activation ofSMNin spinal muscular atrophy carriers and patients treated with valproate. Annals of Neurology, 2006, 59, 970-975.	2.8	133
32	Identification of a candidate modifying gene for spinal muscular atrophy by comparative genomics. Nature Genetics, 1998, 20, 83-86.	9.4	129
33	SMN regulates axonal local translation via miR-183/mTOR pathway. Human Molecular Genetics, 2014, 23, 6318-6331.	1.4	125
34	Acetylcholine Receptor Pathway Mutations Explain Various Fetal Akinesia Deformation Sequence Disorders. American Journal of Human Genetics, 2008, 82, 464-476.	2.6	124
35	The spinal muscular atrophy disease protein SMN is linked to the rho-kinase pathway via profilin. Human Molecular Genetics, 2011, 20, 4865-4878.	1.4	120
36	Clinical Spectrum and Diagnostic Criteria of Infantile Spinal Muscular Atrophy: Further Delineation on the Basis of SMN Gene Deletion Findings. Neuropediatrics, 1996, 27, 8-15.	0.3	119

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37	The benzamide M344, a novel histone deacetylase inhibitor, significantly increases SMN2 RNA/protein levels in spinal muscular atrophy cells. Human Genetics, 2006, 120, 101-110.	1.8	117
38	Survival motor neuron gene 2 silencing by DNA methylation correlates with spinal muscular atrophy disease severity and can be bypassed by histone deacetylase inhibition. Human Molecular Genetics, 2009, 18, 304-317.	1.4	116
39	Plastin 3 ameliorates spinal muscular atrophy via delayed axon pruning and improves neuromuscular junction functionality. Human Molecular Genetics, 2013, 22, 1328-1347.	1.4	116
40	Missense Mutations in Exon 6 of the Survival Motor Neuron Gene in Patients with Spinal Muscular Atrophy (SMA). Human Molecular Genetics, 1997, 6, 821-825.	1.4	110
41	Spinal Muscular Atrophy: From Gene to Therapy. Seminars in Pediatric Neurology, 2006, 13, 121-131.	1.0	110
42	Spinal muscular atrophyâ€"clinical and genetic correlations. Neuromuscular Disorders, 1997, 7, 202-207.	0.3	108
43	Evidence for a modifying pathway in SMA discordant families: reduced SMN level decreases the amount of its interacting partners and Htra2-beta1. Human Genetics, 2003, 114, 11-21.	1.8	108
44	One Year of Newborn Screening for SMA â€“ Results of a German Pilot Project. Journal of Neuromuscular Diseases, 2019, 6, 503-515.	1.1	105
45	Allelic association and deletions in autosomal recessive proximal spinal muscular atrophy: association of marker genotype with disease severity and candidate cDNAs. Human Molecular Genetics, 1995, 4, 1273-1284.	1.4	101
46	Molecular and functional analysis of intragenic SMN1 mutations in patients with spinal muscular atrophy. Human Mutation, 2005, 25, 64-71.	1.1	101
47	LBH589 induces up to 10-fold SMN protein levels by several independent mechanisms and is effective even in cells from SMA patients non-responsive to valproate. Human Molecular Genetics, 2009, 18, 3645-3658.	1.4	100
48	Co-regulation of survival of motor neuron (SMN) protein and its interactor SIP1 during development and in spinal muscular atrophy. Human Molecular Genetics, 2001, 10, 497-505.	1.4	94
49	Valproic acid induces antioxidant effects in X-linked adrenoleukodystrophy. Human Molecular Genetics, 2010, 19, 2005-2014.	1.4	90
50	Reversible molecular pathology of skeletal muscle in spinal muscular atrophy. Human Molecular Genetics, 2011, 20, 4334-4344.	1.4	89
51	Spinal Muscular Atrophy: In the Challenge Lies a Solution. Trends in Neurosciences, 2021, 44, 306-322.	4.2	85
52	Newborn screening for spinal muscular atrophy in Germany: clinical results after 2 years. Orphanet Journal of Rare Diseases, 2021, 16, 153.	1.2	81
53	The gene for autosomal dominant polycystic kidney disease lies in a 750-kb CpG-rich region. Genomics, 1992, 13, 144-151.	1.3	80
54	Diaphragmatic Spinal Muscular Atrophy with Respiratory Distress Is Heterogeneous, and One Form Is Linked to Chromosome 11q13-q21. American Journal of Human Genetics, 1999, 65, 1459-1462.	2.6	80

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55	How genetic modifiers influence the phenotype of spinal muscular atrophy and suggest future therapeutic approaches. Current Opinion in Genetics and Development, 2013, 23, 330-338.	1.5	79
56	Neuromuscular defects and breathing disorders in a new mouse model of spinal muscular atrophy. Neurobiology of Disease, 2010, 38, 125-135.	2.1	71
57	Extended phenotype of pontocerebellar hypoplasia with infantile spinal muscular atrophy. , 2003, 117A, 10-17.		68
58	Genotypeâ€“phenotype studies in infantile spinal muscular atrophy (SMA) type I in Germany: implications for clinical trials and genetic counselling. Clinical Genetics, 2009, 76, 168-178.	1.0	67
59	Identification of Evolutionarily Conserved Exons as Regulated Targets for the Splicing Activator Tra2Î² in Development. PLoS Genetics, 2011, 7, e1002390.	1.5	65
60	Biallelic Loss of Proprioception-Related PIEZO2 Causes Muscular Atrophy with Perinatal Respiratory Distress, Arthrogryposis, and Scoliosis. American Journal of Human Genetics, 2016, 99, 1206-1216.	2.6	65
61	Best practice guidelines for molecular analysis in spinal muscular atrophy. European Journal of Human Genetics, 2001, 9, 484-491.	1.4	63
62	Cognitive training with and without additional physical activity in healthy older adults: cognitive effects, neurobiological mechanisms, and prediction of training success. Frontiers in Aging Neuroscience, 2015, 7, 187.	1.7	63
63	Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. American Journal of Human Genetics, 2018, 103, 431-439.	2.6	62
64	Genetic basis of adult-onset spinal muscular atrophy. Lancet, The, 1995, 346, 1162.	6.3	57
65	Severe SMA mice show organ impairment that cannot be rescued by therapy with the HDACi JNJ-26481585. European Journal of Human Genetics, 2013, 21, 643-652.	1.4	55
66	A Multicopy Dinucleotide Marker That Maps Close to the Spinal Muscular Atrophy Gene. Genomics, 1994, 21, 394-402.	1.3	54
67	Deficiency of the splicing factor Sfrs10 results in early embryonic lethality in mice and has no impact on full-length SMN/Smn splicing. Human Molecular Genetics, 2010, 19, 2154-2167.	1.4	53
68	VPA response in SMA is suppressed by the fatty acid translocase CD36. Human Molecular Genetics, 2013, 22, 398-407.	1.4	50
69	Moving towards treatments for spinal muscular atrophy: hopes and limits. Expert Opinion on Emerging Drugs, 2015, 20, 353-356.	1.0	50
70	218th ENMC International Workshop:. Neuromuscular Disorders, 2017, 27, 596-605.	0.3	49
71	CHP1 reduction ameliorates spinal muscular atrophy pathology by restoring calcineurin activity and endocytosis. Brain, 2018, 141, 2343-2361.	3.7	49
72	Homozygous mutations in <scp>VAMP</scp>1 cause a presynaptic congenital myasthenic syndrome. Annals of Neurology, 2017, 81, 597-603.	2.8	48

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73	The colour theorems of Brooks and Gallai extended. Discrete Mathematics, 1996, 162, 299-303.	0.4	46
74	Large Linkage Analysis in 100 Families with Autosomal Recessive Spinal Muscular Atrophy (SMA) and 11 CEPH Families Using 15 Polymorphic Loci in the Region 5q11.2-q13.3. Genomics, 1994, 20, 84-93.	1.3	45
75	An Â140-kb deletion associated with feline spinal muscular atrophy implies an essential LIX1 function for motor neuron survival. Genome Research, 2006, 16, 1084-1090.	2.4	43
76	Discrepancy in redetermination of SMN2 copy numbers in children with SMA. Neurology, 2019, 93, 267-269.	1.5	43
77	High-throughput genetic newborn screening for spinal muscular atrophy by rapid nucleic acid extraction from dried blood spots and 384-well qPCR. European Journal of Human Genetics, 2020, 28, 23-30.	1.4	43
78	Two different genes for X-linked retinitis pigmentosa. Genomics, 1988, 2, 263-266.	1.3	42
79	Targeted sequencing with expanded gene profile enables high diagnostic yield in non-5q-spinal muscular atrophies. Human Mutation, 2018, 39, 1284-1298.	1.1	42
80	Plastin 3 influences bone homeostasis through regulation of osteoclast activity. Human Molecular Genetics, 2018, 27, 4249-4262.	1.4	41
81	Comparative Sequence Analysis of the Mouse and Human Lgn1/SMA Interval. Genomics, 1999, 60, 137-151.	1.3	39
82	Infants Diagnosed with Spinal Muscular Atrophy and 4 SMN2 Copies through Newborn Screening â€“ Opportunity or Burden?1. Journal of Neuromuscular Diseases, 2020, 7, 109-117.	1.1	39
83	Spinal Muscular Atrophy and Therapeutic Prospects. Progress in Molecular and Subcellular Biology, 2006, 44, 109-132.	0.9	39
84	Fine genetic localization of the gene for autosomal dominant polycystic kidney disease (PKD1) with respect to physically mapped markers. Genomics, 1992, 13, 152-158.	1.3	37
85	Axonal Neuropathy and Predominance of Type II Myofibers in Infantile Spinal Muscular Atrophy. Journal of Child Neurology, 1998, 13, 327-331.	0.7	37
86	Dominant spinal muscular atrophy due to BICD2: a novel mutation refines the phenotype. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 590-592.	0.9	37
87	Autosomal recessive proximal spinal muscular atrophy in 101 sibs out of 48 families: Clinical picture, influence of gender, and genetic implications. American Journal of Medical Genetics Part A, 1994, 51, 70-76.	2.4	36
88	Plastin 3 is upregulated in iPSC-derived motoneurons from asymptomatic SMN1-deleted individuals. Cellular and Molecular Life Sciences, 2016, 73, 2089-2104.	2.4	36
89	Dominant spinal muscular atrophy is caused by mutations in BICD2, an important golgin protein. Frontiers in Neuroscience, 2015, 9, 401.	1.4	35
90	Phenotypic extremes of BICD2-opathies: from lethal, congenital muscular atrophy with arthrogryposis to asymptomatic with subclinical features. European Journal of Human Genetics, 2017, 25, 1040-1048.	1.4	35

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91	Advances in understanding the role of disease-associated proteins in spinal muscular atrophy. Expert Review of Proteomics, 2017, 14, 581-592.	1.3	35
92	Mutations of the LMNA gene can mimic autosomal dominant proximal spinal muscular atrophy. Neurogenetics, 2007, 8, 137-142.	0.7	33
93	Fine Mapping and Narrowing of the Genetic Interval of the Spinal Muscular Atrophy Region by Linkage Studies. Genomics, 1993, 15, 113-118.	1.3	32
94	Nonsense-mediated messenger RNA decay of survival motor neuron 1 causes spinal muscular atrophy. Human Genetics, 2008, 123, 141-153.	1.8	31
95	Splicing factor TRA2B is required for neural progenitor survival. Journal of Comparative Neurology, 2014, 522, 372-392.	0.9	31
96	Plastin 3 in health and disease: a matter of balance. Cellular and Molecular Life Sciences, 2021, 78, 5275-5301.	2.4	31
97	Prenatal prediction in families with autosomal recessive proximal spinal muscular atrophy (5q11.2â€“q13.3): Molecular genetics and clinical experience in 109 cases. Prenatal Diagnosis, 1995, 15, 407-417.	1.1	30
98	UBA1/GARS-dependent pathways drive sensory-motor connectivity defects in spinal muscular atrophy. Brain, 2018, 141, 2878-2894.	3.7	29
99	Biallelic variant in AGTPBP1 causes infantile lower motor neuron degeneration and cerebellar atrophy. American Journal of Medical Genetics, Part A, 2019, 179, 1580-1584.	0.7	29
100	Childhood manifestation of autosomal dominant polycystic kidney disease: no evidence for genetic heterogeneity. Clinical Genetics, 1989, 35, 13-19.	1.0	28
101	Expanding the phenotype of BICD2 mutations toward skeletal muscle involvement. Neurology, 2016, 87, 2235-2243.	1.5	28
102	Neuronal-Specific Deficiency of the Splicing Factor Tra2b Causes Apoptosis in Neurogenic Areas of the Developing Mouse Brain. PLoS ONE, 2014, 9, e89020.	1.1	28
103	X-linked infantile spinal muscular atrophy: Clinical definition and molecular mapping. Genetics in Medicine, 2007, 9, 52-60.	1.1	27
104	Neurocalcin Delta Knockout Impairs Adult Neurogenesis Whereas Half Reduction Is Not Pathological. Frontiers in Molecular Neuroscience, 2019, 12, 19.	1.4	27
105	Effects of a Cognitive Training With and Without Additional Physical Activity in Healthy Older Adults: A Follow-Up 1 Year After a Randomized Controlled Trial. Frontiers in Aging Neuroscience, 2018, 10, 407.	1.7	26
106	NCALD Antisense Oligonucleotide Therapy in Addition to Nusinersen further Ameliorates Spinal Muscular Atrophy in Mice. American Journal of Human Genetics, 2019, 105, 221-230.	2.6	26
107	Mitochondrial defects in the respiratory complex I contribute to impaired translational initiation via ROS and energy homeostasis in SMA motor neurons. Acta Neuropathologica Communications, 2020, 8, 223.	2.4	26
108	Exclusion of the gene locus for spinal muscular atrophy on chromosome 5q in a family with infantile olivopontocerebellar atrophy (OPCA) and anterior horn cell degeneration. Neuromuscular Disorders, 1995, 5, 19-23.	0.3	24

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109	Human and mouse RAD17 genes: identification, localization, genomic structure and histological expression pattern in normal testis and seminoma. Human Genetics, 1999, 105, 17-27.	1.8	24
110	Spinal muscular atrophy: state-of-the-art and therapeutic perspectives. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2002, 3, 87-95.	1.4	23
111	Increased levels of <scp>UCHL</scp>1 are a compensatory response to disrupted ubiquitin homeostasis in spinal muscular atrophy and do not represent a viable therapeutic target. Neuropathology and Applied Neurobiology, 2014, 40, 873-887.	1.8	23
112	Biallelic CHP1 mutation causes human autosomal recessive ataxia by impairing NHE1 function. Neurology: Genetics, 2018, 4, e209.	0.9	23
113	Clinical trial of Lâ€Carnitine and valproic acid in spinal muscular atrophy type I. Muscle and Nerve, 2018, 57, 193-199.	1.0	23
114	Reciprocal Connections Between Cortex and Thalamus Contribute to Retinal Axon Targeting to Dorsal Lateral Geniculate Nucleus. Cerebral Cortex, 2018, 28, 1168-1182.	1.6	22
115	Central synaptopathy is the most conserved feature of motor circuit pathology across spinal muscular atrophy mouse models. IScience, 2021, 24, 103376.	1.9	21
116	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	3.7	20
117	The Transcription Factor-like Nuclear Regulator (TFNR) Contains a Novel 55-Amino-Acid Motif Repeated Nine Times and Maps Closely to SMN1. Genomics, 2000, 70, 315-326.	1.3	19
118	Tra2Î² Protein Is Required for Tissue-specific Splicing of a Smooth Muscle Myosin Phosphatase Targeting Subunit Alternative Exon. Journal of Biological Chemistry, 2012, 287, 16575-16585.	1.6	19
119	Investigational therapies for the treatment of spinal muscular atrophy. Expert Opinion on Investigational Drugs, 2015, 24, 867-881.	1.9	19
120	Autosomal recessive and dominant forms of polycystic kidney disease are not allelic. Human Genetics, 1987, 77, 221-222.	1.8	18
121	Late Manifestation of Autosomal-Recessive Polycystic Kidney Disease in Two Sisters. American Journal of Nephrology, 1988, 8, 194-197.	1.4	18
122	Muscle regulates mTOR dependent axonal local translation in motor neurons via CTRP3 secretion: implications for a neuromuscular disorder, spinal muscular atrophy. Acta Neuropathologica Communications, 2019, 7, 154.	2.4	18
123	Exome sequencing identifies Laing distal myopathy MYH7 mutation in a Roma family previously diagnosed with distal neuronopathy. Neuromuscular Disorders, 2014, 24, 156-161.	0.3	17
124	Metalloprotease-mediated cleavage of PlexinD1 and its sequestration to actin rods in the motoneuron disease spinal muscular atrophy (SMA). Human Molecular Genetics, 2017, 26, 3946-3959.	1.4	17
125	Linkage analysis in X-linked ichthyosis (steroid sulfatase deficiency). Human Genetics, 1988, 80, 191-192.	1.8	16
126	No evidence for DUP25 in patients with panic disorder using a quantitative real-time PCR approach. Human Genetics, 2003, 114, 115-117.	1.8	16

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127	Neuronal activity regulates DROSHA via autophagy in spinal muscular atrophy. Scientific Reports, 2018, 8, 7907.	1.6	16
128	Histone Acetylation as a Potential Therapeutic Target in Motor Neuron Degenerative Diseases. Current Pharmaceutical Design, 2013, 19, 5093-5104.	0.9	16
129	TRA2Î² controls Mypt1 exon 24 splicing in the developmental maturation of mouse mesenteric artery smooth muscle. American Journal of Physiology - Cell Physiology, 2015, 308, C289-C296.	2.1	15
130	Novel insights into SMALED2: BICD2 mutations increase microtubule stability and cause defects in axonal and NMJ development. Human Molecular Genetics, 2018, 27, 1772-1784.	1.4	15
131	Report of a novel ATP7A mutation causing distal motor neuropathy. Neuromuscular Disorders, 2019, 29, 776-785.	0.3	15
132	De Novo and Inherited Variants in GBF1 are Associated with Axonal Neuropathy Caused by Golgi Fragmentation. American Journal of Human Genetics, 2020, 107, 763-777.	2.6	14
133	Increasing SMN levels using the histone deacetylase inhibitor SAHA ameliorates defects in skeletal muscle microvasculature in a mouse model of severe spinal muscular atrophy. Neuroscience Letters, 2013, 544, 100-104.	1.0	13
134	Biallelic MCM3AP mutations cause Charcot-Marie-Tooth neuropathy with variable clinical presentation. Brain, 2017, 140, e65-e65.	3.7	13
135	Genetic modifiers ameliorate endocytic and neuromuscular defects in a model of spinal muscular atrophy. BMC Biology, 2020, 18, 127.	1.7	13
136	Cathepsin D as biomarker in cerebrospinal fluid of nusinersenâ€treated patients with spinal muscular atrophy. European Journal of Neurology, 2022, 29, 2084-2096.	1.7	13
137	Evaluation of potential effects of Plastin 3 overexpression and low-dose SMN-antisense oligonucleotides on putative biomarkers in spinal muscular atrophy mice. PLoS ONE, 2018, 13, e0203398.	1.1	11
138	PRUNE1: a disease-causing gene for secondary microcephaly. Brain, 2017, 140, e61-e61.	3.7	10
139	De novo DNM1L variant presenting with severe muscular atrophy, dystonia and sensory neuropathy. European Journal of Medical Genetics, 2021, 64, 104134.	0.7	9
140	Spinal muscular atrophy (5qSMA): best practice of diagnostics, newborn screening and therapy. Medizinische Genetik, 2020, 32, 263-272.	0.1	9
141	Xmnl polymorphism of the human STS gene. Nucleic Acids Research, 1989, 17, 3326-3326.	6.5	8
142	Hereditary polyneuropathy with optic atrophy due to PDXK variant leading to impaired Vitamin B6 metabolism. Neuromuscular Disorders, 2020, 30, 583-589.	0.3	7
143	Expression and Localization of Thrombospondins, Plastin 3, and STIM1 in Different Cartilage Compartments of the Osteoarthritic Varus Knee. International Journal of Molecular Sciences, 2021, 22, 3073.	1.8	7
144	Maximum bite force in patients with spinal muscular atrophy during the first year of nusinersen therapy - A pilot study. Acta Myologica, 2020, 39, 83-89.	1.5	7

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145	Exclusion of Htra2-Î²1, an up-regulator of full-length SMN2 transcript, as a modifying gene for spinal muscular atrophy. Human Genetics, 2000, 107, 554-558.	1.8	6
146	Reply to Ogino and Wilson. American Journal of Human Genetics, 2002, 70, 1598-1599.	2.6	6
147	Genomic variants causing mitochondrial dysfunction are common in hereditary lower motor neuron disease. Human Mutation, 2021, 42, 460-472.	1.1	6
148	Analysis of the SMN and NAIP Genes in Slovak Spinal Muscular Atrophy Patients. Human Heredity, 2000, 50, 171-174.	0.4	5
149	The zinc finger protein ZNF297B interacts with BDP1, a subunit of TFIIIB. Biological Chemistry, 2006, 387, 277-84.	1.2	4
150	Drug discovery for spinal muscular atrophy. Expert Opinion on Drug Discovery, 2007, 2, 437-451.	2.5	4
151	PLS3 Overexpression Delays Ataxia in Chp1 Mutant Mice. Frontiers in Neuroscience, 2019, 13, 993.	1.4	4
152	Novel variants broaden the phenotypic spectrum of PLEKHG5 â€associated neuropathies. European Journal of Neurology, 2021, 28, 1344-1355.	1.7	4
153	Mapping of the amniotic fluid proteome of fetuses with congenital anomalies of the kidney and urinary tract identifies plastin 3 as a protein involved in glomerular integrity. Journal of Pathology, 2021, 254, 575-588.	2.1	4
154	Giant axonal neuropathy: a differential diagnosis of consideration. Turkish Journal of Pediatrics, 2019, 61, 275.	0.3	4
155	Tongue fasciculations in an infant with spinal muscular atrophy type 1. Clinical Case Reports (discontinued), 2015, 3, 832-834.	0.2	3
156	Author response: Discrepancy in redetermination of SMN2 copy numbers in children with SMA. Neurology, 2020, 95, 145-145.	1.5	2
157	Transformer (Tra2Î²): master regulator of myosin phosphatase alternative splicing and smooth muscle responses to NO/cGMP signaling. BMC Pharmacology, 2011, 11, .	0.4	1
158	Spinale Muskelatrophien. , 2000, , 60-91.		1
159	Twenty different alleles at the locus D5S683 on 5q23â€“31. Human Molecular Genetics, 1993, 2, 2204-2204.	1.4	0
160	Dinucleotide repeat polymorphism at the D12S371 locus. Human Molecular Genetics, 1993, 2, 1754-1754.	1.4	0
161	A Paucisymptomatic Neuromuscular Disease Mimicking Type III 5q-SMA With Complex Rearrangements in the SMN Gene. Journal of Child Neurology, 2014, 29, 254-259.	0.7	0
162	P385â€…Hypotonic infant with riboflavin transporter deficiency due to slc52a2 mutations. , 2017, , .		0

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163	Commemoration of 15 years ESHG SPC member and chair from 2009 to 2016. European Journal of Human Genetics, 2017, 25, S37-S38.	1.4	0
164	Hereditary nodo-paranodopathies: genomic variants, not just autoantibodies, hit the protein. Brain, 2019, 142, 2895-2897.	3.7	0