Christel Depienne

List of Publications by Year
in descending order

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168 papers	12,887 citations	17429 63 h-index	30058 103 g-index
189 all docs	189 docs citations	189 times ranked	18067 citing authors

#	Article	IF	Citations
1	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
2	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. Cell, 2019, 179, 1469-1482.e11.	13.5	935
3	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	1.5	501
4	De Novo Mutations in Synaptic Transmission Genes Including DNM1 Cause Epileptic Encephalopathies. American Journal of Human Genetics, 2014, 95, 360-370.	2.6	388
5	Sporadic Infantile Epileptic Encephalopathy Caused by Mutations in PCDH19 Resembles Dravet Syndrome but Mainly Affects Females. PLoS Genetics, 2009, 5, e1000381.	1.5	304
6	Spectrum of SCN1A gene mutations associated with Dravet syndrome: analysis of 333 patients. Journal of Medical Genetics, 2008, 46, 183-191.	1.5	302
7	The phenotypic spectrum of SCN8A encephalopathy. Neurology, 2015, 84, 480-489.	1.5	246
8	Interrogating the Genetic Determinants of Tourette's Syndrome and Other Tic Disorders Through Genome-Wide Association Studies. American Journal of Psychiatry, 2019, 176, 217-227.	4.0	242
9	Mutations in SPG11 are frequent in autosomal recessive spastic paraplegia with thin corpus callosum, cognitive decline and lower motor neuron degeneration. Brain, 2008, 131, 772-784.	3.7	206
10	De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nature Genetics, 2014, 46, 640-645.	9.4	192
11	GRIN2Bencephalopathy: novel findings on phenotype, variant clustering, functional consequences and treatment aspects. Journal of Medical Genetics, 2017, 54, 460-470.	1.5	190
12	De Novo Loss-of-Function Mutations in CHD2 Cause a Fever-Sensitive Myoclonic Epileptic Encephalopathy Sharing Features with Dravet Syndrome. American Journal of Human Genetics, 2013, 93, 967-975.	2.6	188
13	De novo mutations in ATP1A2 and CACNA1A are frequent in early-onset sporadic hemiplegic migraine. Neurology, 2010, 75, 967-972.	1.5	179
14	Alteration of Fatty-Acid-Metabolizing Enzymes Affects Mitochondrial Form and Function in Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1051-1064.	2.6	179
15	Annonacin, a Natural Mitochondrial Complex I Inhibitor, Causes Tau Pathology in Cultured Neurons. Journal of Neuroscience, 2007, 27, 7827-7837.	1.7	176
16	Hereditary spastic paraplegias: an update. Current Opinion in Neurology, 2007, 20, 674-680.	1.8	174
17	30 years of repeat expansion disorders: What have we learned and what are the remaining challenges?. American Journal of Human Genetics, 2021, 108, 764-785.	2.6	170
18	Delineating the GRIN1 phenotypic spectrum. Neurology, 2016, 86, 2171-2178.	1.5	157

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19	Brain white matter oedema due to ClC-2 chloride channel deficiency: an observational analytical study. Lancet Neurology, The, 2013, 12, 659-668.	4.9	152
20	Spastic paraplegia gene 7 in patients with spasticity and/or optic neuropathy. Brain, 2012, 135, 2980-2993.	3.7	148
21	Genetic Analysis of Inherited Leukodystrophies. JAMA Neurology, 2013, 70, 875.	4.5	147
22	Clinical and genetic heterogeneity in hereditary spastic paraplegias: From SPG1 to SPG72 and still counting. Revue Neurologique, 2015, 171, 505-530.	0.6	143
23	Rare Copy Number Variants in NRXN1 and CNTN6 Increase Risk for Tourette Syndrome. Neuron, 2017, 94, 1101-1111.e7.	3.8	137
24	Genetic and neurodevelopmental spectrum ofSYNGAP1-associated intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 511-522.	1.5	135
25	Screening for Genomic Rearrangements and Methylation Abnormalities of the 15q11-q13 Region in Autism Spectrum Disorders. Biological Psychiatry, 2009, 66, 349-359.	0.7	133
26	Mechanisms for variable expressivity of inherited SCN1A mutations causing Dravet syndrome. Journal of Medical Genetics, 2010, 47, 404-410.	1.5	130
27	DEPDC5 mutations in families presenting as autosomal dominant nocturnal frontal lobe epilepsy. Neurology, 2014, 82, 2101-2106.	1.5	126
28	PRRT2 mutations cause hemiplegic migraine. Neurology, 2012, 79, 2122-2124.	1.5	118
29	PCDH19-related infantile epileptic encephalopathy: An unusual X-linked inheritance disorder. Human Mutation, 2012, 33, 627-634.	1.1	118
30	Complicated forms of autosomal dominant hereditary spastic paraplegia are frequent in SPG10. Human Mutation, 2009, 30, E376-E385.	1.1	115
31	Unstable TTTTA/TTTCA expansions in MARCH6 are associated with Familial Adult Myoclonic Epilepsy type 3. Nature Communications, 2019, 10, 4919.	5.8	111
32	Mutations and deletions in PCDH19 account for various familial or isolated epilepsies in females. Human Mutation, 2011, 32, E1959-E1975.	1.1	109
33	SPG3A is the most frequent cause of hereditary spastic paraplegia with onset before age 10 years. Neurology, 2006, 66, 112-114.	1.5	102
34	Exon deletions of SPG4 are a frequent cause of hereditary spastic paraplegia. Journal of Medical Genetics, 2007, 44, 281-284.	1.5	100
35	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	5.8	99
36	Mutation analysis of the paraplegin gene (SPG7) in patients with hereditary spastic paraplegia. Neurology, 2006, 66, 654-659.	1.5	98

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37	PRRT2 mutations. Neurology, 2012, 79, 170-174.	1.5	98
38	HCN1mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. Brain, 2018, 141, 3160-3178.	3.7	96
39	<scp>PRRT2</scp> mutations and paroxysmal disorders. European Journal of Neurology, 2013, 20, 872-878.	1.7	95
40	Parental mosaicism can cause recurrent transmission ofSCN1A mutations associated with severe myoclonic epilepsy of infancy. Human Mutation, 2006, 27, 389-389.	1.1	93
41	Mutations of protocadherin 19 in female epilepsy (PCDH19-FE) lead to allopregnanolone deficiency. Human Molecular Genetics, 2015, 24, 5250-5259.	1.4	93
42	Biotin-Responsive Basal Ganglia Disease in Ethnic Europeans With Novel SLC19A3 Mutations. Archives of Neurology, 2010, 67, 126-30.	4.9	93
43	STXBP1-related encephalopathy presenting as infantile spasms and generalized tremor in three patients. Epilepsia, 2011, 52, 1820-1827.	2.6	90
44	Analysis of the chromosome X exome in patients with autism spectrum disorders identified novel candidate genes, including TMLHE. Translational Psychiatry, 2012, 2, e179-e179.	2.4	90
45	Cellular Distribution and Karyophilic Properties of Matrix, Integrase, and Vpr Proteins from the Human and Simian Immunodeficiency Viruses. Experimental Cell Research, 2000, 260, 387-395.	1.2	88
46	Spastin mutations are frequent in sporadic spastic paraparesis and their spectrum is different from that observed in familial cases. Journal of Medical Genetics, 2005, 43, 259-265.	1.5	88
47	Alteration of ornithine metabolism leads to dominant and recessive hereditary spastic paraplegia. Brain, 2015, 138, 2191-2205.	3.7	88
48	Familial cortical myoclonic tremor with epilepsy. Neurology, 2010, 74, 2000-2003.	1.5	83
49	REEP1 mutations in SPG31: Frequency, mutational spectrum, and potential association with mitochondrial morpho-functional dysfunction. Human Mutation, 2011, 32, 1118-1127.	1.1	83
50	Loss of Association of REEP2 with Membranes Leads to Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2014, 94, 268-277.	2.6	83
51	Using medical exome sequencing to identify the causes of neurodevelopmental disorders: Experience of 2 clinical units and 216 patients. Clinical Genetics, 2018, 93, 567-576.	1.0	82
52	RAD51 Haploinsufficiency Causes Congenital Mirror Movements in Humans. American Journal of Human Genetics, 2012, 90, 301-307.	2.6	81
53	A novel DCC mutation and genetic heterogeneity in congenital mirror movements. Neurology, 2011, 76, 260-264.	1.5	80
54	Elicited repetitive daily blindness. Neurology, 2009, 72, 1178-1183.	1.5	79

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55	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. American Journal of Human Genetics, 2019, 104, 1060-1072.	2.6	78
56	GABA_AReceptor Î³2 Subunit Mutations Linked to Human Epileptic Syndromes Differentially Affect Phasic and Tonic Inhibition. Journal of Neuroscience, 2007, 27, 14108-14116.	1.7	76
57	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	2.6	76
58	Autism, language delay and mental retardation in a patient with 7q11 duplication. Journal of Medical Genetics, 2007, 44, 452-458.	1.5	75
59	Phylogenetic Analysis of 49 Newly Derived HIV-1 Group O Strains: High Viral Diversity but No Group M-like Subtype Structure. Virology, 2002, 302, 259-273.	1.1	74
60	Spastic paraplegia due to SPAST mutations is modified by the underlying mutation and sex. Brain, 2018, 141, 3331-3342.	3.7	72
61	Characterization of the Nuclear Import Pathway for HIV-1 Integrase. Journal of Biological Chemistry, 2001, 276, 18102-18107.	1.6	70
62	Two novelCLCN2mutations accelerating chloride channel deactivation are associated with idiopathic generalized epilepsy. Human Mutation, 2009, 30, 397-405.	1.1	70
63	Loss of SYNJ1 dual phosphatase activity leads to early onset refractory seizures and progressive neurological decline. Brain, 2016, 139, 2420-2430.	3.7	70
64	Mutations in the SPG3A gene encoding the GTPase atlastin interfere with vesicle trafficking in the ER/Golgi interface and Golgi morphogenesis. Molecular and Cellular Neurosciences, 2007, 35, 1-13.	1.0	69
65	Mutations in DCC cause isolated agenesis of the corpus callosum with incomplete penetrance. Nature Genetics, 2017, 49, 511-514.	9.4	69
66	Utilization of Microsatellite Polymorphism for Differentiating Herpes Simplex Virus Type 1 Strains. Journal of Clinical Microbiology, 2009, 47, 533-540.	1.8	68
67	Congenital mirror movements: a clue to understanding bimanual motor control. Journal of Neurology, 2011, 258, 1911-1919.	1.8	67
68	Pitfalls in genetic testing: the story of missed SCN1A mutations. Molecular Genetics & Genomic Medicine, 2016, 4, 457-464.	0.6	67
69	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	1.8	66
70	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	2.6	65
71	RAD51 deficiency disrupts the corticospinal lateralization of motor control. Brain, 2013, 136, 3333-3346.	3.7	63
72	Prospective diagnostic analysis of copy number variants using SNP microarrays in individuals with autism spectrum disorders. European Journal of Human Genetics, 2014, 22, 71-78.	1.4	60

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73	Paternal age effects on spermFOXK1andKCNA7methylation and transmission into the next generation. Human Molecular Genetics, 2016, 25, ddw328.	1.4	58
74	Genetic association signal near <scp>NTN</scp>4 in <scp>T</scp>ourette syndrome. Annals of Neurology, 2014, 76, 310-315.	2.8	53
75	Recessive loss-of-function mutations in AP4S1 cause mild fever-sensitive seizures, developmental delay and spastic paraplegia through loss of AP-4 complex assembly. Human Molecular Genetics, 2015, 24, 2218-2227.	1.4	53
76	Congenital mirror movements. Neurology, 2014, 82, 1999-2002.	1.5	52
77	Unusual consequences of status epilepticus in Dravet syndrome. Seizure: the Journal of the British Epilepsy Association, 2010, 19, 190-194.	0.9	50
78	Mutations in the netrin-1 gene cause congenital mirror movements. Journal of Clinical Investigation, 2017, 127, 3923-3936.	3.9	48
79	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	1.1	47
80	Two Novel Epilepsy-Linked Mutations Leading to a Loss of Function of LGI1. Archives of Neurology, 2007, 64, 217.	4.9	44
81	Reversible generalized dystonia and encephalopathy from thiamine transporter 2 deficiency. Movement Disorders, 2012, 27, 1295-1298.	2.2	42
82	DCCmutation update: Congenital mirror movements, isolated agenesis of the corpus callosum, and developmental split brain syndrome. Human Mutation, 2018, 39, 23-39.	1.1	41
83	IntragenicCAMTA1rearrangements cause non-progressive congenital ataxia with or without intellectual disability. Journal of Medical Genetics, 2012, 49, 400-408.	1.5	39
84	Annonacin, a natural lipophilic mitochondrial complex I inhibitor, increases phosphorylation of tau in the brain of FTDP-17 transgenic mice. Experimental Neurology, 2014, 253, 113-125.	2.0	39
85	Genetic variants in components of the NALCNâ€“UNC80â€“UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies). Human Genetics, 2018, 137, 753-768.	1.8	38
86	Mutations in the KIF21B kinesin gene cause neurodevelopmental disorders through imbalanced canonical motor activity. Nature Communications, 2020, 11, 2441.	5.8	37
87	A Novel Locus for Generalized Epilepsy With Febrile Seizures Plus in French Families. Archives of Neurology, 2008, 65, 943-51.	4.9	36
88	SNP arrays in Beckwithâ€“Wiedemann syndrome: An improved diagnostic strategy. European Journal of Medical Genetics, 2013, 56, 546-550.	0.7	34
89	Mental deficiency in three families with SPG4 spastic paraplegia. European Journal of Human Genetics, 2008, 16, 97-104.	1.4	33
90	Uncommon nucleotide excision repair phenotypes revealed by targeted high-throughput sequencing. Orphanet Journal of Rare Diseases, 2016, 11, 26.	1.2	32

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91	A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. American Journal of Human Genetics, 2019, 104, 596-610.	2.6	32
92	Biallelic variants in LARS2 and KARS cause deafness and (ovario)leukodystrophy. Neurology, 2019, 92, e1225-e1237.	1.5	32
93	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32
94	Synaptic processes and immune-related pathways implicated in Tourette syndrome. Translational Psychiatry, 2021, 11, 56.	2.4	31
95	The Immune Signaling Adaptor LAT Contributes to the Neuroanatomical Phenotype of 16p11.2 BP2-BP3 CNVs. American Journal of Human Genetics, 2017, 101, 564-577.	2.6	30
96	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	2.6	30
97	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€“2q11.2. Human Genetics, 2016, 135, 1117-1125.	1.8	29
98	Mechanistic basis of an epistatic interaction reducing age at onset in hereditary spastic paraplegia. Brain, 2018, 141, 1286-1299.	3.7	29
99	An unexpected EEG course in Dravet syndrome. Epilepsy Research, 2008, 81, 90-95.	0.8	28
100	Kjellin Syndrome: Long-term Neuro-ophthalmologic Follow-up and Novel Mutations in the SPG11 Gene. Ophthalmology, 2011, 118, 564-573.	2.5	28
101	DYRK1A mutations in two unrelated patients. European Journal of Medical Genetics, 2015, 58, 168-174.	0.7	28
102	Interaction of Human Immunodeficiency Virus Type 1 Vpr with the HHR23A DNA Repair Protein Does Not Correlate with Multiple Biological Functions of Vpr. Virology, 2001, 282, 176-185.	1.1	27
103	A novel locus for autosomal dominant â€œuncomplicatedâ€•hereditary spastic paraplegia maps to chromosome 8p21.1-q13.3. Human Genetics, 2007, 122, 261-273.	1.8	27
104	ARID1Bmutations are the major genetic cause of corpus callosum anomalies in patients with intellectual disability. Brain, 2016, 139, e64-e64.	3.7	26
105	Copy Number Variations Found in Patients with a Corpus Callosum Abnormality and Intellectual Disability. Journal of Pediatrics, 2017, 185, 160-166.e1.	0.9	25
106	Absence of mutations in the LGI1 receptor ADAM22 gene in autosomal dominant lateral temporal epilepsy. Epilepsy Research, 2007, 76, 41-48.	0.8	24
107	A de novo SPAST mutation leading to somatic mosaicism is associated with a later age at onset in HSP. Neurogenetics, 2007, 8, 231-233.	0.7	24
108	Concordance of genetic variation that increases risk for Tourette Syndrome and that influences its underlying neurocircuitry. Translational Psychiatry, 2019, 9, 120.	2.4	24

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109	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	1.1	24
110	Development and Validation of a Prediction Model for Early Diagnosis of SCN1A -Related Epilepsies. Neurology, 2022, 98, .	1.5	24
111	NIPA1 (SPG6) mutations are a rare cause of autosomal dominant spastic paraplegia in Europe. Neurogenetics, 2007, 8, 155-157.	0.7	23
112	K-complex-induced seizures in autosomal dominant nocturnal frontal lobe epilepsy. Clinical Neurophysiology, 2008, 119, 2201-2204.	0.7	23
113	A mutation update for the PCDH19 gene causing early-onset epilepsy in females with an unusual expression pattern. Human Mutation, 2019, 40, 243-257.	1.1	23
114	Variants in the SK2 channel gene (KCNN2) lead to dominant neurodevelopmental movement disorders. Brain, 2020, 143, 3564-3573.	3.7	23
115	Pleiotrophin mediates the neurotrophic effect of cyclic AMP on dopaminergic neurons: Analysis of suppression-subtracted cDNA libraries and confirmation in vitro. Experimental Neurology, 2005, 194, 243-254.	2.0	22
116	Refinement of the 2p11.1-q12.2 locus responsible for cortical tremor associated with epilepsy and exclusion of candidate genes. Neurogenetics, 2008, 9, 69-71.	0.7	22
117	Familial cortical myoclonic tremor with epilepsy (FCMTE): Clinical characteristics and exclusion of linkages to 8q and 2p in a large French family. Revue Neurologique, 2009, 165, 812-820.	0.6	22
118	A standardized patient-centered characterization of the phenotypic spectrum of PCDH19 girls clustering epilepsy. Translational Psychiatry, 2020, 10, 127.	2.4	22
119	Dalfampridine in hereditary spastic paraplegia: a prospective, open study. Journal of Neurology, 2015, 262, 1285-1288.	1.8	21
120	Early-onset encephalopathy with paroxysmal movement disorders and epileptic seizures without hemiplegic attacks: About three children with novel ATP1A3 mutations. Brain and Development, 2018, 40, 768-774.	0.6	21
121	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	1.1	21
122	A <scp>BBS1 SVA</scp> F retrotransposon insertion is a frequent cause of <scp>Bardetâ€Biedl</scp> syndrome. Clinical Genetics, 2021, 99, 318-324.	1.0	21
123	Lafora progressive myoclonus epilepsy: NHLRC1 mutations affect glycogen metabolism. Journal of Molecular Medicine, 2011, 89, 915-925.	1.7	20
124	A novel homozygous change ofCLCN2(p.His590Pro) is associated with a subclinical form of leukoencephalopathy with ataxia (LKPAT). Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 894-896.	0.9	20
125	Deleterious Variation in BRSK2 Associates with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 701-708.	2.6	19
126	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	1.8	18

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127	Hypomorphic variants of cationic amino acid transporter 3 in males with autism spectrum disorders. Amino Acids, 2015, 47, 2647-2658.	1.2	17
128	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. American Journal of Human Genetics, 2020, 106, 830-845.	2.6	17
129	Microsatellite analysis of HSV-1 isolates: From oropharynx reactivation toward lung infection in patients undergoing mechanical ventilation. Journal of Clinical Virology, 2010, 47, 313-320.	1.6	16
130	Evidence against haploinsuffiency of human ataxin 10 as a cause of spinocerebellar ataxia type 10. Neurogenetics, 2010, 11, 273-274.	0.7	15
131	PAK3 mutations responsible for severe intellectual disability and callosal agenesis inhibit cell migration. Neurobiology of Disease, 2020, 136, 104709.	2.1	14
132	Heterogeneous Pattern of Selective Pressure for PRRT2 in Human Populations, but No Association with Autism Spectrum Disorders. PLoS ONE, 2014, 9, e88600.	1.1	14
133	Spastic paraplegia 5: Locus refinement, candidate gene analysis and clinical description. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 854-861.	1.1	13
134	Association of Rare Genetic Variants in Opioid Receptors with Tourette Syndrome. Tremor and Other Hyperkinetic Movements, 2019, 9, .	1.1	13
135	A rare case of SPG11 mutation with multiple sclerosis. Revue Neurologique, 2016, 172, 389-391.	0.6	12
136	Novel GABRA2 variants in epileptic encephalopathy and intellectual disability with seizures. Brain, 2019, 142, e15-e15.	3.7	12
137	Silencing of the Charcotâ€“Marieâ€“Tooth associated MTMR2 gene decreases proliferation and enhances cell death in primary cultures of Schwann cells. Neurobiology of Disease, 2007, 26, 323-331.	2.1	11
138	Callosal agenesis and congenital mirror movements: outcomes associated with DCC mutations. Developmental Medicine and Child Neurology, 2020, 62, 758-762.	1.1	11
139	Novel <scp>SPG</scp>10 mutation associated with dysautonomia, spinal cord atrophy, and skin biopsy abnormality. European Journal of Neurology, 2013, 20, 398-401.	1.7	10
140	Epilepsy genetics: The ongoing revolution. Revue Neurologique, 2015, 171, 539-557.	0.6	10
141	Targeted versus untargeted omics â€" the CAFSA story. Journal of Inherited Metabolic Disease, 2018, 41, 447-456.	1.7	10
142	Pentameric repeat expansions: cortical myoclonus or cortical tremor?. Brain, 2020, 143, e86-e86.	3.7	9
143	Detection of genomic rearrangements by DHPLC: A prospective study of 90 patients with inherited peripheral neuropathies associated with17p11.2 rearrangements. American Journal of Medical Genetics, Part A, 2005, 136A, 136-139.	0.7	8
144	De novo heterozygous missense and lossâ€ofâ€function variants in CDC42BPB are associated with a neurodevelopmental phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 962-973.	0.7	8

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145	Elevated common variant genetic risk for tourette syndrome in a densely-affected pedigree. Molecular Psychiatry, 2021, 26, 7522-7529.	4.1	8
146	Intragenic deletion of UBE3A gene in 2 sisters with Angelman syndrome detected by MLPA. American Journal of Medical Genetics, Part A, 2011, 155, 3170-3173.	0.7	7
147	Application of rare variant transmission disequilibrium tests to epileptic encephalopathy trio sequence data. European Journal of Human Genetics, 2017, 25, 894-899.	1.4	7
148	Familial form of typical childhood absence epilepsy in a consanguineous context. Epilepsia, 2010, 51, 1889-1893.	2.6	6
149	Unlocking the genetics of paroxysmal kinesigenic dyskinesia. Brain, 2011, 134, 3431-3434.	3.7	6
150	Congenital mirror movements: no mutation in DNAL4 in 17 index cases. Journal of Neurology, 2014, 261, 2030-2031.	1.8	6
151	Dravet Syndrome in Lebanon: First Report on Cases with SCN1A Mutations. Case Reports in Medicine, 2019, 2019, 1-4.	0.3	6
152	Tremor-like subcortical myoclonus in STXBP1 encephalopathy. European Journal of Paediatric Neurology, 2021, 34, 62-66.	0.7	6
153	Genes in infantile epileptic encephalopathies. Epilepsia, 2010, 51, 69-69.	2.6	5
154	Protocadherin 19 mutations in girls with infantile-onset epilepsy. Neurology, 2011, 76, 1193-1194.	1.5	5
155	A high-throughput resequencing microarray for autosomal dominant spastic paraplegia genes. Neurogenetics, 2012, 13, 215-227.	0.7	5
156	CDKL5 and ARX mutations are not responsible for early onset severe myoclonic epilepsy in infancy. Epilepsy Research, 2009, 87, 25-30.	0.8	4
157	SCN1A-related epilepsy with recessive inheritance: Two further families. European Journal of Paediatric Neurology, 2021, 33, 121-124.	0.7	4
158	Familial adult myoclonic epilepsy (FAME): clinical features, molecular characteristics, pathophysiological aspects and diagnostic work-up. Medizinische Genetik, 2022, 33, 311-318.	0.1	4
159	Evidence of mosaicism in SPAST variant carriers in four French families. European Journal of Human Genetics, 2021, 29, 1158-1163.	1.4	3
160	MFSD2A-associated primary microcephaly - Expanding the clinical and mutational spectrum of this ultra-rare disease. European Journal of Medical Genetics, 2021, 64, 104310.	0.7	2
161	GC-rich repeat expansions: associated disorders and mechanisms. Medizinische Genetik, 2022, 33, 325-335.	0.1	2
162	Phenotypical and Myopathological Consequences of Compound Heterozygous Missense and Nonsense Variants in SLC18A3. Cells, 2021, 10, 3481.	1.8	1

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163	Tandem repeat expansions: the good, the bad and the hidden. Medizinische Genetik, 2022, 33, 291-292.	0.1	1
164	Tourette syndrome research highlights from 2020. F1000Research, 0, 11, 45.	0.8	1
165	Corpus callosum agenesis with clinically normal people caused by DCC mutations. Prenatal implication. European Journal of Paediatric Neurology, 2017, 21, e43.	0.7	0
166	Clinical, chromosomal and molecular characterization of a cohort of 273 patients with agenesis of the corpus callosum. European Journal of Paediatric Neurology, 2017, 21, e84.	0.7	0
167	Congenital Mirror Movements Due to RAD51: Cosegregation with a Nonsense Mutation in a Norwegian Pedigree and Review of the Literature. Tremor and Other Hyperkinetic Movements, 2016, 6, 424.	1.1	0
168	Tourette syndrome research highlights from 2020. F1000Research, 2022, 11, 45.	0.8	0