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List of Publications by Year
in descending order

Version: 2024-02-01

80 papers	3,883 citations	109321 35 h-index	128289 60 g-index
85 all docs	85 docs citations	85 times ranked	7061 citing authors

#	Article	IF	Citations
1	Infantile SOD1 deficiency syndrome caused by a homozygous SOD1 variant with absence of enzyme activity. Brain, 2022, 145, 872-878.	7.6	10
2	Delineation of the phenotype of MED17-related disease in Caucasus-Jewish families. European Journal of Paediatric Neurology, 2021, 32, 40-45.	1.6	3
3	Homozygous frameshift variant in NTNG2, encoding a synaptic cell adhesion molecule, in individuals with developmental delay, hypotonia, and autistic features. Neurogenetics, 2019, 20, 209-213.	1.4	7
4	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
5	Pathogenic Variants in NUP214 Cause â€œPluggedâ€•Nuclear Pore Channels and Acute Febrile Encephalopathy. American Journal of Human Genetics, 2019, 105, 48-64.	6.2	29
6	Biallelic variants in AGTPBP1, involved in tubulin deglutamylation, are associated with cerebellar degeneration and motor neuropathy. European Journal of Human Genetics, 2019, 27, 1419-1426.	2.8	25
7	Heterozygous RNF13 Gain-of-Function Variants Are Associated with Congenital Microcephaly, Epileptic Encephalopathy, Blindness, and Failure to Thrive. American Journal of Human Genetics, 2019, 104, 179-185.	6.2	10
8	A homozygous deleterious CDK10 mutation in a patient with agenesis of corpus callosum, retinopathy, and deafness. American Journal of Medical Genetics, Part A, 2018, 176, 92-98.	1.2	21
9	Further delineation of the clinical spectrum of de novo TRIM8 truncating mutations. American Journal of Medical Genetics, Part A, 2018, 176, 2470-2478.	1.2	19
10	Homozygous mutation in MFSD2A, encoding a lysolipid transporter for docosahexanoic acid, is associated with microcephaly and hypomyelination. Neurogenetics, 2018, 19, 227-235.	1.4	45
11	A patient-specific induced pluripotent stem cell model for West syndrome caused by ST3GAL3 deficiency. European Journal of Human Genetics, 2018, 26, 1773-1783.	2.8	15
12	tRNA N6-adenosine threonylcarbamoyltransferase defect due to KAE1/TCS3 (OSGEP) mutation manifest by neurodegeneration and renal tubulopathy. European Journal of Human Genetics, 2017, 25, 545-551.	2.8	67
13	Hypomyelinating leukodystrophy associated with a deleterious mutation in the ATRN gene. Neurogenetics, 2017, 18, 135-139.	1.4	8
14	Mutations in the phosphatidylinositol glycan C (PIGC) gene are associated with epilepsy and intellectual disability. Journal of Medical Genetics, 2017, 54, 196-201.	3.2	44
15	Heterozygous De Novo UBTF Gain-of-Function Variant Is Associated with Neurodegeneration in Childhood. American Journal of Human Genetics, 2017, 101, 267-273.	6.2	41
16	Mutations in TRAPPC12 Manifest in Progressive Childhood Encephalopathy and Golgi Dysfunction. American Journal of Human Genetics, 2017, 101, 291-299.	6.2	37
17	Mitochondrial epileptic encephalopathy, 3â€methylglutaconic aciduria and variable complex V deficiency associated with <scp>TIMM50</scp> mutations. Clinical Genetics, 2017, 91, 690-696.	2.0	28
18	Congenital myasthenic syndrome in Israel: Genetic and clinical characterization. Neuromuscular Disorders, 2017, 27, 136-140.	0.6	30

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19	Complex II Deficiency. , 2016, , 265-272.		0
20	Magnetic resonance imaging spectrum of succinate dehydrogenaseâ€"related infantile leukoencephalopathy. Annals of Neurology, 2016, 79, 379-386.	5.3	34
21	Nemaline body myopathy caused by a novel mutation in troponin T1 (TNNT1). Muscle and Nerve, 2016, 53, 564-569.	2.2	39
22	Deficiency of HTRA2/Omi is associated with infantile neurodegeneration and 3-methylglutaconic aciduria. Journal of Medical Genetics, 2016, 53, 690-696.	3.2	30
23	Altered RNA metabolism due to a homozygousRBM7mutation in a patient with spinal motor neuropathy. Human Molecular Genetics, 2016, 25, ddw149.	2.9	35
24	Therapy with eculizumab for patients with CD59 p.Cys89Tyr mutation. Annals of Neurology, 2016, 80, 708-717.	5.3	41
25	PARP10 deficiency manifests by severe developmental delay and DNA repair defect. Neurogenetics, 2016, 17, 227-232.	1.4	17
26	A mutation in the THG1L gene in a family with cerebellar ataxia and developmental delay. Neurogenetics, 2016, 17, 219-225.	1.4	17
27	Postnatal microcephaly and pain insensitivity due to a de novo heterozygous DNM1L mutation causing impaired mitochondrial fission and function. American Journal of Medical Genetics, Part A, 2016, 170, 1603-1607.	1.2	80
28	Microcephaly-dystonia due to mutated PLEKHG2 with impaired actin polymerization. Neurogenetics, 2016, 17, 25-30.	1.4	8
29	Deficiency of the alkaline ceramidase ACER3 manifests in early childhood by progressive leukodystrophy. Journal of Medical Genetics, 2016, 53, 389-396.	3.2	49
30	Leukoencephalopathy and early death associated with an Ashkenazi-Jewish founder mutation in the Hikeshi gene. Journal of Medical Genetics, 2016, 53, 132-137.	3.2	21
31	P164 â€" 2586: Congenital myasthenic syndrome in Israel: Genetic and clinical characterization. European Journal of Paediatric Neurology, 2015, 19, S140.	1.6	0
32	PP05.11 â€" 3025: A new syndrome with postnatal microcephaly, mental retardation, spastic quadriplegia and pontocerebellar atrophy in Caucasus-Jewish families. European Journal of Paediatric Neurology, 2015, 19, S49.	1.6	0
33	Arginine:glycine amidinotransferase (AGAT) deficiency: Clinical features and long term outcomes in 16 patients diagnosed worldwide. Molecular Genetics and Metabolism, 2015, 116, 252-259.	1.1	55
34	Mitochondrial complex IV deficiency, caused by mutated COX6B1, is associated with encephalomyopathy, hydrocephalus and cardiomyopathy. European Journal of Human Genetics, 2015, 23, 159-164.	2.8	82
35	Truncating Mutation in the Nitric Oxide Synthase 1 Gene Is Associated With Infantile Achalasia. Gastroenterology, 2015, 148, 533-536.e4.	1.3	37
36	Hindbrain malformation and myoclonic seizures associated with a deleterious mutation in the INPP4A gene. Neurogenetics, 2015, 16, 23-26.	1.4	10

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37	A defect in the retromer accessory protein, SNX27, manifests by infantile myoclonic epilepsy and neurodegeneration. Neurogenetics, 2015, 16, 215-221.	1.4	44
38	A human laterality disorder caused by a homozygous deleterious mutation inMMP21. Journal of Medical Genetics, 2015, 52, 840-847.	3.2	46
39	Mutations inSLC1A4, encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination. Journal of Medical Genetics, 2015, 52, 541-547.	3.2	68
40	Congenital myasthenic syndromes in Israel: Genetic and clinical characterization. Neuromuscular Disorders, 2015, 25, S211.	0.6	0
41	Deleterious mutation in FDX1L gene is associated with a novel mitochondrial muscle myopathy. European Journal of Human Genetics, 2014, 22, 902-906.	2.8	65
42	Attention Deficit Hyperactivity Disorders Symptomatology Among Individuals With <scp>D</scp>own Syndrome. Journal of Policy and Practice in Intellectual Disabilities, 2014, 11, 58-61.	2.7	9
43	KIF1Cmutations in two families with hereditary spastic paraparesis and cerebellar dysfunction. Journal of Medical Genetics, 2014, 51, 137-142.	3.2	67
44	Conotruncal malformations and absent thymus due to a deleterious NKX2-6 mutation. Journal of Medical Genetics, 2014, 51, 268-270.	3.2	28
45	Diagnosis by whole exome sequencing of atypical infantile onset Alexander disease masquerading as a mitochondrial disorder. European Journal of Paediatric Neurology, 2014, 18, 495-501.	1.6	7
46	West syndrome caused by ST3Galâ€II deficiency. Epilepsia, 2013, 54, e24-7.	5.1	58
47	P181 â€“ 1833 Atypical infantile onset Alexander disease masquerading as a mitochondrial disorder diagnosed by whole exome sequencing. European Journal of Paediatric Neurology, 2013, 17, S103.	1.6	0
48	Agenesis of corpus callosum and optic nerve hypoplasia due to mutations inSLC25A1encoding the mitochondrial citrate transporter. Journal of Medical Genetics, 2013, 50, 240-245.	3.2	60
49	West syndrome, microcephaly, grey matter heterotopia and hypoplasia of corpus callosum due to a novel ARFGEF2 mutation. Journal of Medical Genetics, 2013, 50, 772-775.	3.2	24
50	Mutations in SLC35A3 cause autism spectrum disorder, epilepsy and arthrogryposis. Journal of Medical Genetics, 2013, 50, 733-739.	3.2	55
51	Early infantile epileptic encephalopathy associated with a high voltage gated calcium channelopathy. Journal of Medical Genetics, 2013, 50, 118-123.	3.2	60
52	Highly fatal fast-channel syndrome caused by AChR É› subunit mutation at the agonist binding site. Neurology, 2012, 79, 449-454.	1.1	27
53	Two novel CCDC88C mutations confirm the role of DAPLE in autosomal recessive congenital hydrocephalus. Journal of Medical Genetics, 2012, 49, 708-712.	3.2	67
54	Intractable epilepsy of infancy due to homozygous mutation in the EFHC1 gene. Epilepsia, 2012, 53, 1436-1440.	5.1	16

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55	Natural disease course and genotypeâ€phenotype correlations in Complex I deficiency caused by nuclear gene defects: what we learned from 130 cases. Journal of Inherited Metabolic Disease, 2012, 35, 737-747.	3.6	112
56	Leukoencephalopathy with accumulated succinate is indicative of SDHAF1 related complex II deficiency. Orphanet Journal of Rare Diseases, 2012, 7, 69.	2.7	44
57	A Deleterious Mutation in DNAJC6 Encoding the Neuronal-Specific Clathrin-Uncoating Co-Chaperone Auxilin, Is Associated with Juvenile Parkinsonism. PLoS ONE, 2012, 7, e36458.	2.5	256
58	Hereditary sensory autonomic neuropathy caused by a mutation in dystonin. Annals of Neurology, 2012, 71, 569-572.	5.3	128
59	2FC1.5 Exome sequencing and disease prediction implicate a mutation in KIF1A as a cause of hereditary spastic paraparesis type 30.. European Journal of Paediatric Neurology, 2011, 15, S19.	1.6	0
60	P16.2 Childhood myasthenia in Israel. European Journal of Paediatric Neurology, 2011, 15, S95-S96.	1.6	0
61	2-hydroxylated sphingomyelin profiles in cells from patients with mutated fatty acid 2-hydroxylase. Lipids in Health and Disease, 2011, 10, 84.	3.0	23
62	A deleterious mutation in the LOXHD1 gene causes autosomal recessive hearing loss in Ashkenazi Jews. American Journal of Medical Genetics, Part A, 2011, 155, 1170-1172.	1.2	29
63	Exome sequencing and disease-network analysis of a single family implicate a mutation in KIF1A in hereditary spastic paraparesis. Genome Research, 2011, 21, 658-664.	5.5	172
64	A common pattern of brain MRI imaging in mitochondrial diseases with complex I deficiency. Journal of Medical Genetics, 2011, 48, 16-23.	3.2	89
65	827 Slc25A19 Mutation is a Novel Cause of Neuropathy and Bilateral Striatal Necrosis. Pediatric Research, 2010, 68, 415-415.	2.3	0
66	Joubert Syndrome 2 (JBTS2) in Ashkenazi Jews Is Associated with a TMEM216 Mutation. American Journal of Human Genetics, 2010, 86, 93-97.	6.2	89
67	Joubert Syndrome 2 (JBTS2) in Ashkenazi Jews Is Associated with a TMEM216 Mutation. American Journal of Human Genetics, 2010, 86, 294.	6.2	0
68	Infantile Cerebral and Cerebellar Atrophy Is Associated with a Mutation in the MED17 Subunit of the Transcription Preinitiation Mediator Complex. American Journal of Human Genetics, 2010, 87, 667-670.	6.2	58
69	Defective FA2H leads to a novel form of neurodegeneration with brain iron accumulation (NBIA). Annals of Neurology, 2010, 68, 611-618.	5.3	202
70	l-arginine:glycine amidinotransferase (AGAT) deficiency: Clinical presentation and response to treatment in two patients with a novel mutation. Molecular Genetics and Metabolism, 2010, 101, 228-232.	1.1	44
71	SLC25A19 mutation as a cause of neuropathy and bilateral striatal necrosis. Annals of Neurology, 2009, 66, 419-424.	5.3	74
72	Mitochondrial complex I deficiency caused by a deleterious NDUFA11 mutation. Annals of Neurology, 2008, 63, 405-408.	5.3	103

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73	Mutations in the Fatty Acid 2-Hydroxylase Gene Are Associated with Leukodystrophy with Spastic Paraparesis and Dystonia. American Journal of Human Genetics, 2008, 83, 643-648.	6.2	193
74	C6ORF66 Is an Assembly Factor of Mitochondrial Complex I. American Journal of Human Genetics, 2008, 82, 32-38.	6.2	155
75	The unique neuroradiology of complex I deficiency due to NDUFA12L defect. Molecular Genetics and Metabolism, 2008, 94, 78-82.	1.1	46
76	Deleterious Mutation in the Mitochondrial Arginylâ€“Transfer RNA Synthetase Gene Is Associated with Pontocerebellar Hypoplasia. American Journal of Human Genetics, 2007, 81, 857-862.	6.2	306
77	NMO06 Treatment of congenital myasthenia with ephedrine: a case report. European Journal of Paediatric Neurology, 2007, 11, 38.	1.6	1
78	NMP02 A hereditary disorder of early onset motor neuron disease and epilepsy. European Journal of Paediatric Neurology, 2007, 11, 72.	1.6	0
79	SUBLINGUAL TIZANIDINE FOR TREATMENT OF SPASTIC CP IN CHILDREN: A PILOT STUDY. Neuropediatrics, 2006, 37, .	0.6	0
80	A case of Tourette syndrome presenting with oral self-injurious behaviour. International Journal of Paediatric Dentistry, 2005, 15, 370-374.	1.8	20