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List of Publications by Year
in descending order

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67 papers	3,847 citations	159585 30 h-index	138484 58 g-index
68 all docs	68 docs citations	68 times ranked	8341 citing authors

#	Article	IF	Citations
1	Overlapping cortical malformations in patients with pathogenic variants inGRIN1andGRIN2B. Journal of Medical Genetics, 2023, 60, 183-192.	3.2	1
2	De novo TRPV4 Leu619Pro variant causes a new channelopathy characterised by giant cell lesions of the jaws and skull, skeletal abnormalities and polyneuropathy. Journal of Medical Genetics, 2022, 59, 305-312.	3.2	6
3	A standard of care for individuals with <scp>PIK3CA</scp>â€related disorders: An international expert consensus statement. Clinical Genetics, 2022, 101, 32-47.	2.0	21
4	Defining the phenotypical spectrum associated with variants in TUBB2A. Journal of Medical Genetics, 2021, 58, 33-40.	3.2	11
5	The potential diagnostic yield of whole exome sequencing in pregnancies complicated by fetal ultrasound anomalies. Acta Obstetricia Et Gynecologica Scandinavica, 2021, 100, 1106-1115.	2.8	21
6	ATP1A2- and ATP1A3-associated early profound epileptic encephalopathy and polymicrogyria. Brain, 2021, 144, 1435-1450.	7.6	35
7	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	6.2	17
8	Cystoid maculopathy is a frequent feature of Cohen syndrome-associated retinopathy. Scientific Reports, 2021, 11, 16412.	3.3	7
9	Human TBK1 deficiency leads to autoinflammation driven by TNF-induced cell death. Cell, 2021, 184, 4447-4463.e20.	28.9	64
10	A novel family illustrating the mild phenotypic spectrum of TUBB2B variants. European Journal of Paediatric Neurology, 2021, 35, 35-39.	1.6	2
11	Multidisciplinary interaction and MCD gene discovery. The perspective of the clinical geneticist. European Journal of Paediatric Neurology, 2021, 35, 27-34.	1.6	3
12	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
13	Definitions and classification of malformations of cortical development: practical guidelines. Brain, 2020, 143, 2874-2894.	7.6	145
14	International consensus recommendations on the diagnostic work-up for malformations of cortical development. Nature Reviews Neurology, 2020, 16, 618-635.	10.1	53
15	Human <scp>RAD50</scp> deficiency: Confirmation of a distinctive phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 1378-1386.	1.2	21
16	A recurrent de novo missense pathogenic variant in SMARCB1 causes severe intellectual disability and choroid plexus hyperplasia with resultant hydrocephalus. Genetics in Medicine, 2019, 21, 572-579.	2.4	24
17	Mutations in the Heterotopia Gene Eml1/EML1 Severely Disrupt the Formation of Primary Cilia. Cell Reports, 2019, 28, 1596-1611.e10.	6.4	28
18	Loss of USP18 in microglia induces white matter pathology. Acta Neuropathologica Communications, 2019, 7, 106.	5.2	15

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19	EML1â€associated brain overgrowth syndrome with ribbonâ€like heterotopia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 627-637.	1.6	17
20	Late-onset phenotype associated with a homozygous GJC2 missense mutation in a Turkish family. Parkinsonism and Related Disorders, 2019, 66, 228-231.	2.2	5
21	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48
22	Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. American Journal of Human Genetics, 2019, 105, 844-853.	6.2	17
23	De novo loss-of-function KCNMA1 variants are associated with a new multiple malformation syndrome and a broad spectrum of developmental and neurological phenotypes. Human Molecular Genetics, 2019, 28, 2937-2951.	2.9	76
24	Heterogeneous clinical phenotypes and cerebral malformations reflected by rotatin cellular dynamics. Brain, 2019, 142, 867-884.	7.6	22
25	Cysteinyl-tRNA Synthetase Mutations Cause a Multi-System, Recessive Disease That Includes Microcephaly, Developmental Delay, and Brittle Hair and Nails. American Journal of Human Genetics, 2019, 104, 520-529.	6.2	31
26	TMX2 Is a Crucial Regulator of Cellular Redox State, and Its Dysfunction Causes Severe Brain Developmental Abnormalities. American Journal of Human Genetics, 2019, 105, 1126-1147.	6.2	25
27	Linking the heart and the brain: Neurodevelopmental disorders in patients with catecholaminergic polymorphic ventricular tachycardia. Heart Rhythm, 2019, 16, 220-228.	0.7	29
28	SYNGAP1 encephalopathy. Neurology, 2019, 92, e96-e107.	1.1	131
29	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
30	Mixoploidy combined with aneuploidy in a 13 yearâ€old patient with severe multiple congenital abnormalities and intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 492-495.	1.2	3
31	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.2	59
32	MACF1 Mutations Encoding Highly Conserved Zinc-Binding Residues of the GAR Domain Cause Defects in Neuronal Migration and Axon Guidance. American Journal of Human Genetics, 2018, 103, 1009-1021.	6.2	57
33	Mutated zinc finger protein of the cerebellum 1 leads to microcephaly, cortical malformation, callosal agenesis, cerebellar dysplasia, tethered cord and scoliosis. European Journal of Medical Genetics, 2018, 61, 783-789.	1.3	10
34	Altered distribution of ATG9A and accumulation of axonal aggregates in neurons from a mouse model of AP-4 deficiency syndrome. PLoS Genetics, 2018, 14, e1007363.	3.5	85
35	SYT1-associated neurodevelopmental disorder: a case series. Brain, 2018, 141, 2576-2591.	7.6	98
36	Sodium Channel SCN3A (NaV1.3) Regulation of Human Cerebral Cortical Folding and Oral Motor Development. Neuron, 2018, 99, 905-913.e7.	8.1	109

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37	Neuro-MIG: A European network on brain malformations. European Journal of Medical Genetics, 2018, 61, 741-743.	1.3	6
38	Progressive leukoencephalopathy impairs neurobehavioral development in sialin-deficient mice. Experimental Neurology, 2017, 291, 106-119.	4.1	10
39	Molybdenum cofactor deficiency: Identification of a patient with homozygote mutation in the MOCS3 gene. American Journal of Medical Genetics, Part A, 2017, 173, 1601-1606.	1.2	33
40	XRCC1 mutation is associated with PARP1 hyperactivation and cerebellar ataxia. Nature, 2017, 541, 87-91.	27.8	209
41	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. American Journal of Human Genetics, 2017, 101, 552-563.	6.2	45
42	Altered synaptobrevin-II trafficking in neurons expressing a synaptophysin mutation associated with a severe neurodevelopmental disorder. Neurobiology of Disease, 2017, 108, 298-306.	4.4	25
43	Advanced genomic testing may aid in counseling of isolated agenesis of the corpus callosum on prenatal ultrasound. Prenatal Diagnosis, 2017, 37, 1191-1197.	2.3	22
44	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. American Journal of Human Genetics, 2017, 101, 466-477.	6.2	119
45	Ehlers Danlos syndrome, kyphoscoliotic type due to Lysyl Hydroxylase 1 deficiency in two children without congenital or early onset kyphoscoliosis. European Journal of Medical Genetics, 2017, 60, 536-540.	1.3	9
46	Loss-of-Function and Gain-of-Function Mutations in KCNQ5 Cause Intellectual Disability or Epileptic Encephalopathy. American Journal of Human Genetics, 2017, 101, 65-74.	6.2	99
47	Expanding the Phenotype Associated with NAA10â€Related Nâ€Terminal Acetylation Deficiency. Human Mutation, 2016, 37, 755-764.	2.5	70
48	Human USP18 deficiency underlies type 1 interferonopathy leading to severe pseudo-TORCH syndrome. Journal of Experimental Medicine, 2016, 213, 1163-1174.	8.5	224
49	CSTB null mutation associated with microcephaly, early developmental delay, and severe dyskinesia. Neurology, 2016, 86, 877-878.	1.1	25
50	The expanding phenotypic spectrum of ARFGEF2 gene mutation: Cardiomyopathy and movement disorder. Brain and Development, 2016, 38, 124-127.	1.1	22
51	WDR73Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. Human Mutation, 2015, 36, 1021-1028.	2.5	42
52	The expanding phenotype of COL4A1 and COL4A2 mutations: clinical data on 13 newly identified families and a review of the literature. Genetics in Medicine, 2015, 17, 843-853.	2.4	204
53	<scp>USP</scp> 18 lack in microglia causes destructive interferonopathy of the mouse brain. EMBO Journal, 2015, 34, 1612-1629.	7.8	178
54	Germline activating AKT3 mutation associated with megalencephaly, polymicrogyria, epilepsy and hypoglycemia. Molecular Genetics and Metabolism, 2015, 114, 467-473.	1.1	42

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55	Genotypeâ€“phenotype correlation of contiguous gene deletions of <scp>SLC6A8</scp>, <scp>BCAP31</scp> and <scp>ABCD1</scp>. Clinical Genetics, 2015, 87, 141-147.	2.0	25
56	Severe presentation of WDR62 mutation: Is there a role for modifying genetic factors?. American Journal of Medical Genetics, Part A, 2014, 164, 2161-2171.	1.2	30
57	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. Nature Genetics, 2014, 46, 510-515.	21.4	118
58	Progressive cerebellar atrophy and polyneuropathy: expanding the spectrum of PNKP mutations. Neurogenetics, 2013, 14, 43-51.	1.4	66
59	ACTA2 mutation with childhood cardiovascular, autonomic and brain anomalies and severe outcome. American Journal of Medical Genetics, Part A, 2013, 161, 1376-1380.	1.2	36
60	COL4A2 mutation associated with familial porencephaly and small-vessel disease. European Journal of Human Genetics, 2012, 20, 844-851.	2.8	84
61	Asymmetric polymicrogyria and periventricular nodular heterotopia due to mutation in ARX. American Journal of Medical Genetics, Part A, 2012, 158A, 1472-1476.	1.2	7
62	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. Nature Genetics, 2012, 44, 934-940.	21.4	621
63	Combined cardiological and neurological abnormalities due to filamin A gene mutation. Clinical Research in Cardiology, 2011, 100, 45-50.	3.3	43
64	Periventricular nodular heterotopia and distal limb deficiency: A recurrent association. American Journal of Medical Genetics, Part A, 2010, 152A, 954-959.	1.2	2
65	Unbalanced der(5)t(5;20) translocation associated with megalencephaly, perisylvian polymicrogyria, polydactyly and hydrocephalus. American Journal of Medical Genetics, Part A, 2010, 152A, 1488-1497.	1.2	6
66	[P2.75]: Malformations of cortical development: Genomic analysis by high density microarrays in a large patient cohort. International Journal of Developmental Neuroscience, 2010, 28, 712-713.	1.6	0
67	Re: Polymicrogyria versus pachygyria in 22q11 microdeletion. American Journal of Medical Genetics, Part A, 2005, 136A, 419-419.	1.2	1