Knut Brockmann

List of Publications by Year
in descending order

54	4,127	218677	175258
papers	citations	26 h-index	52 g-index

54	54	54	5847
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Loss-of-function variants in DNM1 cause a specific form of developmental and epileptic encephalopathy only in biallelic state. Journal of Medical Genetics, 2022, 59, 549-553.	3.2	9
2	Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. Genetics in Medicine, 2021, 23, 341-351.	2.4	16
3	A novel homozygous nonsense mutation of VPS13B associated with previously unreported features of Cohen syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 570-575.	1.2	10
4	Comparative analysis of alternating hemiplegia of childhood and rapid-onset dystonia-parkinsonism ATP1A3 mutations reveals functional deficits, which do not correlate with disease severity. Neurobiology of Disease, 2020, 143, 105012.	4.4	8
5	Evidence of pathogenicity for the leaky splice variant c. 1066â€6T >Gâ€‰in ATM. American Journal of Medical Genetics, Part A, 2020, 182, 2971-2975.	1.2	5
6	The Phenotypic Spectrum of PRRT2-Associated Paroxysmal Neurologic Disorders in Childhood. Biomedicines, 2020, 8, 456.	3.2	23
7	Homozygosity for the c.428delG variant in KIAA0586 in a healthy individual: implications for molecular testing in patients with Joubert syndrome. Journal of Medical Genetics, 2019, 56, 261-264.	3.2	15
8	A Novel Gain-of-Function Nav1.9 Mutation in a Child With Episodic Pain. Frontiers in Neuroscience, 2019, 13, 918.	2.8	18
9	Structural brain anomalies in patients with <scp>FOXG</scp>1 syndrome and in Foxg1+/â^' mice. Annals of Clinical and Translational Neurology, 2019, 6, 655-668.	3.7	19
10	Erosive tooth wear and caries experience in children and adolescents with obesity. Journal of Dentistry, 2019, 83, 77-86.	4.1	26
11	FOXG1 syndrome: genotypeâ€"phenotype association in 83 patients with FOXG1 variants. Genetics in Medicine, 2018, 20, 98-108.	2.4	77
12	Nosological delineation of congenital ocular motor apraxia type Cogan: an observational study. Orphanet Journal of Rare Diseases, 2016, 11, 104.	2.7	21
13	Photosensitive form of trichothiodystrophy associated with a novel mutation in the <scp>XPD</scp> gene. Photodermatology Photoimmunology and Photomedicine, 2016, 32, 110-112.	1.5	5
14	Magnetic resonance imaging spectrum of succinate dehydrogenaseâ€"related infantile leukoencephalopathy. Annals of Neurology, 2016, 79, 379-386.	5.3	34
15	Eight further individuals with intellectual disability and epilepsy carrying bi-allelicCNTNAP2aberrations allow delineation of the mutational and phenotypic spectrum. Journal of Medical Genetics, 2016, 53, 820-827.	3.2	45
16	Clinical, neuroradiological and molecular characterization of cerebellar dysplasia with cysts (Porettiâ€"Boltshauser syndrome). European Journal of Human Genetics, 2016, 24, 1262-1267.	2.8	43
17	Obtaining a genetic diagnosis in a child with disability: impact on parental quality of life. Clinical Genetics, 2016, 89, 258-266.	2.0	50
18	Disturbed Neuronal ER-Golgi Sorting of Unassembled Glycine Receptors Suggests Altered Subcellular Processing Is a Cause of Human Hyperekplexia. Journal of Neuroscience, 2015, 35, 422-437.	3.6	26

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19	Genetics of Paroxysmal Dyskinesia. , 2015, , 191-211.		1
20	Comprehensive genotyping and clinical characterisation reveal 27 novel NKX2-1 mutations and expand the phenotypic spectrum. Journal of Medical Genetics, 2014, 51, 375-387.	3.2	77
21	The LYR Factors SDHAF1 and SDHAF3 Mediate Maturation of the Iron-Sulfur Subunit of Succinate Dehydrogenase. Cell Metabolism, 2014, 20, 253-266.	16.2	96
22	Intragenic duplication of EHMT1 gene results in Kleefstra syndrome. Molecular Cytogenetics, 2014, 7, 74.	0.9	11
23	A novel ATP1A3 mutation with unique clinical presentation. Journal of the Neurological Sciences, 2014, 341, 133-135.	0.6	24
24	Phenotypic overlap of alternating hemiplegia of childhood and CAPOS syndrome. Neurology, 2014, 83, 861-863.	1.1	42
25	The expanding clinical and genetic spectrum of ATP1A3-related disorders. Neurology, 2014, 82, 945-955.	1.1	98
26	Episodic Movement Disorders: From Phenotype to Genotype and Back. Current Neurology and Neuroscience Reports, 2013, 13, 379.	4.2	9
27	Leukoencephalopathy with accumulated succinate is indicative of SDHAF1 related complex II deficiency. Orphanet Journal of Rare Diseases, 2012, 7, 69.	2.7	44
28	Heterozygous de-novo mutations in ATP1A3 in patients with alternating hemiplegia of childhood: a whole-exome sequencing gene-identification study. Lancet Neurology, The, 2012, 11, 764-773.	10.2	223
29	PRRT2 Mutations are the major cause of benign familial infantile seizures. Human Mutation, 2012, 33, 1439-1443.	2.5	93
30	A Mosaic Activating Mutation inAKT1Associated with the Proteus Syndrome. New England Journal of Medicine, 2011, 365, 611-619.	27.0	800
31	Towards a more palatable treatment for Glut1 deficiency syndrome. Developmental Medicine and Child Neurology, 2011, 53, 580-581.	2.1	4
32	Leukodystrophies and other genetic metabolic leukoencephalopathies in children and adults. Brain and Development, 2010, 32, 82-89.	1.1	26
33	Unilateral Dilation of Virchow-Robin Spaces in Early Childhood. Neuropediatrics, 2009, 40, 234-238.	0.6	9
34	Overlap of Moebius and oromandibular limb hypogenesis syndrome with gastroschisis and pulmonary hypoplasia. American Journal of Medical Genetics, Part A, 2009, 149A, 2832-2837.	1.2	13
35	The expanding phenotype of GLUT1-deficiency syndrome. Brain and Development, 2009, 31, 545-552.	1.1	216
36	Cerebral involvement in axonal Charcot-Marie-Tooth neuropathy caused by mitofusin2 mutations. Journal of Neurology, 2008, 255, 1049-58.	3.6	66

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37	Monozygotic twins discordant for Proteus syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 2122-2125.	1.2	27
38	GLUT1 mutations are a cause of paroxysmal exertion-induced dyskinesias and induce hemolytic anemia by a cation leak. Journal of Clinical Investigation, 2008, 118, 2157-2168.	8.2	321
39	Sensitivity and specificity of qualitative muscle ultrasound in assessment of suspected neuromuscular disease in childhood. Neuromuscular Disorders, 2007, 17, 517-523.	0.6	64
40	Quantitative proton MRS of cerebral metabolites in laminin Î±2 chain deficiency. Brain and Development, 2007, 29, 357-364.	1.1	9
41	Cerebral metabolic and structural alterations in hereditary spastic paraplegia with thin corpus callosum assessed by MRS and DTI. Neuroradiology, 2006, 48, 893-898.	2.2	35
42	Moyamoya Syndrome Associated With Hemolytic Anemia Due to Hb Alesha. Journal of Pediatric Hematology/Oncology, 2005, 27, 436-440.	0.6	24
43	Visually Self-induced Seizures Sensitive to Round Objects. Epilepsia, 2005, 46, 786-789.	5.1	3
44	Xâ€“linked paroxysmal dyskinesia and severe global retardation caused by defective MCT8 gene. Journal of Neurology, 2005, 252, 663-666.	3.6	89
45	Complicated Hereditary Spastic Paraplegia with Thin Corpus Callosum (HSPâ€TCC) and Childhood Onset. Neuropediatrics, 2005, 36, 274-278.	0.6	21
46	Tarsal tunnel syndrome in a 7-year-old boy. European Journal of Pediatrics, 2004, 163, 46-47.	2.7	3
47	A Novel Syndrome Combining Thyroid and Neurological Abnormalities Is Associated with Mutations in a Monocarboxylate Transporter Gene. American Journal of Human Genetics, 2004, 74, 168-175.	6.2	613
48	Cerebral proton magnetic resonance spectroscopy of a patient with giant axonal neuropathy. Brain and Development, 2003, 25, 45-50.	1.1	15
49	A Novel GFAP Mutation and Disseminated White Matter Lesions: Adult Alexander Disease?. European Neurology, 2003, 50, 100-105.	1.4	25
50	Succinate in dystrophic white matter: A proton magnetic resonance spectroscopy finding characteristic for complex II deficiency. Annals of Neurology, 2002, 52, 38-46.	5.3	88
51	EEG Features of Glutâ€1 Deficiency Syndrome. Epilepsia, 2002, 43, 941-945.	5.1	52
52	Autosomal dominant Glutâ€1 deficiency syndrome and familial epilepsy. Annals of Neurology, 2001, 50, 476-485.	5.3	153
53	Regional Age Dependence of Human Brain Metabolites from Infancy to Adulthood as Detected by Quantitative Localized Proton MRS. Pediatric Research, 1999, 46, 474-474.	2.3	283
54	Magnetic resonance spectroscopy in pediatric white matter disease. , 0, , 806-822.		0