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	A Mosaic Activating Mutation inAKT1Associated with the Proteus Syndrome. New England Journal of Medicine, 2011, 365, 611-619.	27.0	800
2	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
3	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
4	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
5	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
6	The expanding phenotype of GLUT1-deficiency syndrome. Brain and Development, 2009, 31, 545-552.	1.1	216
7	Autosomal dominant Glutâ€1 deficiency syndrome and familial epilepsy. Annals of Neurology, 2001, 50, 476-485.	5.3	153
8	The expanding clinical and genetic spectrum of ATP1A3-related disorders. Neurology, 2014, 82, 945-955.	1.1	98
9	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
10	PRRT2 Mutations are the major cause of benign familial infantile seizures. Human Mutation, 2012, 33, 1439-1443.	2.5	93
11	Xâ€“linked paroxysmal dyskinesia and severe global retardation caused by defective MCT8 gene. Journal of Neurology, 2005, 252, 663-666.	3.6	89
12	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
13	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
14	FOXG1 syndrome: genotypeâ€“phenotype association in 83 patients with FOXG1 variants. Genetics in Medicine, 2018, 20, 98-108.	2.4	77
15	Cerebral involvement in axonal Charcot-Marie-Tooth neuropathy caused by mitofusin2 mutations. Journal of Neurology, 2008, 255, 1049-58.	3.6	66
16	Sensitivity and specificity of qualitative muscle ultrasound in assessment of suspected neuromuscular disease in childhood. Neuromuscular Disorders, 2007, 17, 517-523.	0.6	64
17	EEG Features of Glutâ€1 Deficiency Syndrome. Epilepsia, 2002, 43, 941-945.	5.1	52
18	Obtaining a genetic diagnosis in a child with disability: impact on parental quality of life. Clinical Genetics, 2016, 89, 258-266.	2.0	50

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19	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
20	Leukoencephalopathy with accumulated succinate is indicative of SDHAF1 related complex II deficiency. Orphanet Journal of Rare Diseases, 2012, 7, 69.	2.7	44
21	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
22	Phenotypic overlap of alternating hemiplegia of childhood and CAPOS syndrome. Neurology, 2014, 83, 861-863.	1.1	42
23	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
24	Magnetic resonance imaging spectrum of succinate dehydrogenaseâ€“related infantile leukoencephalopathy. Annals of Neurology, 2016, 79, 379-386.	5.3	34
25	Monozygotic twins discordant for Proteus syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 2122-2125.	1.2	27
26	Leukodystrophies and other genetic metabolic leukoencephalopathies in children and adults. Brain and Development, 2010, 32, 82-89.	1.1	26
27	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
28	Erosive tooth wear and caries experience in children and adolescents with obesity. Journal of Dentistry, 2019, 83, 77-86.	4.1	26
29	A Novel GFAP Mutation and Disseminated White Matter Lesions: Adult Alexander Disease?. European Neurology, 2003, 50, 100-105.	1.4	25
30	Moyamoya Syndrome Associated With Hemolytic Anemia Due to Hb Alesha. Journal of Pediatric Hematology/Oncology, 2005, 27, 436-440.	0.6	24
31	A novel ATP1A3 mutation with unique clinical presentation. Journal of the Neurological Sciences, 2014, 341, 133-135.	0.6	24
32	The Phenotypic Spectrum of PRRT2-Associated Paroxysmal Neurologic Disorders in Childhood. Biomedicines, 2020, 8, 456.	3.2	23
33	Complicated Hereditary Spastic Paraplegia with Thin Corpus Callosum (HSPâ€TCC) and Childhood Onset. Neuropediatrics, 2005, 36, 274-278.	0.6	21
34	Nosological delineation of congenital ocular motor apraxia type Cogan: an observational study. Orphanet Journal of Rare Diseases, 2016, 11, 104.	2.7	21
35	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
36	A Novel Gain-of-Function Nav1.9 Mutation in a Child With Episodic Pain. Frontiers in Neuroscience, 2019, 13, 918.	2.8	18

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37	Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. Genetics in Medicine, 2021, 23, 341-351.	2.4	16
38	Cerebral proton magnetic resonance spectroscopy of a patient with giant axonal neuropathy. Brain and Development, 2003, 25, 45-50.	1.1	15
39	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
40	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
41	Intragenic duplication of EHMT1 gene results in Kleefstra syndrome. Molecular Cytogenetics, 2014, 7, 74.	0.9	11
42	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
43	Quantitative proton MRS of cerebral metabolites in laminin Î±2 chain deficiency. Brain and Development, 2007, 29, 357-364.	1.1	9
44	Unilateral Dilation of Virchow-Robin Spaces in Early Childhood. Neuropediatrics, 2009, 40, 234-238.	0.6	9
45	Episodic Movement Disorders: From Phenotype to Genotype and Back. Current Neurology and Neuroscience Reports, 2013, 13, 379.	4.2	9
46	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
47	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
48	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
49	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
50	Towards a more palatable treatment for Glut1 deficiency syndrome. Developmental Medicine and Child Neurology, 2011, 53, 580-581.	2.1	4
51	Tarsal tunnel syndrome in a 7-year-old boy. European Journal of Pediatrics, 2004, 163, 46-47.	2.7	3
52	Visually Self-induced Seizures Sensitive to Round Objects. Epilepsia, 2005, 46, 786-789.	5.1	3
53	Genetics of Paroxysmal Dyskinesia. , 2015, , 191-211.		1
54	Magnetic resonance spectroscopy in pediatric white matter disease. , 0, , 806-822.		0