Malin Kvarnung

List of Publications by Year
in descending order

23 papers	1,698 citations	471509 17 h-index	610901 24 g-index
25 all docs	25 docs citations	25 times ranked	4157 citing authors

#	Article	IF	Citations
1	Increasing involvement of CAPN1 variants in spastic ataxias and phenotype-genotype correlations. Neurogenetics, 2021, 22, 71-79.	1.4	11
2	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
3	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40.	8.2	116
4	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
5	TUBB3 Arg262His causes a recognizable syndrome including CFEOM3, facial palsy, joint contractures, and early-onset peripheral neuropathy. Human Genetics, 2021, 140, 1709-1731.	3.8	13
6	Pathogenic variants in TNRC6B cause a genetic disorder characterised by developmental delay/intellectual disability and a spectrum of neurobehavioural phenotypes including autism and ADHD. Journal of Medical Genetics, 2020, 57, 717-724.	3.2	14
7	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
8	Ataxia in Patients With Bi-Allelic NFASC Mutations and Absence of Full-Length NF186. Frontiers in Genetics, 2019, 10, 896.	2.3	7
9	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. Genome Medicine, 2019, 11, 68.	8.2	88
10	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
11	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
12	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
13	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	2.8	32
14	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87
15	Genomic screening in rare disorders: New mutations and phenotypes, highlighting ALG14 as a novel cause of severe intellectual disability. Clinical Genetics, 2018, 94, 528-537.	2.0	29
16	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
17	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
18	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. American Journal of Human Genetics, 2017, 100, 650-658.	6.2	56

#	Article	IF	Citations
19	Intellectual Disability & Rare Disorders: A Diagnostic Challenge. Advances in Experimental Medicine and Biology, 2017, 1031, 39-54.	1.6	27
20	Choline transporter mutations in severe congenital myasthenic syndrome disrupt transporter localization. Brain, 2017, 140, 2838-2850.	7.6	24
21	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
22	A novel intellectual disability syndrome caused by GPI anchor deficiency due to homozygous mutations inPIGT. Journal of Medical Genetics, 2013, 50, 521-528.	3.2	108
23	Inherited mosaicism for the supernumerary marker chromosome in cat eye syndrome: Interâ€•and intraâ€•individual variation and correlation to the phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 1111-1117.	1.2	9