

Malin Kvarnung

List of Publications by Year in descending order

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Version: 2024-02-01

23
papers

1,698
citations

471061

17
h-index

610482

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. <i>Neurogenetics</i> , 2021, 22, 71-79.	0.7	11
2	DLG4-related synaptopathy: a new rare brain disorder. <i>Genetics in Medicine</i> , 2021, 23, 888-899.	1.1	16
3	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , 2021, 13, 40.	3.6	116
4	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 63.	3.6	50
5	TUBB3 Arg262His causes a recognizable syndrome including CFEOM3, facial palsy, joint contractures, and early-onset peripheral neuropathy. <i>Human Genetics</i> , 2021, 140, 1709-1731.	1.8	13
6	Pathogenic variants in <i>TNRC6B</i> cause a genetic disorder characterised by developmental delay/intellectual disability and a spectrum of neurobehavioural phenotypes including autism and ADHD. <i>Journal of Medical Genetics</i> , 2020, 57, 717-724.	1.5	14
7	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. <i>Nature Communications</i> , 2020, 11, 4932.	5.8	105
8	Ataxia in Patients With Bi-Allelic NFASC Mutations and Absence of Full-Length NF186. <i>Frontiers in Genetics</i> , 2019, 10, 896.	1.1	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. <i>Genome Medicine</i> , 2019, 11, 68.	3.6	88
10	Disruptive variants of <i>CSDE1</i> associate with autism and interfere with neuronal development and synaptic transmission. <i>Science Advances</i> , 2019, 5, eaax2166.	4.7	35
11	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	2.6	30
12	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. <i>Biological Psychiatry</i> , 2019, 85, 287-297.	0.7	108
13	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. <i>European Journal of Human Genetics</i> , 2018, 26, 54-63.	1.4	32
14	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018, 103, 666-678.	2.6	87
15	Genomic screening in rare disorders: New mutations and phenotypes, highlighting <i>ALG14</i> as a novel cause of severe intellectual disability. <i>Clinical Genetics</i> , 2018, 94, 528-537.	1.0	29
16	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. <i>Nature Genetics</i> , 2017, 49, 515-526.	9.4	443
17	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. <i>Nature Neuroscience</i> , 2017, 20, 1043-1051.	7.1	152
18	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 650-658.	2.6	56

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19	Intellectual Disability & Rare Disorders: A Diagnostic Challenge. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 39-54.	0.8	27
20	Choline transporter mutations in severe congenital myasthenic syndrome disrupt transporter localization. <i>Brain</i> , 2017, 140, 2838-2850.	3.7	24
21	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2016, 98, 541-552.	2.6	132
22	A novel intellectual disability syndrome caused by GPI anchor deficiency due to homozygous mutations in <i>PIGT</i> . <i>Journal of Medical Genetics</i> , 2013, 50, 521-528.	1.5	108
23	Inherited mosaicism for the supernumerary marker chromosome in cat eye syndrome: Inter- and intra-individual variation and correlation to the phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1111-1117.	0.7	9