Marisa A Vineyard

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5004255/publications.pdf

Version: 2024-02-01

12	495	840776 11	1199594 12
papers	citations	h-index	g-index

12	12	12	1671
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
2	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. Genetics in Medicine, 2017, 19, 1040-1048.	2.4	85
3	A Novel Mutation in Isoform 3 of the Plasma Membrane Ca2+ Pump Impairs Cellular Ca2+ Homeostasis in a Patient with Cerebellar Ataxia and Laminin Subunit 1Î± Mutations. Journal of Biological Chemistry, 2015, 290, 16132-16141.	3.4	41
4	Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders. Genome Medicine, 2017, 9, 73.	8.2	39
5	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
6	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	6.2	32
7	Two novel RAD21 mutations in patients with mild Cornelia de Lange syndrome-like presentation and report of the first familial case. Gene, 2014, 537, 279-284.	2.2	31
8	Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). Bone, 2018, 107, 161-171.	2.9	23
9	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042.	2.4	23
10	Multigenerational autosomal dominant inheritance of 5p chromosomal deletions. American Journal of Medical Genetics, Part A, 2016, 170, 583-593.	1.2	21
11	De Novo Variants Disrupting the HX Repeat Motif of ATN1 Cause a Recognizable Non-Progressive Neurocognitive Syndrome. American Journal of Human Genetics, 2019, 104, 542-552.	6.2	19
12	Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. Genetics in Medicine, 2021, 23, 1465-1473.	2.4	10