Marisa A Vineyard

List of Publications by Year
in descending order

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

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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	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
2	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
3	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
4	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
5	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
6	Gnathodiaphyseal dysplasia: Severe atypical presentation with novel heterozygous mutation of the anoctamin gene (ANO5). Bone, 2018, 107, 161-171.	2.9	23
7	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
8	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
9	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
10	Multigenerational autosomal dominant inheritance of 5p chromosomal deletions. American Journal of Medical Genetics, Part A, 2016, 170, 583-593.	1.2	21
11	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
12	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