Neda Mazaheri

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6050947/publications.pdf

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20	424	840776 11	794594 19
papers	citations	h-index	g-index

22	22	22	907
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	8.2	68
2	Mutations in INPP5K Cause a Form of Congenital Muscular Dystrophy Overlapping Marinesco-SjÃ¶gren Syndrome and Dystroglycanopathy. American Journal of Human Genetics, 2017, 100, 537-545.	6.2	67
3	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625.	12.8	47
4	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056.	6.2	30
5	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26
6	Analysis of enriched rare variants in JPH2-encoded junctophilin-2 among Greater Middle Eastern individuals reveals a novel homozygous variant associated with neonatal dilated cardiomyopathy. Scientific Reports, 2019, 9, 9038.	3.3	22
7	Clinical presentation and natural history of infantile-onset ascending spastic paralysis from three families with an ALS2 founder variant. Neurological Sciences, 2018, 39, 1917-1925.	1.9	18
8	TMEM63C mutations cause mitochondrial morphology defects and underlie hereditary spastic paraplegia. Brain, 2022, 145, 3095-3107.	7.6	17
9	Autosomal recessive cardiomyopathy and sudden cardiac death associated with variants in MYL3. Genetics in Medicine, 2021, 23, 787-792.	2.4	16
10	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
11	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. Human Genetics, 2021, 140, 915-931.	3.8	16
12	Biallelic variants in ADARB1, encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy. Journal of Medical Genetics, 2021, 58, 495-504.	3.2	14
13	Biallelic MCM3AP mutations cause Charcot-Marie-Tooth neuropathy with variable clinical presentation. Brain, 2017, 140, e65-e65.	7.6	13
14	Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Genetics in Medicine, 2021, 23, 1933-1943.	2.4	11
15	Ameliorating Effect of Osteopontin on H2O2-Induced Apoptosis of Human Oligodendrocyte Progenitor Cells. Cellular and Molecular Neurobiology, 2018, 38, 891-899.	3.3	8
16	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. European Journal of Human Genetics, 2021, 29, 271-279.	2.8	8
17	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	6.2	8
18	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004.	1.3	7

#	Article	IF	Citations
19	Novel Homozygous Missense Mutation in RYR1 Leads to Severe Congenital Ptosis, Ophthalmoplegia, and Scoliosis in the Absence of Myopathy. Molecular Syndromology, 2018, 9, 25-29.	0.8	3
20	Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDCA) syndrome. Journal of Medical Genetics, 2021, 58, 815-831.	3.2	3