

Kristin G Monaghan

List of Publications by Year in descending order

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Version: 2024-02-01

16
papers

1,877
citations

687363

13
h-index

940533

16
g-index

18
all docs

18
docs citations

18
times ranked

5108
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical application of whole-exome sequencing across clinical indications. <i>Genetics in Medicine</i> , 2016, 18, 696-704.	2.4	780
2	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	6.2	230
3	Points to consider in the reevaluation and reanalysis of genomic test results: a statement of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2019, 21, 1267-1270.	2.4	147
4	Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. <i>American Journal of Human Genetics</i> , 2015, 97, 457-464.	6.2	134
5	The use of fetal exome sequencing in prenatal diagnosis: a points to consider document of the American College of Medical Genetics and Genomics (ACMG). <i>Genetics in Medicine</i> , 2020, 22, 675-680.	2.4	128
6	Whole-exome sequencing on deceased fetuses with ultrasound anomalies: expanding our knowledge of genetic disease during fetal development. <i>Genetics in Medicine</i> , 2017, 19, 1171-1178.	2.4	121
7	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 100, 352-363.	6.2	86
8	Mutations in <i>COQ4</i> , an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. <i>Journal of Medical Genetics</i> , 2015, 52, 627-635.	3.2	48
9	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epistatue of X chromosomes in females. <i>American Journal of Human Genetics</i> , 2021, 108, 502-516.	6.2	48
10	The role of objective facial analysis using FDNA in making diagnoses following whole exome analysis. Report of two patients with mutations in the BAF complex genes. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1754-1762.	1.2	39
11	Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. <i>Acta Neuropathologica</i> , 2020, 139, 415-442.	7.7	38
12	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. <i>Human Mutation</i> , 2021, 42, 445-459.	2.5	26
13	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. <i>Genetics in Medicine</i> , 2019, 21, 2036-2042.	2.4	23
14	Variants in GNAI1 cause a syndrome associated with variable features including developmental delay, seizures, and hypotonia. <i>Genetics in Medicine</i> , 2021, 23, 881-887.	2.4	13
15	<i>BET1</i> variants establish impaired vesicular transport as a cause for muscular dystrophy with epilepsy. <i>EMBO Molecular Medicine</i> , 2021, 13, e13787.	6.9	9
16	Genetic variants in DGAT1 cause diverse clinical presentations of malnutrition through a specific molecular mechanism. <i>European Journal of Medical Genetics</i> , 2020, 63, 103817.	1.3	6