Kristin G Monaghan

List of Publications by Year
in descending order

16	1,877	687363 13	940533 16
papers	citations	h-index	g-index

18	18	18	5108
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Variants in GNAI1 cause a syndrome associated with variable features including developmental delay, seizures, and hypotonia. Genetics in Medicine, 2021, 23, 881-887.	2.4	13
2	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
3	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	2.5	26
4	BET1 variants establish impaired vesicular transport as a cause for muscular dystrophy with epilepsy. EMBO Molecular Medicine, 2021, 13, e13787.	6.9	9
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). Genetics in Medicine, 2020, 22, 675-680.	2.4	128
6	Genetic variants in DGAT1 cause diverse clinical presentations of malnutrition through a specific molecular mechanism. European Journal of Medical Genetics, 2020, 63, 103817.	1.3	6
7	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. Acta Neuropathologica, 2020, 139, 415-442.	7.7	38
8	Points to consider in the reevaluation and reanalysis of genomic test results: a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2019, 21, 1267-1270.	2.4	147
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	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
11	Whole-exome sequencing on deceased fetuses with ultrasound anomalies: expanding our knowledge of genetic disease during fetal development. Genetics in Medicine, 2017, 19, 1171-1178.	2.4	121
12	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. American Journal of Medical Genetics, Part A, 2016, 170, 1754-1762.	1.2	39
13	Clinical application of whole-exome sequencing across clinical indications. Genetics in Medicine, 2016, 18, 696-704.	2.4	780
14	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
15	Mutations inCOQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. Journal of Medical Genetics, 2015, 52, 627-635.	3.2	48
16	Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. American Journal of Human Genetics, 2015, 97, 457-464.	6.2	134