Maria Bitner-Glindzicz

List of Publications by Year
in descending order

17	744	840776 11	888059 17
papers	citations	h-index	g-index

18	18	18	2136
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	3.2	141
2	Rapid Paediatric Sequencing (RaPS): comprehensive real-life workflow for rapid diagnosis of critically ill children. Journal of Medical Genetics, 2018, 55, 721-728.	3.2	98
3	Mutations in SNX14 Cause a Distinctive Autosomal-Recessive Cerebellar Ataxia and Intellectual Disability Syndrome. American Journal of Human Genetics, 2014, 95, 611-621.	6.2	89
4	The Oculome Panel Test. Ophthalmology, 2019, 126, 888-907.	5.2	77
5	SNX14 mutations affect endoplasmic reticulum-associated neutral lipid metabolism in autosomal recessive spinocerebellar ataxia 20. Human Molecular Genetics, 2018, 27, 1927-1940.	2.9	71
6	STAG3 truncating variant as the cause of primary ovarian insufficiency. European Journal of Human Genetics, 2016, 24, 135-138.	2.8	53
7	Genetic investigations in childhood deafness. Archives of Disease in Childhood, 2015, 100, 271-278.	1.9	47
8	Mitochondrial m.1584A 12S m62A rRNA methylation in families with m.1555A>G associated hearing loss. Human Molecular Genetics, 2015, 24, 1036-1044.	2.9	43
9	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	6.2	37
10	The effect of the common c.2299delG mutation in USH2A on RNA splicing. Experimental Eye Research, 2014, 122, 9-12.	2.6	23
11	Lamination of the Outer Plexiform Layer in Optic Atrophy Caused by Dominant WFS1 Mutations. Ophthalmology, 2016, 123, 1624-1626.	5.2	19
12	Exome sequencing identifies variants in FKBP4 that are associated with recurrent fetal loss in humans. Human Molecular Genetics, 2019, 28, 3466-3474.	2.9	13
13	An example of the utility of genomic analysis for fast and accurate clinical diagnosis of complex rare phenotypes. Orphanet Journal of Rare Diseases, 2017, 12, 24.	2.7	10
14	WFS1-Associated Optic Neuropathy: Genotype-Phenotype Correlations and Disease Progression. American Journal of Ophthalmology, 2022, 241, 9-27.	3.3	8
15	Clinical utility gene card for: Wolfram syndrome. European Journal of Human Genetics, 2016, 24, 1-4.	2.8	6
16	The timing of auditory sensory deficits in Norrie disease has implications for therapeutic intervention. JCI Insight, 2022, 7, .	5.0	6
17	Republished: Genetic investigations in childhood deafness. Postgraduate Medical Journal, 2015, 91, 395-402.	1.8	3