Stephan Waldmueller

List of Publications by Year
in descending order

7 papers	110 citations	1684188 5 h-index	1872680 6 g-index
7 all docs	7 docs citations	7 times ranked	276 citing authors

#	Article	IF	Citations
1	Bi-allelic loss-of-function variants inKIF21Acause severe fetal akinesia with arthrogryposis multiplex. Journal of Medical Genetics, 2023, 60, 48-56.	3.2	26
2	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	7.6	8
3	A single center experience of prenatal parentâ€fetus trio exome sequencing for pregnancies with congenital anomalies. Prenatal Diagnosis, 2022, 42, 901-910.	2.3	4
4	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
5	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004.	1.3	7
6	Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions. American Journal of Human Genetics, 2017, 101, 985-994.	6.2	44
7	Recognizable pattern of arthrogryposis and congenital myopathy caused by the recurrent TTN metatranscript-only c.39974-11T>G splice variant. Neuropediatrics, 0, 0, .	0.6	5