

Stephan Waldmueller

List of Publications by Year in descending order

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Version: 2024-02-01

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	Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions. <i>American Journal of Human Genetics</i> , 2017, 101, 985-994.	6.2	44
2	Bi-allelic loss-of-function variants in <i>KIF21A</i> cause severe fetal akinesia with arthrogyryposis multiplex. <i>Journal of Medical Genetics</i> , 2023, 60, 48-56.	3.2	26
3	DLG4-related synaptopathy: a new rare brain disorder. <i>Genetics in Medicine</i> , 2021, 23, 888-899.	2.4	16
4	Biallelic <i>ADAM22</i> pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. <i>Brain</i> , 2022, 145, 2301-2312.	7.6	8
5	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). <i>European Journal of Medical Genetics</i> , 2020, 63, 104004.	1.3	7
6	Recognizable pattern of arthrogyryposis and congenital myopathy caused by the recurrent TTN metatranscript-only c.39974-11T>G splice variant. <i>Neuropediatrics</i> , 0, 0, .	0.6	5
7	A single center experience of prenatal parentâ€fetus trio exome sequencing for pregnancies with congenital anomalies. <i>Prenatal Diagnosis</i> , 2022, 42, 901-910.	2.3	4