Stefanie Brock

List of Publications by Year in descending order

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Version: 2024-02-01

1478505 1588992 10 118 8 6 citations h-index g-index papers 10 10 10 243 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Tubulinopathies continued: refining the phenotypic spectrum associated with variants in TUBG1. European Journal of Human Genetics, 2018, 26, 1132-1142.	2.8	30
2	TMX2 Is a Crucial Regulator of Cellular Redox State, and Its Dysfunction Causes Severe Brain Developmental Abnormalities. American Journal of Human Genetics, 2019, 105, 1126-1147.	6.2	25
3	Genetic heterogeneity of polymicrogyria: study of 123 patients using deep sequencing. Brain Communications, 2021, 3, fcaa221.	3.3	22
4	Recurrent NEDD4L Variant in Periventricular Nodular Heterotopia, Polymicrogyria and Syndactyly. Frontiers in Genetics, 2020, 11, 26.	2.3	12
5	Defining the phenotypical spectrum associated with variants in <i>TUBB2A</i> . Journal of Medical Genetics, 2021, 58, 33-40.	3.2	11
6	Neuropathology of genetically defined malformations of cortical developmentâ€"A systematic literature review. Neuropathology and Applied Neurobiology, 2021, 47, 585-602.	3.2	9
7	Overcoming the Challenges of High Quality RNA Extraction from Core Needle Biopsy. Biomolecules, 2021, 11, 621.	4.0	7
8	Malformations of cerebral development and clues from the peripheral nervous system: A systematic literature review. European Journal of Paediatric Neurology, 2022, 37, 155-164.	1.6	1
9	Overlapping cortical malformations in patients with pathogenic variants in <i>GRIN1</i> and <i>GRIN2B</i> . Journal of Medical Genetics, 2023, 60, 183-192.	3.2	1
10	A rare cause of postmenopausal hyperandrogenism. BMJ Case Reports, 2021, 14, e237505.	0.5	0