Daniele Frattini

List of Publications by Year
in descending order

12	84	1478505 6	1474206 9
papers	citations	h-index	g-index

12	12	12	199
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical and radiological correlates of activities of daily living in cerebellar atrophy caused by PMM2 mutations (PMM2-CDG). Cerebellum, 2021, 20, 596-605.	2.5	8
2	Further delineation of PIGB-related early infantile epileptic encephalopathy. European Journal of Medical Genetics, 2021, 64, 104268.	1.3	1
3	Pharmacological Treatment of Severe Breathing Abnormalities in a Case of HNRNPU Epileptic Encephalopathy. Molecular Syndromology, 2021, 12, 101-105.	0.8	4
4	Beneficial effects of the ketogenic diet on drugâ€resistant epileptic encephalopathy associated with a de novo NBEA pathogenic variant. Epileptic Disorders, 2021, 23, 739-743.	1.3	3
5	SPG6 (NIPA1 variant): A report of a case with early-onset complex hereditary spastic paraplegia and brief literature review. Journal of Clinical Neuroscience, 2021, 94, 281-285.	1.5	11
6	New biallelic GBA2 variant in a patient with SPG46. Clinical Neurology and Neurosurgery, 2020, 191, 105676.	1.4	8
7	Paediatricâ€onset hereditary spastic paraplegias: a retrospective cohort study. Developmental Medicine and Child Neurology, 2020, 62, 1068-1074.	2.1	12
8	Neuropsychological and behavioral disorders as presentation symptoms in two brothers with early-infantile Niemann-Pick type C. Acta Biomedica, 2020, 91, e2020075.	0.3	0
9	Long-term follow-up until early adulthood in autosomal dominant, complex SPG30 with a novel KIF1A variant: a case report. Italian Journal of Pediatrics, 2019, 45, 155.	2.6	10
10	KCNQ2 encephalopathy: A case due to a de novo deletion. Brain and Development, 2018, 40, 65-68.	1.1	18
11	Long-term follow-up in spastic paraplegia due to SPG56/CYP2U1: age-dependency rather than genetic variability?. Journal of Neurology, 2017, 264, 586-588.	3.6	9
12	Dilated Virchow-Robin spaces mimicking white matter disease in a XYY syndrome. Journal of Pediatric Neurology, 2015, 09, 359-363.	0.2	0