

Daniele Frattini

List of Publications by Year in descending order

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Version: 2024-02-01

12
papers

84
citations

1478505

6
h-index

1474206

9
g-index

12
all docs

12
docs citations

12
times ranked

199
citing authors

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