

Anne Maria Rochtus

List of Publications by Year in descending order

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Version: 2024-02-01

14
papers

384
citations

1040056

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h-index

1199594

12
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15
all docs

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docs citations

15
times ranked

909
citing authors

#	ARTICLE	IF	CITATIONS
1	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. <i>Annals of Neurology</i> , 2019, 86, 821-831.	5.3	96
2	Genetic diagnoses in epilepsy: The impact of dynamic exome analysis in a pediatric cohort. <i>Epilepsia</i> , 2020, 61, 249-258.	5.1	85
3	Genome-wide DNA methylation analysis of pseudohypoparathyroidism patients with GNAS imprinting defects. <i>Clinical Epigenetics</i> , 2016, 8, 10.	4.1	53
4	Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphate-binding region. <i>Epilepsia</i> , 2019, 60, 406-418.	5.1	53
5	Mutations in NRXN1 and NRXN2 in a patient with early-onset epileptic encephalopathy and respiratory depression. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003442.	1.2	20
6	Methylome analysis for spina bifida shows SOX18 hypomethylation as a risk factor with evidence for a complex (epi)genetic interplay to affect neural tube development. <i>Clinical Epigenetics</i> , 2016, 8, 108.	4.1	18
7	<i>BRAT1</i> encephalopathy: a recessive cause of epilepsy of infancy with migrating focal seizures. <i>Developmental Medicine and Child Neurology</i> , 2020, 62, 1096-1099.	2.1	18
8	The role of sodium channels in sudden unexpected death in pediatrics. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1309.	1.2	14
9	Antiepileptic Drug Teratogenicity and De Novo Genetic Variation Load. <i>Annals of Neurology</i> , 2020, 87, 897-906.	5.3	9
10	Antiseizure medications and thyroid hormone homeostasis: Literature review and practical recommendations. <i>Epilepsia</i> , 2022, 63, 259-270.	5.1	9
11	Hypothalamic lipoma and growth hormone deficiency. <i>International Journal of Pediatric Endocrinology (Springer)</i> , 2020, 2020, 4.	1.6	5
12	Expanding the clinical spectrum of Fowler syndrome: Three siblings with survival into adulthood and systematic review of the literature. <i>Clinical Genetics</i> , 2020, 98, 423-432.	2.0	4
13	SAT-240 Hypothalamic Lipoma and Growth Hormone Deficiency. <i>Journal of the Endocrine Society</i> , 2020, 4, .	0.2	0
14	Antiepileptic Drugs and Thyroid Hormone Homeostasis: Literature Review and Practical Guideline. <i>Journal of the Endocrine Society</i> , 2021, 5, A824-A824.	0.2	0