

Raffaella Minardi

List of Publications by Year in descending order

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Version: 2024-02-01

18
papers

554
citations

1040056

9
h-index

888059

17
g-index

21
all docs

21
docs citations

21
times ranked

1153
citing authors

#	ARTICLE	IF	CITATIONS
1	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. <i>American Journal of Human Genetics</i> , 2019, 105, 267-282.	6.2	237
2	Polygenic burden in focal and generalized epilepsies. <i>Brain</i> , 2019, 142, 3473-3481.	7.6	90
3	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17,458 subjects. <i>Brain</i> , 2020, 143, 2106-2118.	7.6	47
4	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. <i>American Journal of Human Genetics</i> , 2021, 108, 965-982.	6.2	35
5	Gene Panel Analysis in a Large Cohort of Patients With Autosomal Dominant Polycystic Kidney Disease Allows the Identification of 80 Potentially Causative Novel Variants and the Characterization of a Complex Genetic Architecture in a Subset of Families. <i>Frontiers in Genetics</i> , 2020, 11, 464.	2.3	26
6	Sleep-related hypermotor epilepsy (SHE): Contribution of known genes in 103 patients. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2020, 74, 60-64.	2.0	25
7	Whole-exome sequencing in adult patients with developmental and epileptic encephalopathy: It is never too late. <i>Clinical Genetics</i> , 2020, 98, 477-485.	2.0	25
8	Contribution of ultrarare variants in mTOR pathway genes to sporadic focal epilepsies. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 475-485.	3.7	15
9	Epilepsy in <i>MT-ATP6</i> -related mfs/NARP: correlation of electroclinical features with heteroplasmy. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 704-710.	3.7	10
10	Autosomal dominant lateral temporal lobe epilepsy associated with a novel reelin mutation. <i>Epileptic Disorders</i> , 2020, 22, 443-448.	1.3	8
11	Epilepsy with eyelid myoclonias and Sotos syndrome features in a patient with compound heterozygous missense variants in <i>APC2</i> gene. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2020, 83, 169-171.	2.0	7
12	Epilepsy with auditory features: Contribution of known genes in 112 patients. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2021, 85, 115-118.	2.0	6
13	Accurate Detection of Hot-Spot MTOR Somatic Mutations in Archival Surgical Specimens of Focal Cortical Dysplasia by Molecular Inversion Probes. <i>Molecular Diagnosis and Therapy</i> , 2020, 24, 571-577.	3.8	5
14	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. <i>Epilepsia</i> , 2021, 62, 1518-1527.	5.1	5
15	<i>SCN1A</i> mutations in focal epilepsy with auditory features: widening the spectrum of GEFS <i>plus</i> . <i>Epileptic Disorders</i> , 2019, 21, 185-191.	1.3	5
16	Epilepsy With Auditory Features: From Etiology to Treatment. <i>Frontiers in Neurology</i> , 2021, 12, 807939.	2.4	2
17	A novel compound heterozygous mutation in an adolescent with insulin-dependent diabetes: The challenge of characterizing Wolfram syndrome. <i>Diabetes Research and Clinical Practice</i> , 2016, 121, 59-61.	2.8	1
18	Mild neurological phenotype in a family carrying a novel N-terminal null <i>GRIN2A</i> variant. <i>European Journal of Medical Genetics</i> , 2022, 65, 104500.	1.3	1