Raffaella Minardi

List of Publications by Year
in descending order

18	554	1040056 9	888059 17
papers	citations	h-index	g-index

21	21	21	1153
all docs	docs citations	times ranked	citing authors

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