

Caroline Nava

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

21
papers

1,114
citations

13
h-index

24
g-index

24
ext. papers

1,516
ext. citations

8.7
avg, IF

2.48
L-index

#	Paper	IF	Citations
21	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. <i>Human Genetics</i> , 2021 , 140, 1109-1120	6.3	4
20	Variants in the degnon of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. <i>American Journal of Human Genetics</i> , 2021 , 108, 857-873	11.1	2
19	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. <i>Brain</i> , 2021 ,	11.2	6
18	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. <i>Genetics in Medicine</i> , 2021 , 23, 653-660	8.1	5
17	Lessons learned from 40 novel PIGA patients and a review of the literature. <i>Epilepsia</i> , 2020 , 61, 1142-1155	5.4	15
16	PAK3 mutations responsible for severe intellectual disability and callosal agenesis inhibit cell migration. <i>Neurobiology of Disease</i> , 2020 , 136, 104709	7.5	8
15	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. <i>American Journal of Human Genetics</i> , 2019 , 104, 1210-1222	11	31
14	Novel GABRA2 variants in epileptic encephalopathy and intellectual disability with seizures. <i>Brain</i> , 2019 , 142, e15	11.2	5
13	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. <i>Nature Communications</i> , 2019 , 10, 3094	17.4	76
12	Re-annotation of 191 developmental and epileptic encephalopathy-associated genes unmasks de novo variants in. <i>Npj Genomic Medicine</i> , 2019 , 4, 31	6.2	12
11	Epilepsy with migrating focal seizures: KCNT1 mutation hotspots and phenotype variability. <i>Neurology: Genetics</i> , 2019 , 5, e363	3.8	18
10	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. <i>Genetics in Medicine</i> , 2019 , 21, 837-849	8.1	32
9	Defining the phenotypic spectrum of SLC6A1 mutations. <i>Epilepsia</i> , 2018 , 59, 389-402	6.4	54
8	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. <i>Nature Communications</i> , 2018 , 9, 4619	17.4	39
7	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. <i>PLoS Genetics</i> , 2018 , 14, e1007671	6	11
6	HCN1 mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. <i>Brain</i> , 2018 , 141, 3160-3178	11.2	48
5	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. <i>Brain</i> , 2017 , 140, 1316-1336	11.2	285

4	Copy Number Variations Found in Patients with a Corpus Callosum Abnormality and Intellectual Disability. <i>Journal of Pediatrics</i> , 2017 , 185, 160-166.e1	3.6	16
3	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017 , 101, 664-685	11	214
2	Human Pluripotent Stem Cell-derived Cortical Neurons for High Throughput Medication Screening in Autism: A Proof of Concept Study in SHANK3 Haploinsufficiency Syndrome. <i>EBioMedicine</i> , 2016 , 9, 293-305	8.8	55
1	Cardio-facio-cutaneous and Noonan syndromes due to mutations in the RAS/MAPK signalling pathway: genotype-phenotype relationships and overlap with Costello syndrome. <i>Journal of Medical Genetics</i> , 2007 , 44, 763-71	5.8	178