Caroline Nava

List of Publications by Citations

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	Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. *Brain*, **2017**, 140, 1316-1336	11.2	285
20	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
19	Cardio-facio-cutaneous and Noonan syndromes due to mutations in the RAS/MAPK signalling pathway: genotype-phenotype relationships and overlap with Costello syndrome. *Journal of Medical Genetics*, **2007**, 44, 763-71	5.8	178
18	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. *Nature Communications*, **2019**, 10, 3094	17.4	76
17	Human Pluripotent Stem Cell-derived Cortical Neurons for High Throughput Medication Screening in Autism: A Proof of Concept Study in SHANK3 Haploinsufficiency Syndrome. *EBioMedicine*, **2016**, 9, 293-305	8.8	55
16	Defining the phenotypic spectrum of SLC6A1 mutations. *Epilepsia*, **2018**, 59, 389-402	6.4	54
15	HCN1 mutation spectrum: from neonatal epileptic encephalopathy to benign generalized epilepsy and beyond. *Brain*, **2018**, 141, 3160-3178	11.2	48
14	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. *Nature Communications*, **2018**, 9, 4619	17.4	39
13	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. *Genetics in Medicine*, **2019**, 21, 837-849	8.1	32
12	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
11	Epilepsy with migrating focal seizures: KCNT1 mutation hotspots and phenotype variability. *Neurology: Genetics*, **2019**, 5, e363	3.8	18
10	Copy Number Variations Found in Patients with a Corpus Callosum Abnormality and Intellectual Disability. *Journal of Pediatrics*, **2017**, 185, 160-166.e1	3.6	16
9	Lessons learned from 40 novel PIGA patients and a review of the literature. *Epilepsia*, **2020**, 61, 1142-1154	5.4	15
8	Re-annotation of 191 developmental and epileptic encephalopathy-associated genes unmasks de novo variants in. *Npj Genomic Medicine*, **2019**, 4, 31	6.2	12
7	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. *PLoS Genetics*, **2018**, 14, e1007671	6	11
6	PAK3 mutations responsible for severe intellectual disability and callosal agenesis inhibit cell migration. *Neurobiology of Disease*, **2020**, 136, 104709	7.5	8
5	KCNT1-related epilepsies and epileptic encephalopathies: phenotypic and mutational spectrum. *Brain*, **2021**,	11.2	6

List of Publications

4	Novel GABRA2 variants in epileptic encephalopathy and intellectual disability with seizures. *Brain*, **2019**, 142, e15	11.2	5
3	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. *Genetics in Medicine*, **2021**, 23, 653-660	8.1	5
2	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. *Human Genetics*, **2021**, 140, 1109-1120	6.3	4
1	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. *American Journal of Human Genetics*, **2021**, 108, 857-873	11	2