Identification of new risk factors for rolandic epilepsy: cholinergic synapses

Journal of Medical Genetics

55, 607-616

DOI: 10.1136/jmedgenet-2018-105319

Citation Report

#	Article	IF	Citations
1	Epilepsy phenotype in patients with Xp22.31 microduplication. Epilepsy & Behavior Case Reports, 2019, 11, 31-34.	1.5	3
2	Neurodevelopmental and associated changes in a patient with Xp22.31 duplication. Neurological Sciences, 2020, 41, 713-716.	0.9	2
3	Arfgef1 haploinsufficiency in mice alters neuronal endosome composition and decreases membrane surface postsynaptic GABAA receptors. Neurobiology of Disease, 2020, 134, 104632.	2.1	8
4	Medical and neurobehavioural phenotypes in male and female carriers of Xp22.31 duplications in the UK Biobank. Human Molecular Genetics, 2020, 29, 2872-2881.	1.4	11
5	Exome sequencing in 57 patients with self-limited focal epilepsies of childhood with typical or atypical presentations suggests novel candidate genes. European Journal of Paediatric Neurology, 2020, 27, 104-110.	0.7	17
6	Temporal trends in incidence of Rolandic epilepsy, prevalence of comorbidities and prescribing trends: birth cohort study. Archives of Disease in Childhood, 2020, 105, 569-574.	1.0	8
7	Case Report: Whole Exome Sequencing Revealed Disease-Causing Variants in Two Genes in a Patient With Autism Spectrum Disorder, Intellectual Disability, Hyperactivity, Sleep and Gastrointestinal Disturbances. Frontiers in Genetics, 2021, 12, 625564.	1.1	8
8	Investigation of long interspersed elementâ€1 retrotransposons as potential risk factors for idiopathic temporal lobe epilepsy. Epilepsia, 2021, 62, 1329-1342.	2.6	6
9	Integrated 16S rRNA Gene Sequencing and LC-MS Analysis Revealed the Interplay Between Gut Microbiota and Plasma Metabolites in Rats With Ischemic Stroke. Journal of Molecular Neuroscience, 2021, 71, 2095-2106.	1.1	19
10	Differential Clinical Features in Colombian Patients With Rolandic Epilepsy and Suggestion of Unlikely Association With GRIN2A, RBFOX1, or RBFOX3 Gene Variants. Journal of Child Neurology, 2021, 36, 875-882.	0.7	0
11	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. Genetics in Medicine, 2021, 23, 1901-1911.	1.1	9
12	<scp>ZMYND11</scp> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. Clinical Genetics, 2021, 100, 412-429.	1.0	5
13	Evaluation of candidate genes in a Chinese cohort of atypical Rolandic epilepsy. Epileptic Disorders, 2021, 23, 623-632.	0.7	3
14	Neurodevelopmental Disorders in Patients With Complex Phenotypes and Potential Complex Genetic Basis Involving Non-Coding Genes, and Double CNVs. Frontiers in Genetics, 2021, 12, 732002.	1.1	12
15	Nonsyndromic Early-Onset Epileptic Encephalopathies: Two Novel <i>KCTD7</i> Pathogenic Variants and a Literature Review. Developmental Neuroscience, 2021, 43, 348-357.	1.0	2
16	Perceived Gaps in Genetics Training Among Audiologists and Speech-Language Pathologists: Lessons From a National Survey. American Journal of Speech-Language Pathology, 2019, 28, 408-423.	0.9	11
18	Expanding the Phenotypic and Genotypic Spectrum of ARFGEF1-Related Neurodevelopmental Disorder. Frontiers in Molecular Neuroscience, 0, 15, .	1.4	1
19	Retrospective analysis of the sex chromosomal copy number variations in 186 fetuses using single nucleotide polymorphism arrays. Frontiers in Genetics, 0, 13, .	1.1	0

#	Article	IF	Citations
20	Xp22.31 copy number variations in 87 fetuses: refined genotypeâ€“phenotype correlations by prenatal and postnatal follow-up. BMC Medical Genomics, 2023, 16, .	0.7	1
21	Case report: Sex-specific characteristics of epilepsy phenotypes associated with Xp22.31 deletion: a case report and review. Frontiers in Genetics, 0, 14, .	1.1	0