Anne Claude Tabet

List of Publications by Year
in descending order

22	995	623734 14	642732 23
papers	citations	h-index	g-index

23	23	23	2302
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Rare and de novo duplications containing <scp>TCF20</scp> are associated with a neurodevelopmental disorder. Clinical Genetics, 2022, 101, 364-370.	2.0	7
2	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. Journal of Clinical Investigation, 2021, 131, .	8.2	24
3	The phenotypic and genetic spectrum of patients with heterozygous mutations in cyclin M2 (CNNM2). Human Mutation, 2021, 42, 473-486.	2.5	21
4	Operative list of genes associated with autism and neurodevelopmental disorders based on database review. Molecular and Cellular Neurosciences, 2021, 113, 103623.	2.2	51
5	<scp>EPHA7</scp> haploinsufficiency is associated with a neurodevelopmental disorder. Clinical Genetics, 2021, 100, 396-404.	2.0	3
6	A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder. Npj Genomic Medicine, 2021, 6, 91.	3.8	9
7	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
8	Telomere and Centromere Staining Followed by M-FISH Improves Diagnosis of Chromosomal Instability and Its Clinical Utility. Genes, 2020, 11, 475.	2.4	17
9	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	2.4	14
10	LEF1 haploinsufficiency causes ectodermal dysplasia. Clinical Genetics, 2020, 97, 595-600.	2.0	11
11	INTUâ€related oralâ€facialâ€digital syndrome type VI: A confirmatory report. Clinical Genetics, 2018, 93, 1205-1209.	2.0	7
12	EFNB2 haploinsufficiency causes a syndromic neurodevelopmental disorder. Clinical Genetics, 2018, 93, 1141-1147.	2.0	18
13	NR4A2 haploinsufficiency is associated with intellectual disability and autism spectrum disorder. Clinical Genetics, 2018, 94, 264-268.	2.0	22
14	Molecular and clinical delineation of 2p15p16.1 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2081-2087.	1.2	18
15	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	3.8	58
16	Complex nature of apparently balanced chromosomal rearrangements in patients with autism spectrum disorder. Molecular Autism, 2015, 6, 19.	4.9	29
17	Clinical and molecular delineation of Tetrasomy 9p syndrome: Report of 12 new cases and literature review. American Journal of Medical Genetics, Part A, 2015, 167, 1252-1261.	1.2	20
18	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	3.5	501

#	Article	IF	Citations
19	De novo deletion of TBL1XR1 in a child with nonâ€specific developmental delay supports its implication in intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 2335-2337.	1.2	24
20	Duplication of the 15q11-q13 region: Clinical and genetic study of 30 new cases. European Journal of Medical Genetics, 2014, 57, 5-14.	1.3	68
21	Autism multiplex family with 16p11.2p12.2 microduplication syndrome in monozygotic twins and distal 16p11.2 deletion in their brother. European Journal of Human Genetics, 2012, 20, 540-546.	2.8	38
22	Molecular characterization of a de novo 6q24.2q25.3 duplication interrupting UTRN in a patient with arthrogryposis. American Journal of Medical Genetics, Part A, 2010, 152A, 1781-1788.	1.2	13