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List of Publications by Year in descending order

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ΔΝΗ-ΤΗΠ ΝΙΔΜ

#	Article	IF	CITATIONS
1	Transcriptome analysis of human induced excitatory neurons supports a strong effect of clozapine on cholesterol biosynthesis. Schizophrenia Research, 2021, 228, 324-326.	2.0	13
2	Transcriptomic data of Clozapine-treated Ngn2-induced Human Excitatory Neurons. Data in Brief, 2021, 35, 106897.	1.0	4
3	SLC26A9 SNP rs7512462 is not associated with lung disease severity or lung function response to ivacaftor in cystic fibrosis patients with G551D-CFTR. Journal of Cystic Fibrosis, 2021, 20, 851-856.	0.7	11
4	Homozygous deletions implicate non-coding epigenetic marks in Autism spectrum disorder. Scientific Reports, 2020, 10, 14045.	3.3	12
5	Increased expression of anion transporter SLC26A9 delays diabetes onset in cystic fibrosis. Journal of Clinical Investigation, 2019, 130, 272-286.	8.2	33
6	Capitalizing on the heterogeneous effects of CFTR nonsense and frameshift variants to inform therapeutic strategy for cystic fibrosis. PLoS Genetics, 2018, 14, e1007723.	3.5	44
7	Biallelic mutations in human DCC cause developmental split-brain syndrome. Nature Genetics, 2017, 49, 606-612.	21.4	62
8	Integrated genome and transcriptome sequencing identifies a noncoding mutation in the genome replication factor <i>DONSON</i> as the cause of microcephaly-micromelia syndrome. Genome Research, 2017, 27, 1323-1335.	5.5	40
9	Systematic Computational Identification of Variants That Activate Exonic and Intronic Cryptic Splice Sites. American Journal of Human Genetics, 2017, 100, 751-765.	6.2	68
10	Mutations in mitochondrial enzyme GPT2 cause metabolic dysfunction and neurological disease with developmental and progressive features. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E5598-607.	7.1	51
11	Targeted DNA Sequencing from Autism Spectrum Disorder Brains Implicates Multiple Genetic Mechanisms. Neuron, 2015, 88, 910-917.	8.1	142
12	Mutations in QARS, Encoding Glutaminyl-tRNA Synthetase, Cause Progressive Microcephaly, Cerebral-Cerebellar Atrophy, and Intractable Seizures. American Journal of Human Genetics, 2014, 94, 547-558.	6.2	106
13	Somatic Mutations in Cerebral Cortical Malformations. New England Journal of Medicine, 2014, 371, 733-743.	27.0	326