

Anh-Thu N Lam

List of Publications by Year in descending order

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Version: 2024-02-01

13
papers

912
citations

933447

10
h-index

1125743

13
g-index

13
all docs

13
docs citations

13
times ranked

2561
citing authors

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