Ashleigh E Schaffer

List of Publications by Year
in descending order

21 papers	2,147 citations	516710 16 h-index	677142 22 g-index
22 all docs	22 docs citations	22 times ranked	4714 citing authors

#	Article	IF	Citations
1	Suppression of premature transcription termination leads to reduced mRNA isoform diversity and neurodegeneration. Neuron, 2022, 110, 1340-1357.e7.	8.1	12
2	An epilepsyâ€associated ACTL6B variant captures neuronal hyperexcitability in a human induced pluripotent stem cell model. Journal of Neuroscience Research, 2021, 99, 110-123.	2.9	7
3	Bi-allelic TTC5 variants cause delayed developmental milestones and intellectual disability. Journal of Medical Genetics, 2021, 58, 237-246.	3.2	4
4	tRNA Metabolism and Neurodevelopmental Disorders. Annual Review of Genomics and Human Genetics, 2019, 20, 359-387.	6.2	65
5	Biallelic loss of human CTNNA2, encoding Î±N-catenin, leads to ARP2/3 complex overactivity and disordered cortical neuronal migration. Nature Genetics, 2018, 50, 1093-1101.	21.4	70
6	Biallelic mutations in the 3â€² exonuclease TOE1 cause pontocerebellar hypoplasia and uncover a role in snRNA processing. Nature Genetics, 2017, 49, 457-464.	21.4	66
7	The RNA-binding protein, ZC3H14, is required for proper poly(A) tail length control, expression of synaptic proteins, and brain function in mice. Human Molecular Genetics, 2017, 26, 3663-3681.	2.9	31
8	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534.	21.4	111
9	An AKT3-FOXG1-reelin network underlies defective migration in human focal malformations of cortical development. Nature Medicine, 2015, 21, 1445-1454.	30.7	101
10	Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. Neuron, 2014, 84, 1226-1239.	8.1	95
11	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663.	28.9	228
12	Novel mutation in the fukutin gene in an Egyptian family with Fukuyama congenital muscular dystrophy and microcephaly. Gene, 2014, 539, 279-282.	2.2	5
13	Nkx6.1 Controls a Gene Regulatory Network Required for Establishing and Maintaining Pancreatic Beta Cell Identity. PLoS Genetics, 2013, 9, e1003274.	3.5	212
14	Exome Sequencing Can Improve Diagnosis and Alter Patient Management. Science Translational Medicine, 2012, 4, 138ra78.	12.4	226
15	A homozygous IER3IP1 mutation causes microcephaly with simplified gyral pattern, epilepsy, and permanent neonatal diabetes syndrome (MEDS). American Journal of Medical Genetics, Part A, 2012, 158A, 2788-2796.	1.2	42
16	Transgenic Overexpression of the Transcription Factor Nkx6.1 in Î²-Cells of Mice Does Not Increase Î²-Cell Proliferation, Î²-Cell Mass, or Improve Glucose Clearance. Molecular Endocrinology, 2011, 25, 1904-1914.	3.7	25
17	Sox9+ ductal cells are multipotent progenitors throughout development but do not produce new endocrine cells in the normal or injured adult pancreas. Development (Cambridge), 2011, 138, 653-665.	2.5	403
18	A Drosophila behavioral mutant, down and out (dao), is defective in an essential regulator of Erg potassium channels. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 5617-5621.	7.1	12

#	Article	IF	Citations
19	Nkx6 Transcription Factors and Ptf1a Function as Antagonistic Lineage Determinants in Multipotent Pancreatic Progenitors. Developmental Cell, 2010, 18, 1022-1029.	7.0	234
20	The transcription factors Nkx6.1 and Nkx6.2 possess equivalent activities in promoting beta-cell fate specification in Pdx1+ pancreatic progenitor cells. Development (Cambridge), 2007, 134, 2491-2500.	2.5	108
21	Requirements for endoderm and BMP signaling in sensory neurogenesis in zebrafish. Development (Cambridge), 2005, 132, 3731-3742.	2.5	82