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	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
2	Nkx6 Transcription Factors and Ptf1a Function as Antagonistic Lineage Determinants in Multipotent Pancreatic Progenitors. Developmental Cell, 2010, 18, 1022-1029.	7.0	234
3	CLP1 Founder Mutation Links tRNA Splicing and Maturation to Cerebellar Development and Neurodegeneration. Cell, 2014, 157, 651-663.	28.9	228
4	Exome Sequencing Can Improve Diagnosis and Alter Patient Management. Science Translational Medicine, 2012, 4, 138ra78.	12.4	226
5	Nkx6.1 Controls a Gene Regulatory Network Required for Establishing and Maintaining Pancreatic Beta Cell Identity. PLoS Genetics, 2013, 9, e1003274.	3.5	212
6	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534.	21.4	111
7	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
8	An AKT3-FOXG1-reelin network underlies defective migration in human focal malformations of cortical development. Nature Medicine, 2015, 21, 1445-1454.	30.7	101
9	Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. Neuron, 2014, 84, 1226-1239.	8.1	95
10	Requirements for endoderm and BMP signaling in sensory neurogenesis in zebrafish. Development (Cambridge), 2005, 132, 3731-3742.	2.5	82
11	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
12	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
13	tRNA Metabolism and Neurodevelopmental Disorders. Annual Review of Genomics and Human Genetics, 2019, 20, 359-387.	6.2	65
14	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
15	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
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	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
18	Suppression of premature transcription termination leads to reduced mRNA isoform diversity and neurodegeneration. Neuron, 2022, 110, 1340-1357.e7.	8.1	12

#	Article	IF	Citations
19	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
20	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
21	Bi-allelic TTC5 variants cause delayed developmental milestones and intellectual disability. Journal of Medical Genetics, 2021, 58, 237-246.	3.2	4