Susan A Slaugenhaupt

List of Publications by Year
in descending order

45	3,596	172457	265206
papers	citations	29 h-index	42 g-index

48	48	48	3780
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genome-wide association study reveals novel genetic loci: a new polygenic risk score for mitral valve prolapse. European Heart Journal, 2022, 43, 1668-1680.	2.2	25
2	Developmental regulation of neuronal gene expression by Elongator complex protein 1 dosage. Journal of Genetics and Genomics, 2022, 49, 654-665.	3.9	6
3	Selective retinal ganglion cell loss and optic neuropathy in a humanized mouse model of familial dysautonomia. Human Molecular Genetics, 2022, 31, 1776-1787.	2.9	7
4	Expanding the Genotypic Spectrum of Congenital Sensory and Autonomic Neuropathies Using Whole-Exome Sequencing. Neurology: Genetics, 2021, 7, e568.	1.9	6
5	A deep learning approach to identify gene targets of a therapeutic for human splicing disorders. Nature Communications, 2021, 12, 3332.	12.8	26
6	Development of a Screening Platform to Identify Small Molecules That Modify ELP1 Pre-mRNA Splicing in Familial Dysautonomia. SLAS Discovery, 2019, 24, 57-67.	2.7	18
7	Primary cilia defects causing mitral valve prolapse. Science Translational Medicine, 2019, 11, .	12.4	76
8	Genome-Wide Association Studyâ€“Driven Gene-Set Analyses, Genetic, and Functional Follow-Up Suggest GLIS1 as a Susceptibility Gene for Mitral Valve Prolapse. Circulation Genomic and Precision Medicine, 2019, 12, e002497.	3.6	31
9	ELP1 Splicing Correction Reverses Proprioceptive Sensory Loss in Familial Dysautonomia. American Journal of Human Genetics, 2019, 104, 638-650.	6.2	32
10	Exon-specific U1 snRNAs improve ELP1 exon 20 definition and rescue ELP1 protein expression in a familial dysautonomia mouse model. Human Molecular Genetics, 2018, 27, 2466-2476.	2.9	43
11	New insights into mitral valve dystrophy: a Filamin-A genotypeâ€“phenotype and outcome study. European Heart Journal, 2018, 39, 1269-1277.	2.2	44
12	Fingolimod phosphate inhibits astrocyte inflammatory activity in mucolipidosis IV. Human Molecular Genetics, 2018, 27, 2725-2738.	2.9	22
13	Familial dysautonomia: History, genotype, phenotype and translational research. Progress in Neurobiology, 2017, 152, 131-148.	5.7	87
14	Small Molecules for Early Endosome-Specific Patch Clamping. Cell Chemical Biology, 2017, 24, 907-916.e4.	5.2	34
15	Sensory and autonomic deficits in a new humanized mouse model of familial dysautonomia. Human Molecular Genetics, 2016, 25, 1116-1128.	2.9	40
16	Retinal Dystrophy and Optic Nerve Pathology inÂthe Mouse Model of Mucolipidosis IV. American Journal of Pathology, 2016, 186, 199-209.	3.8	22
17	Response to Letter Regarding Article, â€œFamilial Clustering of Mitral Valve Prolapse in the Communityâ€• Circulation, 2015, 132, e187-8.	1.6	0
18	Mitral valve diseaseâ€”morphology and mechanisms. Nature Reviews Cardiology, 2015, 12, 689-710.	13.7	281

#	Article	IF	Citations
19	Mutations in DCHS1 cause mitral valve prolapse. Nature, 2015, 525, 109-113.	27.8	150
20	Genetic association analyses highlight biological pathways underlying mitral valve prolapse. Nature Genetics, 2015, 47, 1206-1211.	21.4	103
21	Familial Clustering of Mitral Valve Prolapse in the Community. Circulation, 2015, 131, 263-268.	1.6	61
22	Developmental basis for filamin-A-associated myxomatous mitral valve disease. Cardiovascular Research, 2012, 96, 109-119.	3.8	68
23	Atrioventricular valve development: New perspectives on an old theme. Differentiation, 2012, 84, 103-116.	1.9	92
24	Specific correction of a splice defect in brain by nutritional supplementation. Human Molecular Genetics, 2011, 20, 4093-4101.	2.9	33
25	Kinetin Improves IKBKAP mRNA Splicing in Patients With Familial Dysautonomia. Pediatric Research, 2011, 70, 480-483.	2.3	83
26	Trace metal dyshomeostasis is associated with loss of TRPML1 ion channel function. FASEB Journal, 2010, 24, 708.3.	0.5	0
27	Kinetin in Familial Dysautonomia Carriers: Implications for a New Therapeutic Strategy Targeting mRNA Splicing. Pediatric Research, 2009, 65, 341-346.	2.3	38
28	Loss of Mouse Ikbkap, a Subunit of Elongator, Leads to Transcriptional Deficits and Embryonic Lethality That Can Be Rescued by Human IKBKAP. Molecular and Cellular Biology, 2009, 29, 736-744.	2.3	102
29	A humanized IKBKAP transgenic mouse models a tissue-specific human splicing defect. Genomics, 2007, 90, 389-396.	2.9	49
30	Weak definition ofIKBKAPexon 20 leads to aberrant splicing in familial dysautonomia. Human Mutation, 2007, 28, 41-53.	2.5	44
31	Therapeutic potential and mechanism of kinetin as a treatment for the human splicing disease familial dysautonomia. Journal of Molecular Medicine, 2007, 85, 149-161.	3.9	58
32	Transcription Impairment and Cell Migration Defects in Elongator-Depleted Cells: Implication for Familial Dysautonomia. Molecular Cell, 2006, 22, 521-531.	9.7	191
33	New Locus for Autosomal Dominant Mitral Valve Prolapse on Chromosome 13. Circulation, 2005, 112, 2022-2030.	1.6	142
34	Identification of the first non-Jewish mutation in familial Dysautonomia. American Journal of Medical Genetics Part A, 2003, 118A, 305-308.	2.4	64
35	Tissue-Specific Reduction in Splicing Efficiency of IKBKAP Due to the Major Mutation Associated with Familial Dysautonomia. American Journal of Human Genetics, 2003, 72, 749-758.	6.2	125
36	Rescue of a human mRNA splicing defect by the plant cytokinin kinetin. Human Molecular Genetics, 2003, 13, 429-436.	2.9	139

#	Article	IF	Citations
37	Genetics of familial dysautonomia. Tissue-specific expression of a splicing mutation in the IKBKAP gene. Clinical Autonomic Research, 2002, 12, I15-I19.	2.5	8
38	Familial dysautonomia. Current Opinion in Genetics and Development, 2002, 12, 307-311.	3.3	332
39	Targeted genome screen of panic disorder and anxiety disorder proneness using homology to murine QTL regions. American Journal of Medical Genetics Part A, 2001, 105, 195-206.	2.4	85
40	Tissue-Specific Expression of a Splicing Mutation in the Gene Causes Familial Dysautonomia. American Journal of Human Genetics, 2001, 68, 598-605.	6.2	558
41	Genetic association analysis of behavioral inhibition using candidate loci from mouse models. American Journal of Medical Genetics Part A, 2001, 105, 226-235.	2.4	58
42	Intestinal cell kinase (ICK) localizes to the crypt region and requires a dual phosphorylation site found in map kinases. , 2000, 183, 129-139.		47
43	Cloning, mapping, and expression of a novel brain-specific transcript in the Familial Dysautonomia candidate region on Chromosome 9q31. Mammalian Genome, 2000, 11, 81-83.	2.2	5
44	Prenatal diagnostic testing for familial dysautonomia using linked genetic markers. Prenatal Diagnosis, 1995, 15, 817-826.	2.3	11
45	Localization of the gene for familial dysautonomia on chromosome 9 and definition of DNA markers for genetic diagnosis. Nature Genetics, 1993, 4, 160-164.	21.4	149