Miriam H Meisler

List of Publications by Year
in descending order

82	9,791	81900 39	56724 83
papers	citations	h-index	g-index

85	85	85	14772
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	9.1	3,122
2	Mutations of SCN1A, encoding a neuronal sodium channel, in two families with GEFS+2. Nature Genetics, 2000, 24, 343-345.	21.4	910
3	Sodium channel mutations in epilepsy and other neurological disorders. Journal of Clinical Investigation, 2005, 115, 2010-2017.	8.2	427
4	Altered Subthreshold Sodium Currents and Disrupted Firing Patterns in Purkinje Neurons of Scn8a Mutant Mice. Neuron, 1997, 19, 881-891.	8.1	367
5	De Novo Pathogenic SCN8A Mutation Identified by Whole-Genome Sequencing of a Family Quartet Affected by Infantile Epileptic Encephalopathy and SUDEP. American Journal of Human Genetics, 2012, 90, 502-510.	6.2	365
6	Mutation of a new sodium channel gene, Scn8a, in the mouse mutant â€˜motor endplate diseaseâ€™. Nature Genetics, 1995, 10, 461-465.	21.4	286
7	Functional Analysis of the Mouse Scn8a Sodium Channel. Journal of Neuroscience, 1998, 18, 6093-6102.	3.6	227
8	Altered Function of the SCN1A Voltage-gated Sodium Channel Leads to Î³-Aminobutyric Acid-ergic (GABAergic) Interneuron Abnormalities. Journal of Biological Chemistry, 2010, 285, 9823-9834.	3.4	200
9	The splicing regulator Rbfox2 is required for both cerebellar development and mature motor function. Genes and Development, 2012, 26, 445-460.	5.9	186
10	Sodium channel gene family: epilepsy mutations, gene interactions and modifier effects. Journal of Physiology, 2010, 588, 1841-1848.	2.9	183
11	Incidence of Dravet Syndrome in a US Population. Pediatrics, 2015, 136, e1310-e1315.	2.1	178
12	Functional Effects of Two Voltage-Gated Sodium Channel Mutations That Cause Generalized Epilepsy with Febrile Seizures Plus Type 2. Journal of Neuroscience, 2001, 21, 7481-7490.	3.6	173
13	Alternative Splicing of the Sodium Channel SCN8A Predicts a Truncated Two-domain Protein in Fetal Brain and Non-neuronal Cells. Journal of Biological Chemistry, 1997, 272, 24008-24015.	3.4	135
14	Sodium channel SCN8A (Nav1.6): properties and de novo mutations in epileptic encephalopathy and intellectual disability. Frontiers in Genetics, 2013, 4, 213.	2.3	127
15	Convulsive seizures and SUDEP in a mouse model of SCN8A epileptic encephalopathy. Human Molecular Genetics, 2015, 24, 506-515.	2.9	124
16	De novo gain-of-function and loss-of-function mutations ofSCN8Ain patients with intellectual disabilities and epilepsy. Journal of Medical Genetics, 2015, 52, 330-337.	3.2	124
17	SCNM1, a Putative RNA Splicing Factor That Modifies Disease Severity in Mice. Science, 2003, 301, 967-969.	12.6	122
18	Impaired Motor Function in Mice With Cell-Specific Knockout of Sodium Channel Scn8a (NaV1.6) in Cerebellar Purkinje Neurons and Granule Cells. Journal of Neurophysiology, 2006, 96, 785-793.	1.8	111

#	Article	IF	Citations
19	Heterozygous mutations of the voltage-gated sodium channel SCN8A are associated with spike-wave discharges and absence epilepsy in mice. Human Molecular Genetics, 2009, 18, 1633-1641.	2.9	110
20	SCN8A encephalopathy: Research progress and prospects. Epilepsia, 2016, 57, 1027-1035.	5.1	101
21	Recurrent and Non-Recurrent Mutations of SCN8A in Epileptic Encephalopathy. Frontiers in Neurology, 2015, 6, 104.	2.4	99
22	A novel de novo mutation of SCN8A (Nav1.6) with enhanced channel activation in a child with epileptic encephalopathy. Neurobiology of Disease, 2014, 69, 117-123.	4.4	96
23	Pathogenic mechanism of recurrent mutations of <scp>SCN8A</scp> in epileptic encephalopathy. Annals of Clinical and Translational Neurology, 2016, 3, 114-123.	3.7	96
24	Characterization of a de novo SCN8A mutation in a patient with epileptic encephalopathy. Epilepsy Research, 2014, 108, 1511-1518.	1.6	92
25	Molecular and pathological effects of a modifier gene on deficiency of the sodium channel Scn8a (Nav1.6). Human Molecular Genetics, 2002, 11, 2765-2775.	2.9	90
26	Scn8a Antisense Oligonucleotide Is Protective in Mouse Models of SCN8A Encephalopathy and Dravet Syndrome. Annals of Neurology, 2020, 87, 339-346.	5.3	87
27	Exon Organization, Coding Sequence, Physical Mapping, and Polymorphic Intragenic Markers for the Human Neuronal Sodium Channel GeneSCN8A. Genomics, 1998, 54, 287-296.	2.9	86
28	Sodium channelopathies in neurodevelopmental disorders. Nature Reviews Neuroscience, 2021, 22, 152-166.	10.2	79
29	Prominent role of forebrain excitatory neurons inSCN8Aencephalopathy. Brain, 2019, 142, 362-375.	7.6	69
30	Loss-of-function variants of SCN8A in intellectual disability without seizures. Neurology: Genetics, 2017, 3, e170.	1.9	66
31	Neuronal hyperexcitability in a mouse model of SCN8A epileptic encephalopathy. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 2383-2388.	7.1	64
32	Allelic mutations of the sodium channel SCN8A reveal multiple cellular and physiological functions. Genetica, 2004, 122, 37-45.	1.1	60
33	Sodium Channels and Neurological Disease: Insights from Scn8a Mutations in the Mouse. Neuroscientist, 2001, 7, 136-145.	3.5	58
34	C9<scp>ORF</scp>72 expansion in a family with bipolar disorder. Bipolar Disorders, 2013, 15, 326-332.	1.9	58
35	Inactivation of sodium channel Scn8A (Nav1.6) in purkinje neurons impairs learning in Morris Water Maze and delay but not trace eyeblink classical conditioning.. Behavioral Neuroscience, 2006, 120, 229-240.	1.2	54
36	Cardiac arrhythmia in a mouse model of sodium channel SCN8A epileptic encephalopathy. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 12838-12843.	7.1	54

#	Article	IF	Citations
37	The novel sodium channel modulator <scp>GS</scp>â€458967 (<scp>GS</scp>967) is an effective treatment in a mouse model of <scp>SCN</scp>8A encephalopathy. Epilepsia, 2018, 59, 1166-1176.	5.1	53
38	Persistent Nav1.6 current at axon initial segments tunes spike timing of cerebellar granule cells. Journal of Physiology, 2010, 588, 651-670.	2.9	49
39	Biallelic Mutations of VAC14 in Pediatric-Onset Neurological Disease. American Journal of Human Genetics, 2016, 99, 188-194.	6.2	45
40	Rbfox proteins regulate alternative splicing of neuronal sodium channel SCN8A. Molecular and Cellular Neurosciences, 2012, 49, 120-126.	2.2	43
41	Evidence for a direct role of the disease modifier SCNM1 in splicing. Human Molecular Genetics, 2007, 16, 2506-2516.	2.9	41
42	The SCN8A encephalopathy mutation p.Ile1327Val displays elevated sensitivity to the anticonvulsant phenytoin. Epilepsia, 2016, 57, 1458-1466.	5.1	41
43	Aberrant Sodium Channel Currents and Hyperexcitability of Medial Entorhinal Cortex Neurons in a Mouse Model of SCN8A Encephalopathy. Journal of Neuroscience, 2017, 37, 7643-7655.	3.6	41
44	Loss of Fig4 in both Schwann cells and motor neurons contributes to CMT4J neuropathy. Human Molecular Genetics, 2015, 24, 383-396.	2.9	39
45	Partial loss-of-function of sodium channel SCN8A in familial isolated myoclonus. Human Mutation, 2018, 39, 965-969.	2.5	34
46	Antisense Oligonucleotide Therapy for Neurodevelopmental Disorders. Developmental Neuroscience, 2021, 43, 247-252.	2.0	34
47	Interaction of Voltage-gated Sodium Channel Nav1.6 (SCN8A) with Microtubule-associated Protein Map1b. Journal of Biological Chemistry, 2012, 287, 18459-18466.	3.4	32
48	SCN8A encephalopathy: Mechanisms and models. Epilepsia, 2019, 60, S86-S91.	5.1	32
49	Ion Channel Mutations in Mouse Models of Inherited Neurological Disease. Annals of Medicine, 1997, 29, 569-574.	3.8	31
50	Evaluation of the Golgi trafficking protein VPS54 (wobbler) as a candidate for ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 141-148.	2.1	31
51	Altered gene expression profile in a mouse model of SCN8A encephalopathy. Experimental Neurology, 2017, 288, 134-141.	4.1	27
52	Mouse Models of PI(3,5)P2 Deficiency with Impaired Lysosome Function. Methods in Enzymology, 2014, 534, 245-260.	1.0	25
53	PI(3,5)P2 biosynthesis regulates oligodendrocyte differentiation by intrinsic and extrinsic mechanisms. ELife, 2016, 5, .	6.0	25
54	Postictal Death Is Associated with Tonic Phase Apnea in a Mouse Model of Sudden Unexpected Death in Epilepsy. Annals of Neurology, 2021, 89, 1023-1035.	5.3	25

#	Article	IF	Citations
55	The MAP1B Binding Domain of Na_v1.6 Is Required for Stable Expression at the Axon Initial Segment. Journal of Neuroscience, 2019, 39, 4238-4251.	3.6	24
56	Single amino acid deletion in transmembrane segment D4S6 of sodium channel Scn8a (Nav1.6) in a mouse mutant with a chronic movement disorder. Neurobiology of Disease, 2016, 89, 36-45.	4.4	23
57	PIKfyve complex regulates early melanosome homeostasis required for physiological amyloid formation. Journal of Cell Science, 2019, 132, .	2.0	22
58	CRISPR knockout screen implicates three genes in lysosome function. Scientific Reports, 2019, 9, 9609.	3.3	21
59	Mouse Chromosome 3. Mammalian Genome, 1992, 3, S44-S54.	2.2	20
60	Mutations of Voltage-gated Sodium Channels in Movement Disorders and Epilepsy. Novartis Foundation Symposium, 2008, , 72-86.	1.1	20
61	Biallelic inherited SCN8A variants, a rare cause of SCN8A â€related developmental and epileptic encephalopathy. Epilepsia, 2019, 60, 2277-2285.	5.1	18
62	Cerebral hypomyelination associated with biallelic variants of FIG4. Human Mutation, 2019, 40, 619-630.	2.5	18
63	Whole exome sequencing identifies three recessive FIG4 mutations in an apparently dominant pedigree with Charcotâ€“Marieâ€“Tooth disease. Neuromuscular Disorders, 2014, 24, 666-670.	0.6	17
64	Evaluation of SCN8A as a candidate gene for autosomal dominant essential tremor. Parkinsonism and Related Disorders, 2009, 15, 321-323.	2.2	16
65	Reduced Nav1.6 Sodium Channel Activity in Mice Increases In Vivo Sensitivity to Volatile Anesthetics. PLoS ONE, 2015, 10, e0134960.	2.5	15
66	Gabra2 is a genetic modifier of Scn8a encephalopathy in the mouse*. Epilepsia, 2020, 61, 2847-2856.	5.1	15
67	Rescue of neurodegeneration in theFig4null mouse by a catalytically inactive FIG4 transgene. Human Molecular Genetics, 2016, 25, 340-347.	2.9	14
68	Protective role of the lipid phosphatase Fig4 in the adult nervous system. Human Molecular Genetics, 2018, 27, 2443-2453.	2.9	13
69	INTERSTRAIN VARIATION IN AMYLASE GENE COPY NUMBER AND mRNA ABUNDANCE IN THREE MOUSE TISSUES. Genetics, 1986, 113, 713-722.	2.9	13
70	Mutation watch: Mouse brachyury (T), the T-box gene family, and human disease. Mammalian Genome, 1997, 8, 799-800.	2.2	12
71	SCN8A mutation in a child presenting with seizures and developmental delays. Journal of Physical Education and Sports Management, 2016, 2, a001073.	1.2	12
72	Severe bone loss and multiple fractures in SCN8A-related epileptic encephalopathy. Bone, 2017, 103, 136-143.	2.9	11

#	Article	IF	Citations
73	DQX1, an RNA-dependent ATPase homolog with a novel DEAQ box: expression pattern and genomic sequence comparison of the human and mouse genes. Mammalian Genome, 2001, 12, 456-461.	2.2	9
74	Mutations of voltage-gated sodium channels in movement disorders and epilepsy. Novartis Foundation Symposium, 2002, 241, 72-81; discussion 82-6, 226-32.	1.1	9
75	A Targeted Deleterious Allele of the Splicing Factor SCNM1 in the Mouse. Genetics, 2008, 180, 1419-1427.	2.9	8
76	Screening for novel hexanucleotide repeat expansions at ALS- and FTD-associated loci. Neurology: Genetics, 2016, 2, e71.	1.9	6
77	Identification of genes within the Krd deletion on mouse Chromosome 19. Mammalian Genome, 1999, 10, 399-401.	2.2	5
78	Spontaneous seizures and elevated seizure susceptibility in response to somatic mutation of sodium channel Scn8a in the mouse. Human Molecular Genetics, 2021, 30, 902-907.	2.9	4
79	Gene interactions and modifiers in epilepsy. Epilepsia, 2010, 51, 66-66.	5.1	3
80	Social Deficits and Cerebellar Degeneration in Purkinje Cell Scn8a Knockout Mice. Frontiers in Molecular Neuroscience, 2022, 15, 822129.	2.9	2
81	Sudden Cardiac Death in a Severe Form of Childhood Epilepsy: Mice & Men. FASEB Journal, 2013, 27, 706.4.	0.5	0
82	Correction of the hypomorphic Gabra2 splice site variant in mouse strain C57BL/6J modifies the severity of Scn8a encephalopathy. Human Genetics and Genomics Advances, 2022, 3, 100064.	1.7	0