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	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
2	Social Deficits and Cerebellar Degeneration in Purkinje Cell Scn8a Knockout Mice. Frontiers in Molecular Neuroscience, 2022, 15, 822129.	2.9	2
3	Antisense Oligonucleotide Therapy for Neurodevelopmental Disorders. Developmental Neuroscience, 2021, 43, 247-252.	2.0	34
4	Sodium channelopathies in neurodevelopmental disorders. Nature Reviews Neuroscience, 2021, 22, 152-166.	10.2	79
5	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
6	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
7	Gabra2 is a genetic modifier of Scn8a encephalopathy in the mouse*. Epilepsia, 2020, 61, 2847-2856.	5.1	15
8	Scn8a Antisense Oligonucleotide Is Protective in Mouse Models of SCN8A Encephalopathy and Dravet Syndrome. Annals of Neurology, 2020, 87, 339-346.	5.3	87
9	CRISPR knockout screen implicates three genes in lysosome function. Scientific Reports, 2019, 9, 9609.	3.3	21
10	Biallelic inherited SCN8A variants, a rare cause of SCN8A â€related developmental and epileptic encephalopathy. Epilepsia, 2019, 60, 2277-2285.	5.1	18
11	PIKfyve complex regulates early melanosome homeostasis required for physiological amyloid formation. Journal of Cell Science, 2019, 132, .	2.0	22
12	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
13	Cerebral hypomyelination associated with biallelic variants of FIG4. Human Mutation, 2019, 40, 619-630.	2.5	18
14	SCN8A encephalopathy: Mechanisms and models. Epilepsia, 2019, 60, S86-S91.	5.1	32
15	Prominent role of forebrain excitatory neurons inSCN8Aencephalopathy. Brain, 2019, 142, 362-375.	7.6	69
16	Protective role of the lipid phosphatase Fig4 in the adult nervous system. Human Molecular Genetics, 2018, 27, 2443-2453.	2.9	13
17	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
18	Partial loss-of-function of sodium channel SCN8A in familial isolated myoclonus. Human Mutation, 2018, 39, 965-969.	2.5	34

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19	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
20	Loss-of-function variants of SCN8A in intellectual disability without seizures. Neurology: Genetics, 2017, 3, e170.	1.9	66
21	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
22	Severe bone loss and multiple fractures in SCN8A-related epileptic encephalopathy. Bone, 2017, 103, 136-143.	2.9	11
23	Altered gene expression profile in a mouse model of SCN8A encephalopathy. Experimental Neurology, 2017, 288, 134-141.	4.1	27
24	PI(3,5)P2 biosynthesis regulates oligodendrocyte differentiation by intrinsic and extrinsic mechanisms. ELife, 2016, 5, .	6.0	25
25	SCN8A encephalopathy: Research progress and prospects. Epilepsia, 2016, 57, 1027-1035.	5.1	101
26	Screening for novel hexanucleotide repeat expansions at ALS- and FTD-associated loci. Neurology: Genetics, 2016, 2, e71.	1.9	6
27	SCN8A mutation in a child presenting with seizures and developmental delays. Journal of Physical Education and Sports Management, 2016, 2, a001073.	1.2	12
28	The SCN8A encephalopathy mutation p.Ile1327Val displays elevated sensitivity to the anticonvulsant phenytoin. Epilepsia, 2016, 57, 1458-1466.	5.1	41
29	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
30	Biallelic Mutations of VAC14 in Pediatric-Onset Neurological Disease. American Journal of Human Genetics, 2016, 99, 188-194.	6.2	45
31	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
32	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
33	Rescue of neurodegeneration in theFig4null mouse by a catalytically inactive FIG4 transgene. Human Molecular Genetics, 2016, 25, 340-347.	2.9	14
34	Recurrent and Non-Recurrent Mutations of SCN8A in Epileptic Encephalopathy. Frontiers in Neurology, 2015, 6, 104.	2.4	99
35	Reduced Nav1.6 Sodium Channel Activity in Mice Increases In Vivo Sensitivity to Volatile Anesthetics. PLoS ONE, 2015, 10, e0134960.	2.5	15
36	Convulsive seizures and SUDEP in a mouse model of SCN8A epileptic encephalopathy. Human Molecular Genetics, 2015, 24, 506-515.	2.9	124

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37	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
38	Loss of Fig4 in both Schwann cells and motor neurons contributes to CMT4J neuropathy. Human Molecular Genetics, 2015, 24, 383-396.	2.9	39
39	Incidence of Dravet Syndrome in a US Population. Pediatrics, 2015, 136, e1310-e1315.	2.1	178
40	Mouse Models of PI(3,5)P2 Deficiency with Impaired Lysosome Function. Methods in Enzymology, 2014, 534, 245-260.	1.0	25
41	Characterization of a de novo SCN8A mutation in a patient with epileptic encephalopathy. Epilepsy Research, 2014, 108, 1511-1518.	1.6	92
42	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
43	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
44	C9<scp>ORF</scp>72 expansion in a family with bipolar disorder. Bipolar Disorders, 2013, 15, 326-332.	1.9	58
45	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
46	Sudden Cardiac Death in a Severe Form of Childhood Epilepsy: Mice & Men. FASEB Journal, 2013, 27, 706.4.	0.5	0
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	The splicing regulator Rbfox2 is required for both cerebellar development and mature motor function. Genes and Development, 2012, 26, 445-460.	5.9	186
49	Rbfox proteins regulate alternative splicing of neuronal sodium channel SCN8A. Molecular and Cellular Neurosciences, 2012, 49, 120-126.	2.2	43
50	Guidelines for the use and interpretation of assays for monitoring autophagy. Autophagy, 2012, 8, 445-544.	9.1	3,122
51	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
52	Gene interactions and modifiers in epilepsy. Epilepsia, 2010, 51, 66-66.	5.1	3
53	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
54	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

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55	Sodium channel gene family: epilepsy mutations, gene interactions and modifier effects. Journal of Physiology, 2010, 588, 1841-1848.	2.9	183
56	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
57	Evaluation of SCN8A as a candidate gene for autosomal dominant essential tremor. Parkinsonism and Related Disorders, 2009, 15, 321-323.	2.2	16
58	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
59	A Targeted Deleterious Allele of the Splicing Factor SCNM1 in the Mouse. Genetics, 2008, 180, 1419-1427.	2.9	8
60	Mutations of Voltage-gated Sodium Channels in Movement Disorders and Epilepsy. Novartis Foundation Symposium, 2008, , 72-86.	1.1	20
61	Evidence for a direct role of the disease modifier SCNM1 in splicing. Human Molecular Genetics, 2007, 16, 2506-2516.	2.9	41
62	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
63	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
64	Sodium channel mutations in epilepsy and other neurological disorders. Journal of Clinical Investigation, 2005, 115, 2010-2017.	8.2	427
65	Allelic mutations of the sodium channel SCN8A reveal multiple cellular and physiological functions. Genetica, 2004, 122, 37-45.	1.1	60
66	SCNM1, a Putative RNA Splicing Factor That Modifies Disease Severity in Mice. Science, 2003, 301, 967-969.	12.6	122
67	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
68	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
69	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
70	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
71	Sodium Channels and Neurological Disease: Insights from Scn8a Mutations in the Mouse. Neuroscientist, 2001, 7, 136-145.	3.5	58
72	Mutations of SCN1A, encoding a neuronal sodium channel, in two families with GEFS+2. Nature Genetics, 2000, 24, 343-345.	21.4	910

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73	Identification of genes within the Krd deletion on mouse Chromosome 19. Mammalian Genome, 1999, 10, 399-401.	2.2	5
74	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
75	Functional Analysis of the Mouse Scn8a Sodium Channel. Journal of Neuroscience, 1998, 18, 6093-6102.	3.6	227
76	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
77	Ion Channel Mutations in Mouse Models of Inherited Neurological Disease. Annals of Medicine, 1997, 29, 569-574.	3.8	31
78	Altered Subthreshold Sodium Currents and Disrupted Firing Patterns in Purkinje Neurons of Scn8a Mutant Mice. Neuron, 1997, 19, 881-891.	8.1	367
79	Mutation watch: Mouse brachyury (T), the T-box gene family, and human disease. Mammalian Genome, 1997, 8, 799-800.	2.2	12
80	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
81	Mouse Chromosome 3. Mammalian Genome, 1992, 3, S44-S54.	2.2	20
82	INTERSTRAIN VARIATION IN AMYLASE GENE COPY NUMBER AND mRNA ABUNDANCE IN THREE MOUSE TISSUES. Genetics, 1986, 113, 713-722.	2.9	13