Joanna C Jen

List of Publications by Year
in descending order

35 papers	2,700 citations	331670 21 h-index	395702 33 g-index
35 all docs	35 docs citations	35 times ranked	3618 citing authors

#	Article	IF	Citations
1	Novel de novo TREX1 mutation in a patient with retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations mimicking demyelinating disease. Multiple Sclerosis and Related Disorders, 2021, 52, 103015.	2.0	4
2	Lesion evolution and neurodegeneration in RVCL-S. Neurology, 2020, 95, e1918-e1931.	1.1	13
3	CANVAS with cerebellar/sensory/vestibular dysfunction from RFC1 intronic pentanucleotide expansion. Brain, 2020, 143, 386-390.	7.6	5
4	A novel PUS7 mutation causes intellectual disability with autistic and aggressive behaviors. Neurology: Genetics, 2019, 5, e356.	1.9	18
5	<scp>TREX</scp>1 is expressed by microglia in normal human brain and increases in regions affected by ischemia. Brain Pathology, 2018, 28, 806-821.	4.1	15
6	Episodic ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 155, 205-215.	1.8	46
7	Episodic ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 148, 521-529.	1.8	9
8	Rapid degradation of mutant SLC25A46 by the ubiquitin-proteasome system results in MFN1/2-mediated hyperfusion of mitochondria. Molecular Biology of the Cell, 2017, 28, 600-612.	2.1	61
9	Vestibular paroxysmia: Diagnostic criteria. Journal of Vestibular Research: Equilibrium and Orientation, 2017, 26, 409-415.	2.0	149
10	The RNA Exosome Syncs IAV-RNAPII Transcription to Promote Viral Ribogenesis and Infectivity. Cell, 2017, 169, 679-692.e14.	28.9	48
11	Impaired K+ binding to glial glutamate transporter EAAT1 in migraine. Scientific Reports, 2017, 7, 13913.	3.3	50
12	Bilateral vestibulopathy: Diagnostic criteria Consensus document of the Classification Committee of the BÃ¡rÃ¡ny Society1. Journal of Vestibular Research: Equilibrium and Orientation, 2017, 27, 177-189.	2.0	364
13	Late-onset episodic ataxia associated with SLC1A3 mutation. Journal of Human Genetics, 2017, 62, 443-446.	2.3	41
14	Mutations of EXOSC3/Rrp40p associated with neurological diseases impact ribosomal RNA processing functions of the exosome in S. cerevisiae. Rna, 2017, 23, 466-472.	3.5	33
15	Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations. Brain, 2016, 139, 2909-2922.	7.6	114
16	Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. Brain, 2016, 139, 2877-2890.	7.6	74
17	Rare neurological channelopathies â€" networks to study patients, pathogenesis and treatment. Nature Reviews Neurology, 2016, 12, 195-203.	10.1	8
18	Mutations in XPR1 cause primary familial brain calcification associated with altered phosphate export. Nature Genetics, 2015, 47, 579-581.	21.4	237

#	Article	IF	Citations
19	Opsoclonus: Clinical and immunological features. Journal of the Neurological Sciences, 2012, 320, 61-65.	0.6	24
20	Genetics of Vestibulopathies. Advances in Oto-Rhino-Laryngology, 2011, 70, 130-134.	1.6	7
21	Episodic Ataxia Associated With EAAT1 Mutation C186S Affecting Glutamate Reuptake. Archives of Neurology, 2009, 66, 97-101.	4.5	122
22	Hereditary Episodic Ataxias. Annals of the New York Academy of Sciences, 2008, 1142, 250-253.	3.8	61
23	Effects of failure of development of crossing brainstem pathways on ocular motor control. Progress in Brain Research, 2008, 171, 137-141.	1.4	7
24	Recent advances in the genetics of recurrent vertigo and vestibulopathy. Current Opinion in Neurology, 2008, 21, 3-7.	3.6	23
25	C-terminal truncations in human 3â€²-5â€² DNA exonuclease TREX1 cause autosomal dominant retinal vasculopathy with cerebral leukodystrophy. Nature Genetics, 2007, 39, 1068-1070.	21.4	366
26	Dejerineâ€“Sottas syndrome and vestibular loss due to a point mutation in the PMP22 gene. Journal of the Neurological Sciences, 2005, 237, 21-24.	0.6	21
27	Mutations in a Human ROBO Gene Disrupt Hindbrain Axon Pathway Crossing and Morphogenesis. Science, 2004, 304, 1509-1513.	12.6	361
28	No Mutations in CACNA1A and ATP1A2 in Probands With Common Types of Migraine. Archives of Neurology, 2004, 61, 926.	4.5	69
29	Familial horizontal gaze palsy with progressive scoliosis maps to chromosome 11q23-25. Neurology, 2002, 59, 432-435.	1.1	62
30	Genetics of episodic ataxia. Advances in Neurology, 2002, 89, 459-61.	0.8	11
31	Familial benign recurrent vertigo. American Journal of Medical Genetics Part A, 2001, 100, 287-291.	2.4	107
32	Familial episodic ataxias and related ion channel disorders. Current Treatment Options in Neurology, 2000, 2, 429-431.	1.8	19
33	De novo mutation in CACNA1A caused acetazolamide-responsive episodic ataxia. , 1998, 77, 298-301.		73
34	Familial migraine with vertigo: No mutations found in CACNA1A. American Journal of Medical Genetics Part A, 1998, 79, 148-151.	2.4	78
35	Episodic Ataxia Type 1: Natural History and Effect on Quality of Life. Cerebellum, 0, , .	2.5	0