# Joanna C Jen

List of Publications by Year
in descending order



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|---|---|---|---|
| 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 | 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 |
| 2 | 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 |
| 3 | Mutations in a Human ROBO Gene Disrupt Hindbrain Axon Pathway Crossing and Morphogenesis. Science, 2004, 304, 1509-1513. | 12.6 | 361 |
| 4 | Mutations in XPR1 cause primary familial brain calcification associated with altered phosphate export. Nature Genetics, 2015, 47, 579-581. | 21.4 | 237 |
| 5 | Vestibular paroxysmia: Diagnostic criteria. Journal of Vestibular Research: Equilibrium and Orientation, 2017, 26, 409-415. | 2.0 | 149 |
| 6 | Episodic Ataxia Associated With EAAT1 Mutation C186S Affecting Glutamate Reuptake. Archives of Neurology, 2009, 66, 97-101. | 4.5 | 122 |
| 7 | Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations. Brain, 2016, 139, 2909-2922. | 7.6 | 114 |
| 8 | Familial benign recurrent vertigo. American Journal of Medical Genetics Part A, 2001, 100, 287-291. | 2.4 | 107 |
| 9 | Familial migraine with vertigo: No mutations found in CACNA1A. American Journal of Medical Genetics Part A, 1998, 79, 148-151. | 2.4 | 78 |
| 10 | Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. Brain, 2016, 139, 2877-2890. | 7.6 | 74 |
| 11 | De novo mutation in CACNA1A caused acetazolamide-responsive episodic ataxia. , 1998, 77, 298-301. | | 73 |
| 12 | No Mutations in CACNA1A and ATP1A2 in Probands With Common Types of Migraine. Archives of Neurology, 2004, 61, 926. | 4.5 | 69 |
| 13 | Familial horizontal gaze palsy with progressive scoliosis maps to chromosome 11q23-25. Neurology, 2002, 59, 432-435. | 1.1 | 62 |
| 14 | Hereditary Episodic Ataxias. Annals of the New York Academy of Sciences, 2008, 1142, 250-253. | 3.8 | 61 |
| 15 | 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 |
| 16 | Impaired K+ binding to glial glutamate transporter EAAT1 in migraine. Scientific Reports, 2017, 7, 13913. | 3.3 | 50 |
| 17 | The RNA Exosome Syncs IAV-RNAPII Transcription to Promote Viral Ribogenesis and Infectivity. Cell, 2017, 169, 679-692.e14. | 28.9 | 48 |
| 18 | Episodic ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 155, 205-215. | 1.8 | 46 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Late-onset episodic ataxia associated with SLC1A3 mutation. Journal of Human Genetics, 2017, 62, 443-446. | 2.3 | 41 |
| 20 | 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 |
| 21 | Opsoclonus: Clinical and immunological features. Journal of the Neurological Sciences, 2012, 320, 61-65. | 0.6 | 24 |
| 22 | Recent advances in the genetics of recurrent vertigo and vestibulopathy. Current Opinion in Neurology, 2008, 21, 3-7. | 3.6 | 23 |
| 23 | 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 |
| 24 | Familial episodic ataxias and related ion channel disorders. Current Treatment Options in Neurology, 2000, 2, 429-431. | 1.8 | 19 |
| 25 | A novel PUS7 mutation causes intellectual disability with autistic and aggressive behaviors. Neurology: Genetics, 2019, 5, e356. | 1.9 | 18 |
| 26 | <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 |
| 27 | Lesion evolution and neurodegeneration in RVCL-S. Neurology, 2020, 95, e1918-e1931. | 1.1 | 13 |
| 28 | Genetics of episodic ataxia. Advances in Neurology, 2002, 89, 459-61. | 0.8 | 11 |
| 29 | Episodic ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 148, 521-529. | 1.8 | 9 |
| 30 | Rare neurological channelopathies â€" networks to study patients, pathogenesis and treatment. Nature Reviews Neurology, 2016, 12, 195-203. | 10.1 | 8 |
| 31 | Effects of failure of development of crossing brainstem pathways on ocular motor control. Progress in Brain Research, 2008, 171, 137-141. | 1.4 | 7 |
| 32 | Genetics of Vestibulopathies. Advances in Oto-Rhino-Laryngology, 2011, 70, 130-134. | 1.6 | 7 |
| 33 | CANVAS with cerebellar/sensory/vestibular dysfunction from RFC1 intronic pentanucleotide expansion. Brain, 2020, 143, 386-390. | 7.6 | 5 |
| 34 | 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 |
| 35 | Episodic Ataxia Type 1: Natural History and Effect on Quality of Life. Cerebellum, 0, , . | 2.5 | 0 |