Akiko Yoshimura

List of Publications by Year
in descending order

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23 papers	773 citations	687220 13 h-index	752573 20 g-index
24 all docs	24 docs citations	24 times ranked	976 citing authors

#	Article	IF	Citations
1	An MFN2-related Charcot-Marie-Tooth Disease Patient with Optic Nerve Atrophy, Neurogenic Bladder Dysfunction, and Diaphragmatic Weakness. Internal Medicine, 2022, , .	0.3	0
2	Elderly patients with suspected Charcot-Marie-Tooth disease should be tested for the TTR gene for effective treatments. Journal of Human Genetics, 2022, , .	1.1	4
3	An NEFH founder mutation causes broad phenotypic spectrum in multiple Japanese families. Journal of Human Genetics, 2022, 67, 399-403.	1.1	5
4	A case of adult-onset Wolfram syndrome with compound heterozygous mutations of the WFS1 gene. American Journal of Ophthalmology Case Reports, 2022, 25, 101315.	0.4	0
5	Genetic spectrum of <scp>Charcotâ€“Marieâ€“Tooth</scp> disease associated with myelin protein zero gene variants in Japan. Clinical Genetics, 2021, 99, 359-375.	1.0	18
6	Benzalkonium Chloride Resistance in Staphylococcus epidermidis on the Ocular Surface of Glaucoma Patients Under Long-Term Administration of Eye Drops. Translational Vision Science and Technology, 2020, 9, 9.	1.1	10
7	Genetic profile and onset features of 1005 patients with Charcot-Marie-Tooth disease in Japan. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 195-202.	0.9	61
8	Clinical and genetic features of Charcotâ€Marieâ€Tooth disease 2F and hereditary motor neuropathy 2B in Japan. Journal of the Peripheral Nervous System, 2018, 23, 40-48.	1.4	17
9	Clinical and mutational spectrum of Japanese patients with recessive variants in SH3TC2. Journal of Human Genetics, 2018, 63, 281-287.	1.1	7
10	Genetic and phenotypic profile of 112 patients with Xâ€linked Charcotâ€“Marieâ€“Tooth disease type 1. European Journal of Neurology, 2018, 25, 1454-1461.	1.7	12
11	Clinical diversity caused by novel IGHMBP2 variants. Journal of Human Genetics, 2017, 62, 599-604.	1.1	18
12	Clinical and mutational spectrum of Japanese patients with Charcotâ€Marieâ€Tooth disease caused by <scp>GDAP1</scp> variants. Clinical Genetics, 2017, 92, 274-280.	1.0	15
13	<scp>WNK1</scp>/<scp>HSN2</scp> founder mutation in patients with hereditary sensory and autonomic neuropathy: A Japanese cohort study. Clinical Genetics, 2017, 92, 659-663.	1.0	8
14	Clinical and mutational spectrum of Charcotâ€“Marieâ€“Tooth disease type 2Z caused by MORC2 variants in Japan. European Journal of Neurology, 2017, 24, 1274-1282.	1.7	32
15	Clinical and genetic diversities of Charcotâ€Marieâ€Tooth disease with MFN2 mutations in a large case study. Journal of the Peripheral Nervous System, 2017, 22, 191-199.	1.4	31
16	Mutations in MME cause an autosomalâ€recessive Charcotâ€“Marieâ€“Tooth disease type 2. Annals of Neurology, 2016, 79, 659-672.	2.8	82
17	Neurofilament light mutation causes hereditary motor and sensory neuropathy with pyramidal signs. Journal of the Peripheral Nervous System, 2014, 19, 311-316.	1.4	25
18	Hereditary sensory and autonomic neuropathy type IID caused by an SCN9A mutation. Neurology, 2013, 80, 1641-1649.	1.5	59

#	Article	IF	Citations
19	Novel mutations identified in patients with a mild phenotype of Ullrich congenital muscular dystrophy through targeted next-generation sequencing. Neurology and Clinical Neuroscience, 2013, 1, 148-153.	0.2	0
20	A novel cytokine-inducible gene CIS encodes an SH2-containing protein that binds to tyrosine-phosphorylated interleukin 3 and erythropoietin receptors. EMBO Journal, 1995, 14, 2816-26.	3.5	199
21	Novel screening method for agents that overcome classical multidrug resistance in a human cell line. Cancer Letters, 1990, 50, 45-51.	3.2	29
22	Biosynthesis, processing and half-life of P-glycoprotein in a human multidrug-resistant KB cell. Biochimica Et Biophysica Acta - General Subjects, 1989, 992, 307-314.	1.1	42
23	Cytoplasmic orientation and two-domain structure of the multidrug transporter, P-glycoprotein, demonstrated with sequence-specific antibodies. Journal of Biological Chemistry, 1989, 264, 16282-91.	1.6	99