Kishin Koh

List of Publications by Year
in descending order

50 papers	679 citations	759233 12 h-index	642732 23 g-index
53 all docs	53 docs citations	53 times ranked	1332 citing authors

#	Article	IF	Citations
1	ChÃ©diakâ€“Higashi syndrome presenting as a hereditary spastic paraplegia. Journal of Human Genetics, 2022, 67, 119-121.	2.3	2
2	A clinical and genetic study of SPG31 in Japan. Journal of Human Genetics, 2022, , .	2.3	1
3	Ageâ€related changes in blood pressure and heart rates of patients with Parkinson's disease. Journal of Clinical Hypertension, 2021, 23, 175-178.	2.0	7
4	Spastic Paraplegia with Paget's Disease of Bone due to a VCP Gene Mutation. Internal Medicine, 2021, 60, 141-144.	0.7	4
5	SPG9A with the new occurrence of an ALDH18A1 mutation in a CMT1A family with PMP22 duplication: case report. BMC Neurology, 2021, 21, 64.	1.8	2
6	A Nepalese family with an REEP2 mutation: clinical and genetic study. Journal of Human Genetics, 2021, 66, 749-752.	2.3	1
7	Sympathetic nerve outflow to skin in a case with dentatorubral-pallidoluysian atrophy. Journal of Clinical Neuroscience, 2021, 87, 80-83.	1.5	2
8	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
9	Paraneoplastic sensorimotor neuropathy associated with mediastinal germ cell tumor: favorable outcome after high-dose intravenous immunoglobulin therapy. Neurological Sciences, 2020, 41, 723-725.	1.9	2
10	VPS13Dâ€related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. Molecular Genetics & Genomic Medicine, 2020, 8, e1108.	1.2	29
11	Identification of a novel mutation in ATP13A2 associated with a complicated form of hereditary spastic paraplegia. Neurology: Genetics, 2020, 6, e514.	1.9	15
12	A case of lateâ€onset Chediakâ€Higashi syndrome with progressive gait disturbance and cognitive dysfunction caused by novel variant in LYST gene. Neurology and Clinical Neuroscience, 2020, 8, 415-418.	0.4	2
13	RFC1 repeat expansion in Japanese patients with late-onset cerebellar ataxia. Journal of Human Genetics, 2020, 65, 1143-1147.	2.3	32
14	A Japanese SPG4 Patient with a Confirmed De Novo Mutation of the SPAST Gene. Internal Medicine, 2020, 59, 2311-2315.	0.7	1
15	Variants in saposin D domain of prosaposin gene linked to Parkinsonâ€™s disease. Brain, 2020, 143, 1190-1205.	7.6	72
16	A novel mutation in the GBA2 gene in a Japanese patient with SPG46: A case report. ENeurologicalSci, 2020, 19, 100238.	1.3	6
17	Sympathetic outflow to skin predicts central autonomic dysfunction in multiple system atrophy. Neurological Sciences, 2020, 41, 2241-2248.	1.9	5
18	Conjugal multiple system atrophy: Computing chance or investigating real patients?. Parkinsonism and Related Disorders, 2020, 75, 122-123.	2.2	1

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19	Conjugal multiple system atrophy: Be cautious when calculating numbers of probability. Parkinsonism and Related Disorders, 2020, 77, 178-179.	2.2	0
20	Clinical and Genetic Study of the First Japanese FTDP-17 Patient with a Mutation of +3 in Intron 10 in the <i>MAPT</i> Gene. Internal Medicine, 2019, 58, 2397-2400.	0.7	2
21	Conjugal cerebellar type of multiple system atrophy: Person-to-person transmission?. Parkinsonism and Related Disorders, 2019, 69, 68-70.	2.2	5
22	UBAP1 mutations cause juvenile-onset hereditary spastic paraplegias (SPG80) and impair UBAP1 targeting to endosomes. Journal of Human Genetics, 2019, 64, 1055-1065.	2.3	15
23	Non-convulsive status epilepticus associated with neuronal intranuclear inclusion disease: A case report and literature review. Epilepsy & Behavior Case Reports, 2019, 11, 103-106.	1.5	10
24	Pathological findings in a patient with non-dystrophic myotonia with a mutation of the SCN4A gene; a case report. BMC Neurology, 2019, 19, 125.	1.8	3
25	A Japanese family with a novel nonsense mutation in the spastin gene associated with both cerebellar ataxia and cognitive impairment. Journal of the Neurological Sciences, 2019, 397, 114-116.	0.6	1
26	Neuroimaging, genetic, and enzymatic study in a Japanese family with a GBA gross deletion. Parkinsonism and Related Disorders, 2019, 61, 57-63.	2.2	6
27	Novel GARS mutation presenting as autosomal dominant intermediate Charcotâ€Marieâ€Tooth disease. Journal of the Peripheral Nervous System, 2019, 24, 156-160.	3.1	8
28	A novel homozygous mutation of the TFG gene in a patient with early onset spastic paraplegia and later onset sensorimotor polyneuropathy. Journal of Human Genetics, 2019, 64, 171-176.	2.3	4
29	Exome Sequencing Reveals a Novel Homozygous Frameshift Mutation in the <i>CYP7B1</i> Gene in a Japanese Patient with SPG5. Internal Medicine, 2019, 58, 719-722.	0.7	2
30	Decreasing 123I-ioflupane SPECT accumulation and 123I-MIBG myocardial scintigraphy uptake in a patient with a novel homozygous mutation in the ZFYVE26 gene. Neurological Sciences, 2019, 40, 429-431.	1.9	2
31	PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 55-59.	2.3	17
32	ExpansionsÂofÂintronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. Nature Genetics, 2018, 50, 581-590.	21.4	238
33	Sympathetic neurograms showing characteristics of both muscle and skin sympathetic nerve activity in a case with pure autonomic failure. Clinical Autonomic Research, 2018, 28, 347-349.	2.5	0
34	Novel <i>SPG11</i> Mutations in a Patient with Symptoms Mimicking Multiple Sclerosis. Internal Medicine, 2018, 57, 3183-3186.	0.7	7
35	Novel SLC20A2 mutation in primary familial brain calcification with disturbance of sustained phonation and orofacial apraxia. Journal of the Neurological Sciences, 2018, 390, 1-3.	0.6	2
36	JASPAC: Japan Spastic Paraplegia Research Consortium. Brain Sciences, 2018, 8, 153.	2.3	41

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37	Novel mutations in the ALDH18A1 gene in complicated hereditary spastic paraplegia with cerebellar ataxia and cognitive impairment. Journal of Human Genetics, 2018, 63, 1009-1013.	2.3	18
38	Vasomotor regulation in patients with multiple system atrophy. Journal of Neural Transmission, 2017, 124, 477-481.	2.8	4
39	Pre- and postganglionic vasomotor dysfunction causes distal limb coldness in multiple system atrophy. Journal of the Neurological Sciences, 2017, 380, 191-195.	0.6	3
40	Co-existence of spastic paraplegia-30 with novel KIF1A mutation and spinocerebellar ataxia 31 with intronic expansion of BEAN and TK2 in a family. Journal of the Neurological Sciences, 2017, 372, 128-130.	0.6	9
41	Exome sequencing reveals a novel missense mutation in the KIAA0196 gene in a Japanese patient with SPG8. Clinical Neurology and Neurosurgery, 2016, 144, 36-38.	1.4	11
42	Hot cross bun sign in a late-onset SCA1 patient. Neurological Sciences, 2016, 37, 1873-1874.	1.9	7
43	Wholeâ€exome sequencing reveals a missense mutation in the <scp>KCND</scp>3 gene in a patient with <scp>SCA</scp>19/22. Neurology and Clinical Neuroscience, 2015, 3, 197-199.	0.4	4
44	Japanese amyotrophic lateral sclerosis patient with learning disabilities with a deletion mutation in the Câ€terminal of the <scp>FUS</scp>/<scp>TLS</scp> gene. Neurology and Clinical Neuroscience, 2015, 3, 192-193.	0.4	2
45	No relation between sympathetic outflow to muscles and respiratory function in amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2015, 358, 66-71.	0.6	1
46	Novel mutations in the PNPLA6 gene in Boucher-NeuhÃuser syndrome. Journal of Human Genetics, 2015, 60, 217-220.	2.3	15
47	A Japanese SCA5 family with a novel three-nucleotide in-frame deletion mutation in the SPTBN2 gene: a clinical and genetic study. Journal of Human Genetics, 2014, 59, 569-573.	2.3	20
48	Exome sequencing shows a novel de novo mutation in <scp>ATL</scp>1. Neurology and Clinical Neuroscience, 2014, 2, 1-4.	0.4	2
49	A MOG antibodyâ€related disorder associated with peripheral facial nerve palsy. Clinical and Experimental Neuroimmunology, 0, , .	1.0	0
50	Novel heterozygous variants of <scp> SLC12A6 </scp> in Japanese families with <scp>Charcotâ€“Marieâ€“Tooth</scp> disease. Annals of Clinical and Translational Neurology, 0, , .	3.7	1