# Kishin Koh

List of Publications by Year
in descending order



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|---|---|---|---|
| 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 | ExpansionsÂofÂintronic TTTCA and TTTTA repeats in benign adult familial myoclonic epilepsy. Nature Genetics, 2018, 50, 581-590. | 21.4 | 238 |
| 2 | Variants in saposin D domain of prosaposin gene linked to Parkinsonâ€™s disease. Brain, 2020, 143, 1190-1205. | 7.6 | 72 |
| 3 | JASPAC: Japan Spastic Paraplegia Research Consortium. Brain Sciences, 2018, 8, 153. | 2.3 | 41 |
| 4 | RFC1 repeat expansion in Japanese patients with late-onset cerebellar ataxia. Journal of Human Genetics, 2020, 65, 1143-1147. | 2.3 | 32 |
| 5 | VPS13Dâ€related disorders presenting as a pure and complicated form of hereditary spastic paraplegia. Molecular Genetics & Genomic Medicine, 2020, 8, e1108. | 1.2 | 29 |
| 6 | Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434. | 7.6 | 22 |
| 7 | 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 |
| 8 | 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 |
| 9 | PLA2G6-associated neurodegeneration presenting as a complicated form of hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 55-59. | 2.3 | 17 |
| 10 | Novel mutations in the PNPLA6 gene in Boucher-NeuhÃuser syndrome. Journal of Human Genetics, 2015, 60, 217-220. | 2.3 | 15 |
| 11 | 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 |
| 12 | Identification of a novel mutation in ATP13A2 associated with a complicated form of hereditary spastic paraplegia. Neurology: Genetics, 2020, 6, e514. | 1.9 | 15 |
| 13 | 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 |
| 14 | 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 |
| 15 | 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 |
| 16 | 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 |
| 17 | Hot cross bun sign in a late-onset SCA1 patient. Neurological Sciences, 2016, 37, 1873-1874. | 1.9 | 7 |
| 18 | Novel <i>SPG11</i> Mutations in a Patient with Symptoms Mimicking Multiple Sclerosis. Internal Medicine, 2018, 57, 3183-3186. | 0.7 | 7 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | 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 |
| 20 | 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 |
| 21 | A novel mutation in the GBA2 gene in a Japanese patient with SPG46: A case report. ENeurologicalSci, 2020, 19, 100238. | 1.3 | 6 |
| 22 | Conjugal cerebellar type of multiple system atrophy: Person-to-person transmission?. Parkinsonism and Related Disorders, 2019, 69, 68-70. | 2.2 | 5 |
| 23 | Sympathetic outflow to skin predicts central autonomic dysfunction in multiple system atrophy. Neurological Sciences, 2020, 41, 2241-2248. | 1.9 | 5 |
| 24 | 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 |
| 25 | Vasomotor regulation in patients with multiple system atrophy. Journal of Neural Transmission, 2017, 124, 477-481. | 2.8 | 4 |
| 26 | 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 |
| 27 | Spastic Paraplegia with Paget's Disease of Bone due to a VCP Gene Mutation. Internal Medicine, 2021, 60, 141-144. | 0.7 | 4 |
| 28 | 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 |
| 29 | 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 |
| 30 | Exome sequencing shows a novel de novo mutation in <scp>ATL</scp>1. Neurology and Clinical Neuroscience, 2014, 2, 1-4. | 0.4 | 2 |
| 31 | 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 |
| 32 | 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 |
| 33 | 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 |
| 34 | 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 |
| 35 | 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 |
| 36 | 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 |

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|---|---|---|---|
| 37 | 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 |
| 38 | 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 |
| 39 | Sympathetic nerve outflow to skin in a case with dentatorubral-pallidoluysian atrophy. Journal of Clinical Neuroscience, 2021, 87, 80-83. | 1.5 | 2 |
| 40 | ChÃ©diakâ€“Higashi syndrome presenting as a hereditary spastic paraplegia. Journal of Human Genetics, 2022, 67, 119-121. | 2.3 | 2 |
| 41 | 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 |
| 42 | 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 |
| 43 | A Japanese SPG4 Patient with a Confirmed De Novo Mutation of the SPAST Gene. Internal Medicine, 2020, 59, 2311-2315. | 0.7 | 1 |
| 44 | Conjugal multiple system atrophy: Computing chance or investigating real patients?. Parkinsonism and Related Disorders, 2020, 75, 122-123. | 2.2 | 1 |
| 45 | A Nepalese family with an REEP2 mutation: clinical and genetic study. Journal of Human Genetics, 2021, 66, 749-752. | 2.3 | 1 |
| 46 | A clinical and genetic study of SPG31 in Japan. Journal of Human Genetics, 2022, , . | 2.3 | 1 |
| 47 | 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 |
| 48 | 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 |
| 49 | Conjugal multiple system atrophy: Be cautious when calculating numbers of probability. Parkinsonism and Related Disorders, 2020, 77, 178-179. | 2.2 | 0 |
| 50 | A MOG antibodyâ€related disorder associated with peripheral facial nerve palsy. Clinical and Experimental Neuroimmunology, 0, , . | 1.0 | 0 |