

# Kazumasa Saigoh

## List of Publications by Year in descending order

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Version: 2024-02-01

37  
papers

2,138  
citations

687363

13  
h-index

330143

37  
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42  
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42  
docs citations

42  
times ranked

3308  
citing authors

#	ARTICLE	IF	CITATIONS
1	Functional consequences of a CK1 $\delta$ mutation causing familial advanced sleep phase syndrome. <i>Nature</i> , 2005, 434, 640-644.	27.8	773
2	Intragenic deletion in the gene encoding ubiquitin carboxy-terminal hydrolase in gad mice. <i>Nature Genetics</i> , 1999, 23, 47-51.	21.4	467
3	Lamin B1 duplications cause autosomal dominant leukodystrophy. <i>Nature Genetics</i> , 2006, 38, 1114-1123.	21.4	365
4	A <i>PERIOD3</i> variant causes a circadian phenotype and is associated with a seasonal mood trait. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E1536-44.	7.1	134
5	Mutations in the gene encoding p62 in Japanese patients with amyotrophic lateral sclerosis. <i>Neurology</i> , 2013, 80, 458-463.	1.1	73
6	Oxidation and interaction of DJ-1 with 20S proteasome in the erythrocytes of early stage Parkinson's disease patients. <i>Scientific Reports</i> , 2016, 6, 30793.	3.3	30
7	Chondroitin beta-1,4-N-acetylgalactosaminyltransferase-1 missense mutations are associated with neuropathies. <i>Journal of Human Genetics</i> , 2011, 56, 143-146.	2.3	27
8	VCP gene analyses in Japanese patients with sporadic amyotrophic lateral sclerosis identify a new mutation. <i>Neurobiology of Aging</i> , 2015, 36, 1604.e1-1604.e6.	3.1	27
9	Noncoding repeat expansions for ALS in Japan are associated with the <i>ATXN8OS</i> gene. <i>Neurology: Genetics</i> , 2018, 4, e252.	1.9	19
10	The novel de novo mutation of KIF1A gene as the cause for Spastic paraplegia 30 in a Japanese case. <i>ENeurologicalSci</i> , 2019, 14, 34-37.	1.3	18
11	Clinical practice guideline for chronic headache 2013. <i>Neurology and Clinical Neuroscience</i> , 2019, 7, 231-259.	0.4	17
12	Diagnosis of Parkinson's disease and the level of oxidized DJ-1 protein. <i>Neuroscience Research</i> , 2018, 128, 58-62.	1.9	15
13	A chondroitin synthase-1 (ChSy-1) missense mutation in a patient with neuropathy impairs the elongation of chondroitin sulfate chains initiated by chondroitin N-acetylgalactosaminyltransferase-1. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 2013, 1830, 4806-4812.	2.4	14
14	A novel mutation in the calcium channel gene in a family with hypokalemic periodic paralysis. <i>Journal of the Neurological Sciences</i> , 2011, 309, 9-11.	0.6	13
15	A New Mouse Allele of Glutamate Receptor Delta 2 with Cerebellar Atrophy and Progressive Ataxia. <i>PLoS ONE</i> , 2014, 9, e107867.	2.5	13
16	Successful Combination of Pallidal and Thalamic Stimulation for Intractable Involuntary Movements in Patients with Neuroacanthocytosis. <i>World Neurosurgery</i> , 2015, 84, 1177.e1-1177.e7.	1.3	13
17	A comparative study of curated contents by knowledge-based curation system in cancer clinical sequencing. <i>Scientific Reports</i> , 2019, 9, 11340.	3.3	12
18	Chondroitin sulfate $\beta$ -1,4-N-acetylgalactosaminyltransferase-1 (ChGn-1) polymorphism: Association with progression of multiple sclerosis. <i>Neuroscience Research</i> , 2016, 108, 55-59.	1.9	11

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19	PSP-Phenotype in SCA8: Case Report and Systemic Review. <i>Cerebellum</i> , 2019, 18, 76-84.	2.5	11
20	A Guillain-Barré syndrome-associated SIGLEC10 rare variant impairs its recognition of gangliosides. <i>Journal of Autoimmunity</i> , 2021, 116, 102571.	6.5	10
21	Tau accumulation in two patients with frontotemporal lobe degeneration showing different types of aphasia using 18F-THK-5351 positron emission tomography: a case report. <i>International Psychogeriatrics</i> , 2018, 30, 641-646.	1.0	9
22	Multiple system involvement in a Japanese patient with a V31A mutation in the <i>SOD1</i> gene. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2014, 15, 312-314.	1.7	8
23	The first Japanese familial case of spinocerebellar ataxia 23 with a novel mutation in the PDYN gene. <i>Parkinsonism and Related Disorders</i> , 2015, 21, 332-334.	2.2	8
24	Phenotypic and molecular diversities of spinocerebellar ataxia type 2 in Japan. <i>Journal of Neurology</i> , 2021, 268, 2933-2942.	3.6	7
25	Unexpectedly mild phenotype in an ataxic family with a two-base deletion in the APTX gene. <i>Journal of the Neurological Sciences</i> , 2017, 378, 75-79.	0.6	5
26	Charcot-Marie-Tooth disease with a mutation in FBLN5 accompanying with the small vasculitis and widespread onion-bulb formations. <i>Journal of the Neurological Sciences</i> , 2020, 410, 116623.	0.6	5
27	Effect of expression alteration in flanking genes on phenotypes of <i>St8sia2</i> -deficient mice. <i>Scientific Reports</i> , 2019, 9, 13634.	3.3	4
28	Neutral Lipid Storage Disease Associated with the <i>PNPLA2</i> Gene: Case Report and Literature Review. <i>European Neurology</i> , 2020, 83, 317-322.	1.4	4
29	Circadian expression and specific localization of a sialyltransferase gene in the suprachiasmatic nucleus. <i>Neuroscience Letters</i> , 2013, 535, 12-17.	2.1	3
30	Time Course of Radiological Imaging and Variable Interindividual Symptoms in Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Associated with p.Arg487His Mutation in the <i>VCP</i> Gene. <i>European Neurology</i> , 2017, 78, 78-83.	1.4	3
31	Novel mutation in the <i>SOD1</i> gene in a patient with early-onset, rapidly progressive amyotrophic lateral sclerosis. <i>Neurology and Clinical Neuroscience</i> , 2017, 5, 189-191.	0.4	3
32	Regional gray matter-dedicated SUVR with 3D-MRI detects positive amyloid deposits in equivocal amyloid PET images. <i>Annals of Nuclear Medicine</i> , 2020, 34, 856-863.	2.2	3
33	Ophthalmologic involvement in Japanese siblings with chorea-acanthocytosis caused by a novel chorein mutation. <i>Parkinsonism and Related Disorders</i> , 2013, 19, 913-915.	2.2	2
34	Hemiplegic migraine type 2 with new mutation of the <i>ATP1A2</i> gene in Japanese cases. <i>Neuroscience Research</i> , 2022, , .	1.9	2
35	YAC/BAC-Based Physical and Transcript Mapping around the Gracile Axonal Dystrophy ( <i>gad</i> ) Locus Identifies <i>Uchl1</i> , <i>Pmx2b</i> , <i>Atp3a2</i> , and <i>Hip2</i> Genes. <i>Genomics</i> , 2000, 66, 333-336.	2.9	1
36	Progressive amnesic cognitive impairment in a middle-aged patient with developmental language disorder: a case report. <i>Journal of Medical Case Reports</i> , 2020, 14, 139.	0.8	0

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37	Longitudinal study of primary progressive aphasia in a patient with pathologically diagnosed Alzheimer's disease: a case report. Journal of Medical Case Reports, 2021, 15, 272.	0.8	0