Satoko Miyatake

List of Publications by Year
in descending order

50 papers	1,474 citations	361296 20 h-index	360920 35 g-index
50 all docs	50 docs citations	50 times ranked	3152 citing authors

#	Article	IF	Citations
1	Dominant mutations in ORAI1 cause tubular aggregate myopathy with hypocalcemia via constitutive activation of store-operated Ca2+ channels. Human Molecular Genetics, 2015, 24, 637-648.	1.4	132
2	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. Cell Reports, 2018, 22, 734-747.	2.9	132
3	Tandem-genotypes: robust detection of tandem repeat expansions from long DNA reads. Genome Biology, 2019, 20, 58.	3.8	103
4	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. Scientific Reports, 2016, 6, 30072.	1.6	102
5	GGC Repeat Expansion of NOTCH2NLC in Adult Patients with Leukoencephalopathy. Annals of Neurology, 2019, 86, 962-968.	2.8	98
6	A Novel Mutation inELOVL4Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratodermia. JAMA Neurology, 2015, 72, 797.	4.5	79
7	Biallelic Mutations in MYPN , Encoding Myopalladin, Are Associated with Childhood-Onset, Slowly Progressive Nemaline Myopathy. American Journal of Human Genetics, 2017, 100, 169-178.	2.6	66
8	De novo KIF1A mutations cause intellectual deficit, cerebellar atrophy, lower limb spasticity and visual disturbance. Journal of Human Genetics, 2015, 60, 739-742.	1.1	58
9	A 12-kb structural variation in progressive myoclonic epilepsy was newly identified by long-read whole-genome sequencing. Journal of Human Genetics, 2019, 64, 359-368.	1.1	48
10	A novel mutation in SLC1A3 causes episodic ataxia. Journal of Human Genetics, 2018, 63, 207-211.	1.1	42
11	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36
12	Long-read sequencing identifies the pathogenic nucleotide repeat expansion in RFC1 in a Japanese case of CANVAS. Journal of Human Genetics, 2020, 65, 475-480.	1.1	35
13	Detecting a long insertion variant in SAMD12 by SMRT sequencing: implications of long-read whole-genome sequencing for repeat expansion diseases. Journal of Human Genetics, 2019, 64, 191-197.	1.1	33
14	Milder progressive cerebellar atrophy caused by biallelic SEPSECS mutations. Journal of Human Genetics, 2016, 61, 527-531.	1.1	30
15	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	1.5	30
16	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. Scientific Reports, 2015, 4, 7132.	1.6	29
17	Identification of novel <scp>SNORD118</scp> mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. Clinical Genetics, 2017, 92, 180-187.	1.0	28
18	A novel SLC9A1 mutation causes cerebellar ataxia. Journal of Human Genetics, 2018, 63, 1049-1054.	1.1	28

#	Article	IF	Citations
19	De novo variants in RHOBTB2 , an atypical Rho GTPase gene, cause epileptic encephalopathy. Human Mutation, 2018, 39, 1070-1075.	1.1	25
20	Gain-of-Function MN1 Truncation Variants Cause a Recognizable Syndrome with Craniofacial and Brain Abnormalities. American Journal of Human Genetics, 2020, 106, 13-25.	2.6	25
21	Complete sequencing of expandedSAMD12repeats by long-read sequencing and Cas9-mediated enrichment. Brain, 2021, 144, 1103-1117.	3.7	25
22	Father-to-offspring transmission of extremely long NOTCH2NLC repeat expansions with contractions: genetic and epigenetic profiling with long-read sequencing. Clinical Epigenetics, 2021, 13, 204.	1.8	22
23	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. Journal of Human Genetics, 2018, 63, 263-270.	1.1	19
24	Repeat conformation heterogeneity in cerebellar ataxia, neuropathy, vestibular areflexia syndrome. Brain, 2022, 145, 1139-1150.	3.7	19
25	Whole exome sequencing of fetal structural anomalies detected by ultrasonography. Journal of Human Genetics, 2021, 66, 499-507.	1.1	18
26	Efficient detection of copyâ€•number variations using exome data: Batchâ€•and sexâ€‐based analyses. Human Mutation, 2021, 42, 50-65.	1.1	18
27	Phenotypeâ€“genotype correlations in patients with GNB1 gene variants, including the first three reported Japanese patients to exhibit spastic diplegia, dyskinetic quadriplegia, and infantile spasms. Brain and Development, 2020, 42, 199-204.	0.6	16
28	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. Journal of Human Genetics, 2022, 67, 157-164.	1.1	16
29	Novel EXOSC9 variants cause pontocerebellar hypoplasia type 1D with spinal motor neuronopathy and cerebellar atrophy. Journal of Human Genetics, 2021, 66, 401-407.	1.1	15
30	Monoallelic and bi-allelic variants in NCDN cause neurodevelopmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 739-748.	2.6	15
31	Actin-binding protein filamin-A drives tau aggregation and contributes to progressive supranuclear palsy pathology. Science Advances, 2022, 8, .	4.7	15
32	De novo ATP1A3 variants cause polymicrogyria. Science Advances, 2021, 7, .	4.7	13
33	Hemorrhagic stroke and renovascular hypertension with Grange syndrome arising from a novel pathogenic variant in YY1AP1. Journal of Human Genetics, 2019, 64, 885-890.	1.1	11
34	Leaky splicing variant in sepiapterin reductase deficiency. Neurology: Genetics, 2019, 5, e319.	0.9	10
35	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	0.7	10
36	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. Journal of Human Genetics, 2017, 62, 997-1000.	1.1	9

#	Article	IF	Citations
37	A novel homozygous mutation of CLCN2 in a patient with characteristic brain MRI images â€“ A first case of CLCN2-related leukoencephalopathy in Japan. Brain and Development, 2019, 41, 101-105.	0.6	9
38	â€˜Cortical cerebellar atrophyâ€™ dwindles away in the era of next-generation sequencing. Journal of Human Genetics, 2014, 59, 589-590.	1.1	8
39	Predominant cerebellar phenotype in spastic paraplegia 7 (SPG7). Human Genome Variation, 2015, 2, 15012.	0.4	7
40	Biallelic null variants in ZNF142 cause global developmental delay with familial epilepsy and dysmorphic features. Journal of Human Genetics, 2022, 67, 169-173.	1.1	7
41	Polymicrogyria in a child with KCNMA1-related channelopathy. Brain and Development, 2022, 44, 173-177.	0.6	7
42	A novel DARS2 mutation in a Japanese patient with leukoencephalopathy with brainstem and spinal cord involvement but no lactate elevation. Human Genome Variation, 2017, 4, 17051.	0.4	6
43	Expanding the <scp>KIF4A</scp>â€associated phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3728-3739.	0.7	6
44	De novo heterozygous variants in KIF5B cause kyphomelic dysplasia. Clinical Genetics, 2022, 102, 3-11.	1.0	5
45	Novel CLTC variants cause new brain and kidney phenotypes. Journal of Human Genetics, 2021, , .	1.1	4
46	Reply to â€œ<scp>GGC</scp> Repeat Expansion of <scp>NOTCH2NLC</scp> is Rare in European Leukoencephalopathyâ€• Annals of Neurology, 2020, 88, 642-643.	2.8	2
47	Cerebrovascular diseases in two patients with entire NSD1 deletion. Human Genome Variation, 2021, 8, 20.	0.4	2
48	A 23-year follow-up report of juvenile-onset Sandhoff disease presenting with a motor neuron disease phenotype and a novel variant. Brain and Development, 2021, 43, 1029-1032.	0.6	1
49	A 2â€yearâ€old patient with a diffuse intrinsic pontine glioma and radiationâ€induced moyamoya syndrome. Pediatric Blood and Cancer, 2020, 67, e28618.	0.8	0
50	A case of epilepsy of infancy with migrating focal seizures caused by mosaic <i>SCN2A</i> mutation. Epilepsy and Seizure, 2022, 14, 17-24.	0.1	0