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	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
2	Repeat conformation heterogeneity in cerebellar ataxia, neuropathy, vestibular areflexia syndrome. Brain, 2022, 145, 1139-1150.	3.7	19
3	Polymicrogyria in a child with KCNMA1-related channelopathy. Brain and Development, 2022, 44, 173-177.	0.6	7
4	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. Journal of Human Genetics, 2022, 67, 157-164.	1.1	16
5	De novo heterozygous variants in KIF5B cause kyphomelic dysplasia. Clinical Genetics, 2022, 102, 3-11.	1.0	5
6	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
7	Actin-binding protein filamin-A drives tau aggregation and contributes to progressive supranuclear palsy pathology. Science Advances, 2022, 8, .	4.7	15
8	Whole exome sequencing of fetal structural anomalies detected by ultrasonography. Journal of Human Genetics, 2021, 66, 499-507.	1.1	18
9	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
10	Efficient detection of copyâ€•number variations using exome data: Batchâ€•and sexâ€•based analyses. Human Mutation, 2021, 42, 50-65.	1.1	18
11	De novo ATP1A3 variants cause polymicrogyria. Science Advances, 2021, 7, .	4.7	13
12	Complete sequencing of expandedSAMD12repeats by long-read sequencing and Cas9-mediated enrichment. Brain, 2021, 144, 1103-1117.	3.7	25
13	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
14	Cerebrovascular diseases in two patients with entire NSD1 deletion. Human Genome Variation, 2021, 8, 20.	0.4	2
15	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
16	Novel CLTC variants cause new brain and kidney phenotypes. Journal of Human Genetics, 2021, , .	1.1	4
17	Expanding the <scp>KIF4A</scp>â€•associated phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3728-3739.	0.7	6
18	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

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19	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
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	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
22	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
23	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
24	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
25	GGC Repeat Expansion of NOTCH2NLC in Adult Patients with Leukoencephalopathy. Annals of Neurology, 2019, 86, 962-968.	2.8	98
26	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
27	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36
28	Tandem-genotypes: robust detection of tandem repeat expansions from long DNA reads. Genome Biology, 2019, 20, 58.	3.8	103
29	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	1.5	30
30	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
31	Leaky splicing variant in sepiapterin reductase deficiency. Neurology: Genetics, 2019, 5, e319.	0.9	10
32	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
33	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	0.7	10
34	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. Journal of Human Genetics, 2018, 63, 263-270.	1.1	19
35	A novel mutation in SLC1A3 causes episodic ataxia. Journal of Human Genetics, 2018, 63, 207-211.	1.1	42
36	De novo variants in RHOBTB2 , an atypical Rho GTPase gene, cause epileptic encephalopathy. Human Mutation, 2018, 39, 1070-1075.	1.1	25

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37	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. Cell Reports, 2018, 22, 734-747.	2.9	132
38	A novel SLC9A1 mutation causes cerebellar ataxia. Journal of Human Genetics, 2018, 63, 1049-1054.	1.1	28
39	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
40	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
41	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. Journal of Human Genetics, 2017, 62, 997-1000.	1.1	9
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	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. Scientific Reports, 2016, 6, 30072.	1.6	102
44	Milder progressive cerebellar atrophy caused by biallelic SEPSECS mutations. Journal of Human Genetics, 2016, 61, 527-531.	1.1	30
45	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. Scientific Reports, 2015, 4, 7132.	1.6	29
46	Predominant cerebellar phenotype in spastic paraplegia 7 (SPG7). Human Genome Variation, 2015, 2, 15012.	0.4	7
47	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
48	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
49	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
50	â€˜Cortical cerebellar atrophyâ€™ dwindles away in the era of next-generation sequencing. Journal of Human Genetics, 2014, 59, 589-590.	1.1	8