

# Satoko Miyatake

## List of Publications by Year in descending order

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Version: 2024-02-01

50  
papers

1,474  
citations

361413

20  
h-index

361022

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. <i>Journal of Human Genetics</i> , 2022, 67, 169-173.	2.3	7
2	Repeat conformation heterogeneity in cerebellar ataxia, neuropathy, vestibular areflexia syndrome. <i>Brain</i> , 2022, 145, 1139-1150.	7.6	19
3	Polymicrogyria in a child with KCNMA1-related channelopathy. <i>Brain and Development</i> , 2022, 44, 173-177.	1.1	7
4	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. <i>Journal of Human Genetics</i> , 2022, 67, 157-164.	2.3	16
5	De novo heterozygous variants in <i>KIF5B</i> cause kyphomelic dysplasia. <i>Clinical Genetics</i> , 2022, 102, 3-11.	2.0	5
6	A case of epilepsy of infancy with migrating focal seizures caused by mosaic <i>SCN2A</i> mutation. <i>Epilepsy and Seizure</i> , 2022, 14, 17-24.	0.2	0
7	Actin-binding protein filamin-A drives tau aggregation and contributes to progressive supranuclear palsy pathology. <i>Science Advances</i> , 2022, 8, .	10.3	15
8	Whole exome sequencing of fetal structural anomalies detected by ultrasonography. <i>Journal of Human Genetics</i> , 2021, 66, 499-507.	2.3	18
9	Novel EXOSC9 variants cause pontocerebellar hypoplasia type 1D with spinal motor neuronopathy and cerebellar atrophy. <i>Journal of Human Genetics</i> , 2021, 66, 401-407.	2.3	15
10	Efficient detection of copy number variations using exome data: Batch- and sex-based analyses. <i>Human Mutation</i> , 2021, 42, 50-65.	2.5	18
11	De novo ATP1A3 variants cause polymicrogyria. <i>Science Advances</i> , 2021, 7, .	10.3	13
12	Complete sequencing of expanded <i>SAMD12</i> repeats by long-read sequencing and Cas9-mediated enrichment. <i>Brain</i> , 2021, 144, 1103-1117.	7.6	25
13	Monoallelic and bi-allelic variants in NCDN cause neurodevelopmental delay, intellectual disability, and epilepsy. <i>American Journal of Human Genetics</i> , 2021, 108, 739-748.	6.2	15
14	Cerebrovascular diseases in two patients with entire NSD1 deletion. <i>Human Genome Variation</i> , 2021, 8, 20.	0.7	2
15	A 23-year follow-up report of juvenile-onset Sandhoff disease presenting with a motor neuron disease phenotype and a novel variant. <i>Brain and Development</i> , 2021, 43, 1029-1032.	1.1	1
16	Novel CLTC variants cause new brain and kidney phenotypes. <i>Journal of Human Genetics</i> , 2021, , .	2.3	4
17	Expanding the <i>KIF4A</i> -associated phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3728-3739.	1.2	6
18	Father-to-offspring transmission of extremely long NOTCH2NLC repeat expansions with contractions: genetic and epigenetic profiling with long-read sequencing. <i>Clinical Epigenetics</i> , 2021, 13, 204.	4.1	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.	1.1	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.	6.2	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.	1.5	0
22	Reply to â€œ<sc>GGC</sc> Repeat Expansion of <sc><i>NOTCH2NLC</i></sc> is Rare in European Leukoencephalopathyâ€“. Annals of Neurology, 2020, 88, 642-643.	5.3	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.	2.3	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.	1.1	9
25	GGC Repeat Expansion of <i>NOTCH2NLC</i> in Adult Patients with Leukoencephalopathy. Annals of Neurology, 2019, 86, 962-968.	5.3	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.	2.3	11
27	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	2.3	36
28	Tandem-genotypes: robust detection of tandem repeat expansions from long DNA reads. Genome Biology, 2019, 20, 58.	8.8	103
29	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	3.2	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.	2.3	48
31	Leaky splicing variant in sepiapterin reductase deficiency. Neurology: Genetics, 2019, 5, e319.	1.9	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.	2.3	33
33	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	1.2	10
34	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. Journal of Human Genetics, 2018, 63, 263-270.	2.3	19
35	A novel mutation in SLC1A3 causes episodic ataxia. Journal of Human Genetics, 2018, 63, 207-211.	2.3	42
36	De novo variants in <i>RHOTB2</i>, an atypical Rho GTPase gene, cause epileptic encephalopathy. Human Mutation, 2018, 39, 1070-1075.	2.5	25

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37	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. <i>Cell Reports</i> , 2018, 22, 734-747.	6.4	132
38	A novel SLC9A1 mutation causes cerebellar ataxia. <i>Journal of Human Genetics</i> , 2018, 63, 1049-1054.	2.3	28
39	Identification of novel <i>SNORD118</i> mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. <i>Clinical Genetics</i> , 2017, 92, 180-187.	2.0	28
40	Biallelic Mutations in MYPN , Encoding Myopalladin, Are Associated with Childhood-Onset, Slowly Progressive Nemaline Myopathy. <i>American Journal of Human Genetics</i> , 2017, 100, 169-178.	6.2	66
41	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. <i>Journal of Human Genetics</i> , 2017, 62, 997-1000.	2.3	9
42	A novel DARS2 mutation in a Japanese patient with leukoencephalopathy with brainstem and spinal cord involvement but no lactate elevation. <i>Human Genome Variation</i> , 2017, 4, 17051.	0.7	6
43	Impaired neuronal KCC2 function by biallelic SLC12A5 mutations in migrating focal seizures and severe developmental delay. <i>Scientific Reports</i> , 2016, 6, 30072.	3.3	102
44	Milder progressive cerebellar atrophy caused by biallelic SEPSECS mutations. <i>Journal of Human Genetics</i> , 2016, 61, 527-531.	2.3	30
45	Late-onset spastic ataxia phenotype in a patient with a homozygous DDHD2 mutation. <i>Scientific Reports</i> , 2015, 4, 7132.	3.3	29
46	Predominant cerebellar phenotype in spastic paraplegia 7 (SPG7). <i>Human Genome Variation</i> , 2015, 2, 15012.	0.7	7
47	Dominant mutations in ORAI1 cause tubular aggregate myopathy with hypocalcemia via constitutive activation of store-operated Ca <sup>2+</sup> channels. <i>Human Molecular Genetics</i> , 2015, 24, 637-648.	2.9	132
48	A Novel Mutation in <i>ELOVL4</i> Leading to Spinocerebellar Ataxia (SCA) With the Hot Cross Bun Sign but Lacking Erythrokeratoderma. <i>JAMA Neurology</i> , 2015, 72, 797.	9.0	79
49	De novo KIF1A mutations cause intellectual deficit, cerebellar atrophy, lower limb spasticity and visual disturbance. <i>Journal of Human Genetics</i> , 2015, 60, 739-742.	2.3	58
50	“Cortical cerebellar atrophy” dwindles away in the era of next-generation sequencing. <i>Journal of Human Genetics</i> , 2014, 59, 589-590.	2.3	8