

# Takeshi Mizuguchi

## List of Publications by Year in descending order

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Version: 2024-02-01

53  
papers

1,846  
citations

430754

18  
h-index

289141

40  
g-index

53  
all docs

53  
docs citations

53  
times ranked

3392  
citing authors

#	ARTICLE	IF	CITATIONS
1	De novo mutations in the gene encoding STXBP1 (MUNC18-1) cause early infantile epileptic encephalopathy. <i>Nature Genetics</i> , 2008, 40, 782-788.	9.4	498
2	Long-read sequencing identifies GGC repeat expansions in NOTCH2NLC associated with neuronal intranuclear inclusion disease. <i>Nature Genetics</i> , 2019, 51, 1215-1221.	9.4	328
3	Tandem-genotypes: robust detection of tandem repeat expansions from long DNA reads. <i>Genome Biology</i> , 2019, 20, 58.	3.8	103
4	GGC Repeat Expansion of <i>NOTCH2NLC</i> in Adult Patients with Leukoencephalopathy. <i>Annals of Neurology</i> , 2019, 86, 962-968.	2.8	98
5	A 12-kb structural variation in progressive myoclonic epilepsy was newly identified by long-read whole-genome sequencing. <i>Journal of Human Genetics</i> , 2019, 64, 359-368.	1.1	48
6	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. <i>Journal of Human Genetics</i> , 2019, 64, 967-978.	1.1	43
7	A novel mutation in SLC1A3 causes episodic ataxia. <i>Journal of Human Genetics</i> , 2018, 63, 207-211.	1.1	42
8	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders. <i>Human Molecular Genetics</i> , 2018, 27, 1421-1433.	1.4	36
9	Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. <i>Journal of Human Genetics</i> , 2019, 64, 1173-1186.	1.1	36
10	Detecting a long insertion variant in SAMD12 by SMRT sequencing: implications of long-read whole-genome sequencing for repeat expansion diseases. <i>Journal of Human Genetics</i> , 2019, 64, 191-197.	1.1	33
11	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. <i>Journal of Medical Genetics</i> , 2019, 56, 396-407.	1.5	30
12	ATP6VOA1 encoding the $\alpha$ 1-subunit of the V0 domain of vacuolar H <sup>+</sup> -ATPases is essential for brain development in humans and mice. <i>Nature Communications</i> , 2021, 12, 2107.	5.8	30
13	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.	1.0	28
14	A novel SLC9A1 mutation causes cerebellar ataxia. <i>Journal of Human Genetics</i> , 2018, 63, 1049-1054.	1.1	28
15	Primary immunodeficiency with chronic enteropathy and developmental delay in a boy arising from a novel homozygous RIPK1 variant. <i>Journal of Human Genetics</i> , 2019, 64, 955-960.	1.1	28
16	De novo variants in <i>RHOBTB2</i> , an atypical Rho GTPase gene, cause epileptic encephalopathy. <i>Human Mutation</i> , 2018, 39, 1070-1075.	1.1	25
17	Gain-of-Function MN1 Truncation Variants Cause a Recognizable Syndrome with Craniofacial and Brain Abnormalities. <i>American Journal of Human Genetics</i> , 2020, 106, 13-25.	2.6	25
18	Complete sequencing of expanded <i>SAMD12</i> repeats by long-read sequencing and Cas9-mediated enrichment. <i>Brain</i> , 2021, 144, 1103-1117.	3.7	25

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19	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.	1.8	22
20	A novel <i>CYCS</i> mutation in the $\alpha$ -helix of the CYCS C-terminal domain causes non-syndromic thrombocytopenia. <i>Clinical Genetics</i> , 2018, 94, 548-553.	1.0	20
21	Neuronal intranuclear inclusion disease presenting with an MELAS-like episode in chronic polyneuropathy. <i>Neurology: Genetics</i> , 2020, 6, e531.	0.9	20
22	Novel recessive mutations in <i>MSTO1</i> cause cerebellar atrophy with pigmentary retinopathy. <i>Journal of Human Genetics</i> , 2018, 63, 263-270.	1.1	19
23	Whole exome sequencing of fetal structural anomalies detected by ultrasonography. <i>Journal of Human Genetics</i> , 2021, 66, 499-507.	1.1	18
24	Efficient detection of copy number variations using exome data: Batch- and sex-based analyses. <i>Human Mutation</i> , 2021, 42, 50-65.	1.1	18
25	Missense and truncating variants in <i>CHD5</i> in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. <i>Human Genetics</i> , 2021, 140, 1109-1120.	1.8	18
26	Phenotype-genotype correlations in patients with <i>GNB1</i> gene variants, including the first three reported Japanese patients to exhibit spastic diplegia, dyskinetic quadriplegia, and infantile spasms. <i>Brain and Development</i> , 2020, 42, 199-204.	0.6	16
27	Two families with <i>TET3</i> -related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. <i>Journal of Human Genetics</i> , 2022, 67, 157-164.	1.1	16
28	Clinical features of <i>SMARCA2</i> duplication overlap with Coffin-Siris syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2662-2670.	0.7	15
29	Novel <i>EXOSC9</i> variants cause pontocerebellar hypoplasia type 1D with spinal motor neuronopathy and cerebellar atrophy. <i>Journal of Human Genetics</i> , 2021, 66, 401-407.	1.1	15
30	Pathogenic variants in the survival of motor neurons complex gene <i>GEMIN5</i> cause cerebellar atrophy. <i>Clinical Genetics</i> , 2021, 100, 722-730.	1.0	15
31	A novel <i>ITPA</i> variant causes epileptic encephalopathy with multiple-organ dysfunction. <i>Journal of Human Genetics</i> , 2020, 65, 751-757.	1.1	13
32	De novo <i>ATP1A3</i> variants cause polymicrogyria. <i>Science Advances</i> , 2021, 7, .	4.7	13
33	A novel <i>STXBP1</i> mutation causes typical Rett syndrome in a Japanese girl. <i>Brain and Development</i> , 2018, 40, 493-497.	0.6	11
34	Hemorrhagic stroke and renovascular hypertension with Grange syndrome arising from a novel pathogenic variant in <i>YY1AP1</i> . <i>Journal of Human Genetics</i> , 2019, 64, 885-890.	1.1	11
35	Pathogenic 12-kb copy-neutral inversion in syndromic intellectual disability identified by high-fidelity long-read sequencing. <i>Genomics</i> , 2021, 113, 1044-1053.	1.3	11
36	SOFT syndrome in a patient from Chile. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 338-340.	0.7	10

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37	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. <i>Journal of Human Genetics</i> , 2017, 62, 997-1000.	1.1	9
38	Entire FGF12 duplication by complex chromosomal rearrangements associated with West syndrome. <i>Journal of Human Genetics</i> , 2019, 64, 1005-1014.	1.1	9
39	Comparison of mitochondrial DNA variants detection using short- and long-read sequencing. <i>Journal of Human Genetics</i> , 2019, 64, 1107-1116.	1.1	8
40	Long-read whole-genome sequencing identified a partial MBD5 deletion in an exome-negative patient with neurodevelopmental disorder. <i>Journal of Human Genetics</i> , 2021, 66, 697-705.	1.1	8
41	Rapid progression of a walking disability in a 5-year-old boy with a CLN6 mutation. <i>Brain and Development</i> , 2019, 41, 726-730.	0.6	7
42	Clinical variations of epileptic syndrome associated with PACS2 variant. <i>Brain and Development</i> , 2021, 43, 343-347.	0.6	7
43	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.	1.1	7
44	Amelioration of a neurodevelopmental disorder by carbamazepine in a case having a gain-of-function GRIA3 variant. <i>Human Genetics</i> , 2022, 141, 283-293.	1.8	6
45	Novel ACOX1 mutations in two siblings with peroxisomal acyl-CoA oxidase deficiency. <i>Brain and Development</i> , 2021, 43, 475-481.	0.6	5
46	Novel CLTC variants cause new brain and kidney phenotypes. <i>Journal of Human Genetics</i> , 2021, , .	1.1	4
47	Intellectual disability and microcephaly associated with a novel CHAMP1 mutation. <i>Human Genome Variation</i> , 2021, 8, 34.	0.4	3
48	Reply to "Repeat Expansion of <i>NOTCH2NLC</i> is Rare in European Leukoencephalopathy". <i>Annals of Neurology</i> , 2020, 88, 642-643.	2.8	2
49	Monogenic causes of pigmentary mosaicism. <i>Human Genetics</i> , 2022, , .	1.8	2
50	Genetic and Imaging Characteristics of a Family With Neuronal Intranuclear Inclusion Disease.		