

Takeshi Mizuguchi

List of Publications by Year in descending order

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Version: 2024-02-01

53
papers

1,846
citations

430442

18
h-index

288905

40
g-index

53
all docs

53
docs citations

53
times ranked

3392
citing authors

#	ARTICLE	IF	CITATIONS
1	Gait disturbance in a patient with de novo 1.0-kb SOX2 microdeletion. <i>Brain and Development</i> , 2022, 44, 68-72.	0.6	1
2	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
3	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. <i>Journal of Human Genetics</i> , 2022, 67, 157-164.	1.1	16
4	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
5	Monogenic causes of pigmentary mosaicism. <i>Human Genetics</i> , 2022, , .	1.8	2
6	Genetic and Imaging Characteristics of a Family With Neuronal Intranuclear Inclusion Disease.		

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19	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
20	Head titubation and irritability as early symptoms of Joubert syndrome with a homozygous NPHP1 variant. <i>Brain and Development</i> , 2021, 43, 863-866.	0.6	1
21	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
22	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
23	Two males with sick sinus syndrome in a family with 0.6-kb deletions involving major domains in MECP2. <i>European Journal of Medical Genetics</i> , 2020, 63, 103769.	0.7	0
24	Phenotype-genotype correlations in patients with GNB1 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
25	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
26	A novel ITPA variant causes epileptic encephalopathy with multiple-organ dysfunction. <i>Journal of Human Genetics</i> , 2020, 65, 751-757.	1.1	13
27	Reply to "GGC Repeat Expansion of <i>NOTCH2NLC</i> is Rare in European Leukoencephalopathy". <i>Annals of Neurology</i> , 2020, 88, 642-643.	2.8	2
28	Neuronal intranuclear inclusion disease presenting with an MELAS-like episode in chronic polyneuropathy. <i>Neurology: Genetics</i> , 2020, 6, e531.	0.9	20
29	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
30	GGC Repeat Expansion of <i>NOTCH2NLC</i> in Adult Patients with Leukoencephalopathy. <i>Annals of Neurology</i> , 2019, 86, 962-968.	2.8	98
31	Entire FGF12 duplication by complex chromosomal rearrangements associated with West syndrome. <i>Journal of Human Genetics</i> , 2019, 64, 1005-1014.	1.1	9
32	Hemorrhagic stroke and renovascular hypertension with Grange syndrome arising from a novel pathogenic variant in YY1AP1. <i>Journal of Human Genetics</i> , 2019, 64, 885-890.	1.1	11
33	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
34	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
35	Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. <i>Journal of Human Genetics</i> , 2019, 64, 1173-1186.	1.1	36
36	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

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37	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
38	Tandem-genotypes: robust detection of tandem repeat expansions from long DNA reads. <i>Genome Biology</i> , 2019, 20, 58.	3.8	103
39	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
40	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
41	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
42	SOFT syndrome in a patient from Chile. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 338-340.	0.7	10
43	A novel STXBP1 mutation causes typical Rett syndrome in a Japanese girl. <i>Brain and Development</i> , 2018, 40, 493-497.	0.6	11
44	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
45	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. <i>Journal of Human Genetics</i> , 2018, 63, 263-270.	1.1	19
46	A novel mutation in SLC1A3 causes episodic ataxia. <i>Journal of Human Genetics</i> , 2018, 63, 207-211.	1.1	42
47	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
48	A novel SLC9A1 mutation causes cerebellar ataxia. <i>Journal of Human Genetics</i> , 2018, 63, 1049-1054.	1.1	28
49	A novel <i>CYCS</i> mutation in the α -helix of the CYCS C-terminal domain causes non-syndromic thrombocytopenia. <i>Clinical Genetics</i> , 2018, 94, 548-553.	1.0	20
50	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
51	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
52	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
53	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