Takeshi Mizuguchi

List of Publications by Year
in descending order

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53	1,846	430874	289244
papers	citations	18 h-index	40 g-index

53	53	53	3392
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	De novo mutations in the gene encoding STXBP1 (MUNC18-1) cause early infantile epileptic encephalopathy. Nature Genetics, 2008, 40, 782-788.	21.4	498
2	Long-read sequencing identifies GGC repeat expansions in NOTCH2NLC associated with neuronal intranuclear inclusion disease. Nature Genetics, 2019, 51, 1215-1221.	21.4	328
3	Tandem-genotypes: robust detection of tandem repeat expansions from long DNA reads. Genome Biology, 2019, 20, 58.	8.8	103
4	GGC Repeat Expansion of NOTCH2NLC in Adult Patients with Leukoencephalopathy. Annals of Neurology, 2019, 86, 962-968.	5.3	98
5	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
6	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978.	2.3	43
7	A novel mutation in SLC1A3 causes episodic ataxia. Journal of Human Genetics, 2018, 63, 207-211.	2.3	42
8	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders. Human Molecular Genetics, 2018, 27, 1421-1433.	2.9	36
9	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	2.3	36
10	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
11	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	3.2	30
12	ATP6V0A1 encoding the a1-subunit of the V0 domain of vacuolar H+-ATPases is essential for brain development in humans and mice. Nature Communications, 2021, 12, 2107.	12.8	30
13	Identification of novel <scp>SNORD118</scp> mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. Clinical Genetics, 2017, 92, 180-187.	2.0	28
14	A novel SLC9A1 mutation causes cerebellar ataxia. Journal of Human Genetics, 2018, 63, 1049-1054.	2.3	28
15	Primary immunodeficiency with chronic enteropathy and developmental delay in a boy arising from a novel homozygous RIPK1 variant. Journal of Human Genetics, 2019, 64, 955-960.	2.3	28
16	De novo variants in RHOBTB2 , an atypical Rho GTPase gene, cause epileptic encephalopathy. Human Mutation, 2018, 39, 1070-1075.	2.5	25
17	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
18	Complete sequencing of expandedSAMD12repeats by long-read sequencing and Cas9-mediated enrichment. Brain, 2021, 144, 1103-1117.	7.6	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. Clinical Epigenetics, 2021, 13, 204.	4.1	22
20	A novel CYCS mutation in the Î±â€helix of the CYCS Câ€terminal domain causes nonâ€syndromic thrombocytopenia. Clinical Genetics, 2018, 94, 548-553.	2.0	20
21	Neuronal intranuclear inclusion disease presenting with an MELAS-like episode in chronic polyneuropathy. Neurology: Genetics, 2020, 6, e531.	1.9	20
22	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. Journal of Human Genetics, 2018, 63, 263-270.	2.3	19
23	Whole exome sequencing of fetal structural anomalies detected by ultrasonography. Journal of Human Genetics, 2021, 66, 499-507.	2.3	18
24	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	2.5	18
25	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	3.8	18
26	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
27	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. Journal of Human Genetics, 2022, 67, 157-164.	2.3	16
28	Clinical features of SMARCA2 duplication overlap with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2662-2670.	1.2	15
29	Novel EXOSC9 variants cause pontocerebellar hypoplasia type 1D with spinal motor neuronopathy and cerebellar atrophy. Journal of Human Genetics, 2021, 66, 401-407.	2.3	15
30	Pathogenic variants in the survival of motor neurons complex gene <scp>GEMIN5</scp> cause cerebellar atrophy. Clinical Genetics, 2021, 100, 722-730.	2.0	15
31	A novel ITPA variant causes epileptic encephalopathy with multiple-organ dysfunction. Journal of Human Genetics, 2020, 65, 751-757.	2.3	13
32	De novo ATP1A3 variants cause polymicrogyria. Science Advances, 2021, 7, .	10.3	13
33	A novel STXBP1 mutation causes typical Rett syndrome in a Japanese girl. Brain and Development, 2018, 40, 493-497.	1.1	11
34	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
35	Pathogenic 12-kb copy-neutral inversion in syndromic intellectual disability identified by high-fidelity long-read sequencing. Genomics, 2021, 113, 1044-1053.	2.9	11
36	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	1.2	10

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37	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. Journal of Human Genetics, 2017, 62, 997-1000.	2.3	9
38	Entire FGF12 duplication by complex chromosomal rearrangements associated with West syndrome. Journal of Human Genetics, 2019, 64, 1005-1014.	2.3	9
39	Comparison of mitochondrial DNA variants detection using short- and long-read sequencing. Journal of Human Genetics, 2019, 64, 1107-1116.	2.3	8
40	Long-read whole-genome sequencing identified a partial MBD5 deletion in an exome-negative patient with neurodevelopmental disorder. Journal of Human Genetics, 2021, 66, 697-705.	2.3	8
41	Rapid progression of a walking disability in a 5-year-old boy with a CLN6 mutation. Brain and Development, 2019, 41, 726-730.	1.1	7
42	Clinical variations of epileptic syndrome associated with PACS2 variant. Brain and Development, 2021, 43, 343-347.	1.1	7
43	Biallelic null variants in ZNF142 cause global developmental delay with familial epilepsy and dysmorphic features. Journal of Human Genetics, 2022, 67, 169-173.	2.3	7
44	Amelioration of a neurodevelopmental disorder by carbamazepine in a case having a gain-of-function GRIA3 variant. Human Genetics, 2022, 141, 283-293.	3.8	6
45	Novel ACOX1 mutations in two siblings with peroxisomal acyl-CoA oxidase deficiency. Brain and Development, 2021, 43, 475-481.	1.1	5
46	Novel CLTC variants cause new brain and kidney phenotypes. Journal of Human Genetics, 2021, , .	2.3	4
47	Intellectual disability and microcephaly associated with a novel CHAMP1 mutation. Human Genome Variation, 2021, 8, 34.	0.7	3
48	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.	5.3	2
49	Monogenic causes of pigmentary mosaicism. Human Genetics, 2022, , .	3.8	2
50	Genetic and Imaging Characteristics of a Family With Neuronal Intranuclear Inclusion Disease.		