Takeshi Mizuguchi

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9574024/publications.pdf

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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	Gait disturbance in a patient with de novo 1.0-kb SOX2 microdeletion. Brain and Development, 2022, 44, 68-72.	1.1	1
2	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
3	Two families with TET3-related disorder showing neurodevelopmental delay with craniofacial dysmorphisms. Journal of Human Genetics, 2022, 67, 157-164.	2.3	16
4	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
5	Monogenic causes of pigmentary mosaicism. Human Genetics, 2022, , .	3.8	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 <scp>GEMIN5</scp> cause cerebellar atrophy. Clinical Genetics, 2021, 100, 722-730.	2.0	15
20	Head titubation and irritability as early symptoms of Joubert syndrome with a homozygous NPHP1 variant. Brain and Development, 2021, 43, 863-866.	1.1	1
21	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
22	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
23	Two males with sick sinus syndrome in a family with 0.6â€‾kb deletions involving major domains in MECP2. European Journal of Medical Genetics, 2020, 63, 103769.	1.3	0
24	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
25	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
26	A novel ITPA variant causes epileptic encephalopathy with multiple-organ dysfunction. Journal of Human Genetics, 2020, 65, 751-757.	2.3	13
27	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
28	Neuronal intranuclear inclusion disease presenting with an MELAS-like episode in chronic polyneuropathy. Neurology: Genetics, 2020, 6, e531.	1.9	20
29	Comparison of mitochondrial DNA variants detection using short- and long-read sequencing. Journal of Human Genetics, 2019, 64, 1107-1116.	2.3	8
30	GGC Repeat Expansion of NOTCH2NLC in Adult Patients with Leukoencephalopathy. Annals of Neurology, 2019, 86, 962-968.	5.3	98
31	Entire FGF12 duplication by complex chromosomal rearrangements associated with West syndrome. Journal of Human Genetics, 2019, 64, 1005-1014.	2.3	9
32	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
33	Long-read sequencing identifies GGC repeat expansions in NOTCH2NLC associated with neuronal intranuclear inclusion disease. Nature Genetics, 2019, 51, 1215-1221.	21.4	328
34	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978.	2.3	43
35	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	2.3	36
36	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

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37	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
38	Tandem-genotypes: robust detection of tandem repeat expansions from long DNA reads. Genome Biology, 2019, 20, 58.	8.8	103
39	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	3.2	30
40	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
41	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
42	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	1.2	10
43	A novel STXBP1 mutation causes typical Rett syndrome in a Japanese girl. Brain and Development, 2018, 40, 493-497.	1.1	11
44	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders. Human Molecular Genetics, 2018, 27, 1421-1433.	2.9	36
45	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. Journal of Human Genetics, 2018, 63, 263-270.	2.3	19
46	A novel mutation in SLC1A3 causes episodic ataxia. Journal of Human Genetics, 2018, 63, 207-211.	2.3	42
47	De novo variants in RHOBTB2 , an atypical Rho GTPase gene, cause epileptic encephalopathy. Human Mutation, 2018, 39, 1070-1075.	2.5	25
48	A novel SLC9A1 mutation causes cerebellar ataxia. Journal of Human Genetics, 2018, 63, 1049-1054.	2.3	28
49	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
50	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
51	An atypical case of SPG56/CYP2U1-related spastic paraplegia presenting with delayed myelination. Journal of Human Genetics, 2017, 62, 997-1000.	2.3	9
52	Clinical features of SMARCA2 duplication overlap with Coffinâ€“Siris syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 2662-2670.	1.2	15
53	De novo mutations in the gene encoding STXBP1 (MUNC18-1) cause early infantile epileptic encephalopathy. Nature Genetics, 2008, 40, 782-788.	21.4	498