Atsushi Takata

List of Publications by Year
in descending order

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23	893	567144 15	677027 22
papers	citations	h-index	g-index

23	23	23	2361
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genetics of bipolar disorder: insights into its complex architecture and biology from common and rare variants. Journal of Human Genetics, 2023, 68, 183-191.	1.1	5
2	Whole exome sequencing of fetal structural anomalies detected by ultrasonography. Journal of Human Genetics, 2021, 66, 499-507.	1.1	18
3	Novel EXOSC9 variants cause pontocerebellar hypoplasia type 1D with spinal motor neuronopathy and cerebellar atrophy. Journal of Human Genetics, 2021, 66, 401-407.	1.1	15
4	Efficient detection of copyâ€•number variations using exome data: Batchâ€•and sexâ€•based analyses. Human Mutation, 2021, 42, 50-65.	1.1	18
5	De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. Human Mutation, 2021, 42, 66-76.	1.1	16
6	De novo ATP1A3 variants cause polymicrogyria. Science Advances, 2021, 7, .	4.7	13
7	Refinement of the clinical variant interpretation framework by statistical evidence and machine learning. Med, 2021, 2, 611-632.e9.	2.2	1
8	Systematic analysis of exonic germline and postzygotic de novo mutations in bipolar disorder. Nature Communications, 2021, 12, 3750.	5.8	15
9	Gain-of-Function MN1 Truncation Variants Cause a Recognizable Syndrome with Craniofacial and Brain Abnormalities. American Journal of Human Genetics, 2020, 106, 13-25.	2.6	25
10	Hemorrhagic stroke and renovascular hypertension with Grange syndrome arising from a novel pathogenic variant in YY1AP1. Journal of Human Genetics, 2019, 64, 885-890.	1.1	11
11	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978.	1.1	43
12	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	1.1	36
13	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.	1.1	28
14	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. Nature Communications, 2019, 10, 2506.	5.8	46
15	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	1.5	30
16	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	0.7	10
17	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. Journal of Human Genetics, 2018, 63, 263-270.	1.1	19
18	A novel mutation in SLC1A3 causes episodic ataxia. Journal of Human Genetics, 2018, 63, 207-211.	1.1	42

#	Article	IF	Citations
19	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. Cell Reports, 2018, 22, 734-747.	2.9	132
20	A novel SLC9A1 mutation causes cerebellar ataxia. Journal of Human Genetics, 2018, 63, 1049-1054.	1.1	28
21	Identification of novel <scp>SNORD118</scp> mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. Clinical Genetics, 2017, 92, 180-187.	1.0	28
22	De Novo Synonymous Mutations in Regulatory Elements Contribute to the Genetic Etiology of Autism and Schizophrenia. Neuron, 2016, 89, 940-947.	3.8	140
23	Loss-of-Function Variants in Schizophrenia Risk and SETD1A as a Candidate Susceptibility Gene. Neuron, 2014, 82, 773-780.	3.8	174