Atsushi Takata

List of Publications by Year in descending order

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567281 677142 23 893 15 22 citations h-index g-index papers 23 23 23 2361 docs citations times ranked citing authors all docs

#	Article	IF	CITATIONS
1	Loss-of-Function Variants in Schizophrenia Risk and SETD1A as a Candidate Susceptibility Gene. Neuron, 2014, 82, 773-780.	8.1	174
2	De Novo Synonymous Mutations in Regulatory Elements Contribute to the Genetic Etiology of Autism and Schizophrenia. Neuron, 2016, 89, 940-947.	8.1	140
3	Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. Cell Reports, 2018, 22, 734-747.	6.4	132
4	Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. Nature Communications, 2019, 10, 2506.	12.8	46
5	Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. Journal of Human Genetics, 2019, 64, 967-978.	2.3	43
6	A novel mutation in SLC1A3 causes episodic ataxia. Journal of Human Genetics, 2018, 63, 207-211.	2.3	42
7	Genetic abnormalities in a large cohort of Coffin–Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	2.3	36
8	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. Journal of Medical Genetics, 2019, 56, 396-407.	3.2	30
9	Identification of novel <i><scp>SNORD118</scp></i> mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. Clinical Genetics, 2017, 92, 180-187.	2.0	28
10	A novel SLC9A1 mutation causes cerebellar ataxia. Journal of Human Genetics, 2018, 63, 1049-1054.	2.3	28
11	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
12	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
13	Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. Journal of Human Genetics, 2018, 63, 263-270.	2.3	19
14	Whole exome sequencing of fetal structural anomalies detected by ultrasonography. Journal of Human Genetics, 2021, 66, 499-507.	2.3	18
15	Efficient detection of copyâ€number variations using exome data: Batch―and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	2,5	18
16	De novo variants in <i>CELF2</i> that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. Human Mutation, 2021, 42, 66-76.	2.5	16
17	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
18	Systematic analysis of exonic germline and postzygotic de novo mutations in bipolar disorder. Nature Communications, 2021, 12, 3750.	12.8	15

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#	Article	IF	CITATION
19	De novo ATP1A3 variants cause polymicrogyria. Science Advances, 2021, 7, .	10.3	13
20	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
21	SOFT syndrome in a patient from Chile. American Journal of Medical Genetics, Part A, 2019, 179, 338-340.	1.2	10
22	Genetics of bipolar disorder: insights into its complex architecture and biology from common and rare variants. Journal of Human Genetics, 2023, 68, 183-191.	2.3	5
23	Refinement of the clinical variant interpretation framework by statistical evidence and machine learning. Med, 2021, 2, 611-632.e9.	4.4	1