Hsiao-Tuan Chao

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3762297/publications.pdf

Version: 2024-02-01

35 papers	3,847 citations	257450 24 h-index	345221 36 g-index
40 all docs	40 docs citations	40 times ranked	6253 citing authors

#	Article	IF	Citations
1	PRUNE1 c.<scp>933G</scp>>A synonymous variant induces exon 7 skipping, disrupts the <scp>DHHA2</scp> domain, and leads to an atypical <scp>NMIHBA</scp> syndrome presentation: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2022, 188, 1868-1874.	1.2	2
2	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Cell Reports, 2022, 38, 110517.	6.4	24
3	An Integrated Phenotypic and Genotypic Approach Reveals a Highâ€Risk Subtype Association for <scp>EBF3</scp> Missense Variants Affecting the Zinc Finger Domain. Annals of Neurology, 2022, 92, 138-153.	5.3	5
4	Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, .	8.2	87
5	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genetics in Medicine, 2021, 23, 1889-1900.	2.4	13
6	EIF2AK2 -related Neurodevelopmental Disorder With Leukoencephalopathy, Developmental Delay, and Episodic Neurologic Regression Mimics Pelizaeus-Merzbacher Disease. Neurology: Genetics, 2021, 7, e539.	1.9	9
7	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
8	De Novo Variants in CDK19 Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. American Journal of Human Genetics, 2020, 106, 717-725.	6.2	23
9	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
10	Stxbp1/Munc18-1 haploinsufficiency impairs inhibition and mediates key neurological features of STXBP1 encephalopathy. ELife, 2020, 9, .	6.0	42
11	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
12	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	1.5	6
13	DYRK1A-related intellectual disability: a syndrome associated with congenital anomalies of the kidney and urinary tract. Genetics in Medicine, 2019, 21, 2755-2764.	2.4	19
14	In Vivo Functional Study of Disease-associated Rare Human Variants Using Drosophila. Journal of Visualized Experiments, 2019, , .	0.3	34
15	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	6.2	29
16	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. American Journal of Human Genetics, 2019, 104, 1127-1138.	6.2	59
17	Recurrent mosaic MTOR c.5930C > T (p.Thr1977Ile) variant causing megalencephaly, asymmetric polymicrogyria, and cutaneous pigmentary mosaicism: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 475-479.	1.2	11
18	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	6.2	181

#	Article	IF	Citations
19	Building dialogues between clinical and biomedical research through cross-species collaborations. Seminars in Cell and Developmental Biology, 2017, 70, 49-57.	5.0	16
20	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
21	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. Genetics, 2017, 207, 9-27.	2.9	165
22	Clinical and molecular characterization of de novo loss of function variants in HNRNPU. American Journal of Medical Genetics, Part A, 2017, 173, 2680-2689.	1.2	34
23	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
24	Investigation of Synapse Formation and Function in a Glutamatergic-GABAergic Two-Neuron Microcircuit. Journal of Neuroscience, 2014, 34, 855-868.	3.6	22
25	Dendritic Arborization and Spine Dynamics Are Abnormal in the Mouse Model ofMECP2Duplication Syndrome. Journal of Neuroscience, 2013, 33, 19518-19533.	3.6	123
26	MeCP2: only 100% will do. Nature Neuroscience, 2012, 15, 176-177.	14.8	59
27	Binding of the complexin N terminus to the SNARE complex potentiates synaptic-vesicle fusogenicity. Nature Structural and Molecular Biology, 2010, 17, 568-575.	8.2	113
28	Dysfunction in GABA signalling mediates autism-like stereotypies and Rett syndrome phenotypes. Nature, 2010, 468, 263-269.	27.8	1,042
29	The yin and yang of MeCP2 phosphorylation. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 4577-4578.	7.1	31
30	Math1 Is Essential for the Development of Hindbrain Neurons Critical for Perinatal Breathing. Neuron, 2009, 64, 341-354.	8.1	146
31	Loss of MeCP2 in aminergic neurons causes cell-autonomous defects in neurotransmitter synthesis and specific behavioral abnormalities. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 21966-21971.	7.1	240
32	Deletion of Mecp2 in Sim1-Expressing Neurons Reveals a Critical Role for MeCP2 in Feeding Behavior, Aggression, and the Response to Stress. Neuron, 2008, 59, 947-958.	8.1	230
33	A partial loss of function allele of Methyl-CpG-binding protein 2 predicts a human neurodevelopmental syndrome. Human Molecular Genetics, 2008, 17, 1718-1727.	2.9	173
34	MeCP2 Controls Excitatory Synaptic Strength by Regulating Glutamatergic Synapse Number. Neuron, 2007, 56, 58-65.	8.1	439
35	Distinct domains of complexin I differentially regulate neurotransmitter release. Nature Structural and Molecular Biology, 2007, 14, 949-958.	8.2	198