# 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<br>papers | 3,847<br>citations | 257450<br>24<br>h-index | 345221<br>36<br>g-index |
| 40<br>all docs | 40<br>docs citations | 40<br>times ranked | 6253<br>citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Dysfunction in GABA signalling mediates autism-like stereotypies and Rett syndrome phenotypes. Nature, 2010, 468, 263-269. | 27.8 | 1,042 |
| 2 | MeCP2 Controls Excitatory Synaptic Strength by Regulating Glutamatergic Synapse Number. Neuron, 2007, 56, 58-65. | 8.1 | 439 |
| 3 | 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 |
| 4 | 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 |
| 5 | Distinct domains of complexin I differentially regulate neurotransmitter release. Nature Structural and Molecular Biology, 2007, 14, 949-958. | 8.2 | 198 |
| 6 | 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 |
| 7 | 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 |
| 8 | Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. Genetics, 2017, 207, 9-27. | 2.9 | 165 |
| 9 | Math1 Is Essential for the Development of Hindbrain Neurons Critical for Perinatal Breathing. Neuron, 2009, 64, 341-354. | 8.1 | 146 |
| 10 | Dendritic Arborization and Spine Dynamics Are Abnormal in the Mouse Model ofMECP2Duplication Syndrome. Journal of Neuroscience, 2013, 33, 19518-19533. | 3.6 | 123 |
| 11 | 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 |
| 12 | A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137. | 6.2 | 96 |
| 13 | Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing. Journal of Clinical Investigation, 2021, 131, . | 8.2 | 87 |
| 14 | MeCP2: only 100% will do. Nature Neuroscience, 2012, 15, 176-177. | 14.8 | 59 |
| 15 | 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 |
| 16 | Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83. | 8.2 | 50 |
| 17 | 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 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Stxbp1/Munc18-1 haploinsufficiency impairs inhibition and mediates key neurological features of STXBP1 encephalopathy. ELife, 2020, 9, . | 6.0 | 42 |
| 20 | 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 |
| 21 | 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 |
| 22 | In Vivo Functional Study of Disease-associated Rare Human Variants Using <em>Drosophila</em>. Journal of Visualized Experiments, 2019, , . | 0.3 | 34 |
| 23 | 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 |
| 24 | 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 |
| 25 | 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 |
| 26 | 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 |
| 27 | Investigation of Synapse Formation and Function in a Glutamatergic-GABAergic Two-Neuron Microcircuit. Journal of Neuroscience, 2014, 34, 855-868. | 3.6 | 22 |
| 28 | 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 |
| 29 | Building dialogues between clinical and biomedical research through cross-species collaborations. Seminars in Cell and Developmental Biology, 2017, 70, 49-57. | 5.0 | 16 |
| 30 | Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genetics in Medicine, 2021, 23, 1889-1900. | 2.4 | 13 |
| 31 | 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 |
| 32 | EIF2AK2 -related Neurodevelopmental Disorder With Leukoencephalopathy, Developmental Delay, and Episodic Neurologic Regression Mimics Pelizaeus-Merzbacher Disease. Neurology: Genetics, 2021, 7, e539. | 1.9 | 9 |
| 33 | Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113. | 1.5 | 6 |
| 34 | 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 |
| 35 | 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 |