# Weiyi Mu

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9577539/publications.pdf

Version: 2024-02-01

| 21 | 555 | 932766 10 | 713013 21 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 21 | 21 | 21 | 1327 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Genetic testing for the epilepsies: A systematic review. Epilepsia, 2022, 63, 375-387. | 2.6 | 53 |
| 2 | Novel variants in KAT6B spectrum of disorders expand our knowledge of clinical manifestations and molecular mechanisms. Molecular Genetics & Genomic Medicine, 2021, 9, e1809. | 0.6 | 4 |
| 3 | Two siblings with a novel variant of EXOSC3 extended phenotypic spectrum of pontocerebellar hypoplasia 1B to an exceptionally mild form. BMJ Case Reports, 2021, 14, e236732. | 0.2 | 2 |
| 4 | Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation. Science Advances, 2021, 7, . | 4.7 | 25 |
| 5 | A structured genetics rotation for pediatric residents: an important educational opportunity. Genetics in Medicine, 2020, 22, 793-796. | 1.1 | 9 |
| 6 | De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism. American Journal of Human Genetics, 2020, 107, 352-363. | 2.6 | 64 |
| 7 | Adult patients with undiagnosed conditions and their responses to unresolved uncertainty from exome sequencing. Journal of Genetic Counseling, 2020, 29, 992-1003. | 0.9 | 3 |
| 8 | Relapsingâ€“remitting clinical course expands the phenotype of Aicardiâ€“GoutiÃ¨res syndrome. Annals of Clinical and Translational Neurology, 2020, 7, 254-258. | 1.7 | 2 |
| 9 | Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. American Journal of Human Genetics, 2020, 106, 484-495. | 2.6 | 22 |
| 10 | Intracranial calcifications and dystonia associated with a novel deletion of chromosome 8p11.2 encompassing SLC20A2 and THAP1. BMJ Case Reports, 2019, 12, e228782. | 0.2 | 7 |
| 11 | Factors affecting quality of life in children and adolescents with hypermobile Ehlersâ€Danlos syndrome/hypermobility spectrum disorders. American Journal of Medical Genetics, Part A, 2019, 179, 561-569. | 0.7 | 39 |
| 12 | Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. American Journal of Human Genetics, 2019, 105, 151-165. | 2.6 | 170 |
| 13 | Expansion of the clinical spectrum associated with AARS2â€related disorders. American Journal of Medical Genetics, Part A, 2019, 179, 1556-1564. | 0.7 | 20 |
| 14 | The utility of whole exome sequencing in diagnosing neurological disorders in adults from a highly consanguineous population. Journal of Neurogenetics, 2019, 33, 21-26. | 0.6 | 10 |
| 15 | Characterization of pulmonary arteriovenous malformations in ACVRL1 versus ENG mutation carriers in hereditary hemorrhagic telangiectasia. Genetics in Medicine, 2018, 20, 639-644. | 1.1 | 14 |
| 16 | Eighteen-year-old man with autism, obsessive compulsive disorder and aSHANK2variant presents with severe anorexia that responds to high-dose fluoxetine. BMJ Case Reports, 2018, 2018, bcr-2018-225119. | 0.2 | 7 |
| 17 | Pain and sleep quality in children with nonâ€vascular Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part A, 2018, 176, 1858-1864. | 0.7 | 12 |
| 18 | De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. Human Genetics, 2016, 135, 1399-1409. | 1.8 | 40 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Genotypeâ€“phenotype correlation of congenital anomalies in multiple congenital anomalies hypotonia seizures syndrome (MCAHS1)/PIGNâ€related epilepsy. American Journal of Medical Genetics, Part A, 2016, 170, 77-86. | 0.7 | 41 |
| 20 | Incontinentia Pigmenti with Persistent Hypercalcemia: Case Report. Pediatric Dermatology, 2016, 33, e315-7. | 0.5 | 3 |
| 21 | An atypical 0.73 MB microduplication of 22q11.21 and a novel SALL4 missense mutation associated with thumb agenesis and radioulnar synostosis. American Journal of Medical Genetics, Part A, 2015, 167, 1644-1649. | 0.7 | 8 |