# Megan T Cho

List of Publications by Year
in descending order



| 16 | 2,091 | 643344 15 | 1051228 16 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 17 | 17 | 17 | 6430 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Clinical application of whole-exome sequencing across clinical indications. Genetics in Medicine, 2016, 18, 696-704. | 1.1 | 780 |
| 2 | High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685. | 2.6 | 337 |
| 3 | Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352. | 2.6 | 230 |
| 4 | De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788. | 2.6 | 136 |
| 5 | Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. American Journal of Human Genetics, 2015, 97, 457-464. | 2.6 | 134 |
| 6 | De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363. | 2.6 | 86 |
| 7 | Mutations inSLC1A4, encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination. Journal of Medical Genetics, 2015, 52, 541-547. | 1.5 | 68 |
| 8 | Mutations in ARID2 are associated with intellectual disabilities. Neurogenetics, 2015, 16, 307-314. | 0.7 | 54 |
| 9 | Mutations inCOQ4, an essential component of coenzyme Q biosynthesis, cause lethal neonatal mitochondrial encephalomyopathy. Journal of Medical Genetics, 2015, 52, 627-635. | 1.5 | 48 |
| 10 | De novo missense variants inHECW2are associated with neurodevelopmental delay and hypotonia. Journal of Medical Genetics, 2017, 54, 84-86. | 1.5 | 46 |
| 11 | Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024. | 4.1 | 43 |
| 12 | A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178. | 0.7 | 32 |
| 13 | Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. Brain, 2019, 142, 2617-2630. | 3.7 | 31 |
| 14 | De novo mutations in MSL3 cause an X-linked syndrome marked by impaired histone H4 lysine 16 acetylation. Nature Genetics, 2018, 50, 1442-1451. | 9.4 | 28 |
| 15 | De novo missense variants in MEIS2 recapitulate the microdeletion phenotype of cardiac and palate abnormalities, developmental delay, intellectual disability and dysmorphic features. American Journal of Medical Genetics, Part A, 2018, 176, 1845-1851. | 0.7 | 21 |
| 16 | A qualitative study of Latinx parents' experiences of clinical exome sequencing. Journal of Genetic Counseling, 2020, 29, 574-586. | 0.9 | 16 |