# Jennifer Friedman

List of Publications by Year
in descending order



| 13 | 938 | 933447 10 | 1058476 14 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 14 | 14 | 14 | 2028 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Rapid whole-genome sequencing decreases infant morbidity and cost of hospitalization. Npj Genomic Medicine, 2018, 3, 10. | 3.8 | 314 |
| 2 | Intrafamilial phenotypic variability of the DYT1 dystonia: From asymptomaticTOR1A gene carrier status to dystonic storm. Movement Disorders, 2002, 17, 339-345. | 3.9 | 142 |
| 3 | TPP1 deficiency: Rare cause of isolated childhood-onset progressive ataxia. Neurology, 2015, 85, 1259-1261. | 1.1 | 138 |
| 4 | YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925. | 6.2 | 125 |
| 5 | KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261. | 7.6 | 57 |
| 6 | Alternating Hemiplegia of Childhood: Retrospective Genetic Study and Genotype-Phenotype Correlations in 187 Subjects from the US AHCF Registry. PLoS ONE, 2015, 10, e0127045. | 2.5 | 53 |
| 7 | Caenorhabditis elegans provides an efficient drug screening platform for GNAO1-related disorders and highlights the potential role of caffeine in controlling dyskinesia. Human Molecular Genetics, 2022, 31, 929-941. | 2.9 | 32 |
| 8 | De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546. | 2.4 | 24 |
| 9 | De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660. | 2.4 | 20 |
| 10 | Urine sepiapterin excretion as a new diagnostic marker for sepiapterin reductase deficiency. Molecular Genetics and Metabolism, 2015, 115, 157-160. | 1.1 | 18 |
| 11 | Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. Movement Disorders, 2021, 36, 690-703. | 3.9 | 7 |
| 12 | Ending a diagnostic odyssey: Moving from exome to genome to identify cockayne syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1623. | 1.2 | 3 |
| 13 | Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome. Brain, 2022, 145, 3383-3390. | 7.6 | 3 |