Jennifer Friedman

List of Publications by Year
in descending order

13	938	933264 10	1058333 14
papers	citations	h-index	g-index

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

#	Article	IF	Citations
1	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.	1.4	32
2	Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome. Brain, 2022, 145, 3383-3390.	3.7	3
3	Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. Movement Disorders, 2021, 36, 690-703.	2.2	7
4	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	1.1	20
5	Ending a diagnostic odyssey: Moving from exome to genome to identify cockayne syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1623.	0.6	3
6	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	1.1	24
7	KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation. Brain, 2020, 143, 3242-3261.	3.7	57
8	Rapid whole-genome sequencing decreases infant morbidity and cost of hospitalization. Npj Genomic Medicine, 2018, 3, 10.	1.7	314
9	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	2.6	125
10	TPP1 deficiency: Rare cause of isolated childhood-onset progressive ataxia. Neurology, 2015, 85, 1259-1261.	1.5	138
11	Urine sepiapterin excretion as a new diagnostic marker for sepiapterin reductase deficiency. Molecular Genetics and Metabolism, 2015, 115, 157-160.	0.5	18
12	Alternating Hemiplegia of Childhood: Retrospective Genetic Study and Genotype-Phenotype Correlations in 187 Subjects from the US AHCF Registry. PLoS ONE, 2015, 10, e0127045.	1.1	53
13	Intrafamilial phenotypic variability of the DYT1 dystonia: From asymptomaticTOR1A gene carrier status to dystonic storm. Movement Disorders, 2002, 17, 339-345.	2.2	142