Laurie A Robak

List of Publications by Year
in descending order

14 papers	1,196 citations	1040056 9 h-index	1058476 14 g-index
16 all docs	16 docs citations	16 times ranked	3326 citing authors

#	Article	IF	Citations
1	Genome Sequencing in the Parkinson Disease Clinic. Neurology: Genetics, 2022, 8, .	1.9	7
2	A novel de novo intronic variant in ITPR1 causes Gillespie syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2315-2324.	1.2	2
3	The expanding clinical phenotype of germline ABL1 â€associated congenital heart defects and skeletal malformations syndrome. Human Mutation, 2020, 41, 1738-1744.	2.5	10
4	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. Neurology: Genetics, 2020, 6, e498.	1.9	11
5	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Populationâ€Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. Movement Disorders, 2019, 34, 1851-1863.	3.9	47
6	De novo missense variant in the GTPase effector domain (GED) of DNM1L leads to static encephalopathy and seizures. Journal of Physical Education and Sports Management, 2019, 5, a003673.	1.2	24
7	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
8	The expanding neurological phenotype of DNM1L-related disorders. Brain, 2018, 141, e28-e28.	7.6	7
9	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. Neurobiology of Aging, 2017, 57, 247.e9-247.e13.	3.1	108
10	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. American Journal of Human Genetics, 2017, 100, 91-104.	6.2	72
11	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
12	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
13	Excessive burden of lysosomal storage disorder gene variants in Parkinsonâ€™s disease. Brain, 2017, 140, 3191-3203.	7.6	323
14	Whole-Exome Sequencing in Familial Parkinson Disease. JAMA Neurology, 2016, 73, 68.	9.0	71