Nicole J Lake

List of Publications by Year
in descending order

14	982	840776 11	1125743 13
papers	citations	h-index	g-index

19	19	19	1802
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Leigh syndrome: One disorder, more than 75 monogenic causes. Annals of Neurology, 2016, 79, 190-203.	5.3	374
2	Leigh Syndrome. Journal of Neuropathology and Experimental Neurology, 2015, 74, 482-492.	1.7	126
3	ATAD3 gene cluster deletions cause cerebellar dysfunction associated with altered mitochondrial DNA and cholesterol metabolism. Brain, 2017, 140, 1595-1610.	7.6	105
4	Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitoribosomal Subunit and Leigh Syndrome. American Journal of Human Genetics, 2017, 101, 239-254.	6.2	83
5	Mitochondrial DNA variation across 56,434 individuals in gnomAD. Genome Research, 2022, 32, 569-582.	5.5	59
6	Estimating prevalence for limb-girdle muscular dystrophy based on public sequencing databases. Genetics in Medicine, 2019, 21, 2512-2520.	2.4	56
7	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
8	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	4.4	33
9	TRAK2, a novel regulator of ABCA1 expression, cholesterol efflux and HDL biogenesis. European Heart Journal, 2017, 38, 3579-3587.	2.2	27
10	Leigh syndrome caused by mutations in <scp>MTFMT</scp> is associated with a better prognosis. Annals of Clinical and Translational Neurology, 2019, 6, 515-524.	3.7	17
11	Whole Exome Sequencing Identifies the Genetic Basis of Late-Onset Leigh Syndrome in a Patient with MRI but Little Biochemical Evidence of a Mitochondrial Disorder. JIMD Reports, 2016, 32, 117-124.	1.5	11
12	A patient with homozygous nonsense variants in two Leigh syndrome disease genes: Distinguishing a dual diagnosis from a hypomorphic proteinâ€truncating variant. Human Mutation, 2019, 40, 893-898.	2.5	8
13	MitoVisualize: a resource for analysis of variants in human mitochondrial RNAs and DNA. Bioinformatics, 2022, 38, 2967-2969.	4.1	1
14	Approaches for Dissection of the Genetic Basis of Complex Disease Development in Human. , 0, , .		0