

# Nicole J Lake

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/9145918/publications.pdf>

Version: 2024-02-01

14  
papers

982  
citations

840776

11  
h-index

1125743

13  
g-index

19  
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19  
docs citations

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times ranked

1802  
citing authors

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