

# Ali B Naini

## List of Publications by Year in descending order

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Version: 2024-02-01

22  
papers

1,415  
citations

687363

13  
h-index

713466

21  
g-index

22  
all docs

22  
docs citations

22  
times ranked

1809  
citing authors

#	ARTICLE	IF	CITATIONS
1	Leigh Syndrome with Nephropathy and CoQ10 Deficiency Due to decaprenyl diphosphate synthase subunit 2 (PDSS2) Mutations. <i>American Journal of Human Genetics</i> , 2006, 79, 1125-1129.	6.2	359
2	A Mutation in Para-Hydroxybenzoate-Polyprenyl Transferase (COQ2) Causes Primary Coenzyme Q10 Deficiency. <i>American Journal of Human Genetics</i> , 2006, 78, 345-349.	6.2	322
3	Increased expression of the pro-inflammatory enzyme cyclooxygenase-2 in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2001, 49, 176-185.	5.3	266
4	Chronic levodopa administration alters cerebral mitochondrial respiratory chain activity. <i>Annals of Neurology</i> , 1993, 34, 715-723.	5.3	144
5	Peripheral and central pharmacokinetics of apomorphine and its effect on dopamine metabolism in humans. <i>Movement Disorders</i> , 1995, 10, 28-36.	3.9	57
6	Muscle Phosphoglycerate Mutase Deficiency Revisited. <i>Archives of Neurology</i> , 2009, 66, 394-8.	4.5	40
7	Lack of Effect of Coenzyme Q10 on Doxorubicin Cytotoxicity in Breast Cancer Cell Cultures. <i>Integrative Cancer Therapies</i> , 2012, 11, 243-250.	2.0	32
8	Functional cellular analyses reveal energy metabolism defect and mitochondrial DNA depletion in a case of mitochondrial aconitase deficiency. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 28-34.	1.1	32
9	Novel recessive mutations in COQ4 cause severe infantile cardiomyopathy and encephalopathy associated with CoQ 10 deficiency. <i>Molecular Genetics and Metabolism Reports</i> , 2017, 12, 23-27.	1.1	27
10	Lack of aprataxin impairs mitochondrial functions via downregulation of the APE1/NRF1/NRF2 pathway. <i>Human Molecular Genetics</i> , 2015, 24, 4516-4529.	2.9	23
11	Whole exome sequencing identifies a homozygous POLG2 missense variant in an infant with fulminant hepatic failure and mitochondrial DNA depletion. <i>European Journal of Medical Genetics</i> , 2016, 59, 540-545.	1.3	21
12	Leber hereditary optic neuropathy plus dystonia, and transverse myelitis due to double mutations in MT-ND4 and MT-ND6. <i>Journal of Neurology</i> , 2020, 267, 823-829.	3.6	17
13	Detection of Mutations in mtDNA. <i>Methods in Cell Biology</i> , 2007, 80, 437-463.	1.1	15
14	A Novel SUCLA2 Mutation Presenting as a Complex Childhood Movement Disorder. <i>Journal of Child Neurology</i> , 2017, 32, 246-250.	1.4	15
15	Characterization of the human homozygous R182W POLG2 mutation in mitochondrial DNA depletion syndrome. <i>PLoS ONE</i> , 2018, 13, e0203198.	2.5	11
16	Identification of a novel D109Y mutation in Cu/Zn superoxide dismutase (sod1) gene associated with amyotrophic lateral sclerosis. <i>Journal of the Neurological Sciences</i> , 2007, 254, 17-21.	0.6	9
17	Compound heterozygous inheritance of two novel COQ2 variants results in familial coenzyme Q deficiency. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 320.	2.7	8
18	Increased expression of the pro-inflammatory enzyme cyclooxygenase-2 in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2001, 49, 176-185.	5.3	6

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19	Detection of mitochondrial DNA (mtDNA) mutations. <i>Methods in Cell Biology</i> , 2020, 155, 383-400.	1.1	5
20	A De Novo Mutation in MTND6 Causes Generalized Dystonia in 2 Unrelated Children. <i>Child Neurology Open</i> , 2016, 3, 2329048X1562793.	1.1	4
21	Whole-Exome Sequencing Identifies a Novel POLG Frameshift Variant in an Adult Patient Presenting with Progressive External Ophthalmoplegia and Mitochondrial DNA Depletion. <i>Case Reports in Genetics</i> , 2021, 2021, 1-7.	0.2	1
22	Whole Exome Sequencing detects PYGM variants in two adults with McArdle disease. <i>Journal of Physical Education and Sports Management</i> , 2022, , mcs.a006173.	1.2	1