

# 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

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g-index

22  
all docs

22  
docs citations

22  
times ranked

1809  
citing authors

#	ARTICLE	IF	CITATIONS
1	Whole Exome Sequencing detects PYGM variants in two adults with McArdle disease. Journal of Physical Education and Sports Management, 2022, , mcs.a006173.	1.2	1
2	Whole-Exome Sequencing Identifies a Novel POLG Frameshift Variant in an Adult Patient Presenting with Progressive External Ophthalmoplegia and Mitochondrial DNA Depletion. Case Reports in Genetics, 2021, 2021, 1-7.	0.2	1
3	Leber hereditary optic neuropathy plus dystonia, and transverse myelitis due to double mutations in MT-ND4 and MT-ND6. Journal of Neurology, 2020, 267, 823-829.	3.6	17
4	Compound heterozygous inheritance of two novel COQ2 variants results in familial coenzyme Q deficiency. Orphanet Journal of Rare Diseases, 2020, 15, 320.	2.7	8
5	Detection of mitochondrial DNA (mtDNA) mutations. Methods in Cell Biology, 2020, 155, 383-400.	1.1	5
6	Characterization of the human homozygous R182W POLG2 mutation in mitochondrial DNA depletion syndrome. PLoS ONE, 2018, 13, e0203198.	2.5	11
7	Novel recessive mutations in COQ4 cause severe infantile cardiomyopathy and encephalopathy associated with CoQ 10 deficiency. Molecular Genetics and Metabolism Reports, 2017, 12, 23-27.	1.1	27
8	A Novel SUCLA2 Mutation Presenting as a Complex Childhood Movement Disorder. Journal of Child Neurology, 2017, 32, 246-250.	1.4	15
9	A De Novo Mutation in MTND6 Causes Generalized Dystonia in 2 Unrelated Children. Child Neurology Open, 2016, 3, 2329048X1562793.	1.1	4
10	Whole exome sequencing identifies a homozygous POLG2 missense variant in an infant with fulminant hepatic failure and mitochondrial DNA depletion. European Journal of Medical Genetics, 2016, 59, 540-545.	1.3	21
11	Functional cellular analyses reveal energy metabolism defect and mitochondrial DNA depletion in a case of mitochondrial aconitase deficiency. Molecular Genetics and Metabolism, 2016, 118, 28-34.	1.1	32
12	Lack of aprataxin impairs mitochondrial functions via downregulation of the APE1/NRF1/NRF2 pathway. Human Molecular Genetics, 2015, 24, 4516-4529.	2.9	23
13	Lack of Effect of Coenzyme Q10 on Doxorubicin Cytotoxicity in Breast Cancer Cell Cultures. Integrative Cancer Therapies, 2012, 11, 243-250.	2.0	32
14	Muscle Phosphoglycerate Mutase Deficiency Revisited. Archives of Neurology, 2009, 66, 394-8.	4.5	40
15	Detection of Mutations in mtDNA. Methods in Cell Biology, 2007, 80, 437-463.	1.1	15
16	Identification of a novel D109Y mutation in Cu/Zn superoxide dismutase (sod1) gene associated with amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2007, 254, 17-21.	0.6	9
17	A Mutation in Para-Hydroxybenzoate-Polyprenyl Transferase (COQ2) Causes Primary Coenzyme Q10 Deficiency. American Journal of Human Genetics, 2006, 78, 345-349.	6.2	322
18	Leigh Syndrome with Nephropathy and CoQ10 Deficiency Due to decaprenyl diphosphate synthase subunit 2 (PDSS2) Mutations. American Journal of Human Genetics, 2006, 79, 1125-1129.	6.2	359

#	ARTICLE	IF	CITATIONS
19	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
20	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
21	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
22	Chronic levodopa administration alters cerebral mitochondrial respiratory chain activity. <i>Annals of Neurology</i> , 1993, 34, 715-723.	5.3	144