

# Emna Mkaouar-Rebai

## List of Publications by Year in descending order

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Version: 2024-02-01

19  
papers

190  
citations

1163117

8  
h-index

1058476

14  
g-index

19  
all docs

19  
docs citations

19  
times ranked

303  
citing authors

#	ARTICLE	IF	CITATIONS
1	The heteroplasmic m.14709T>C mutation in the tRNAGlu gene in two Tunisian families with mitochondrial diabetes. <i>Journal of Diabetes and Its Complications</i> , 2010, 24, 270-277.	2.3	30
2	Mutational analysis of the mitochondrial 12S rRNA and tRNASer(UCN) genes in Tunisian patients with nonsyndromic hearing loss. <i>Biochemical and Biophysical Research Communications</i> , 2006, 340, 1251-1258.	2.1	22
3	Mutational analysis of the mitochondrial tRNA <sup>Leu</sup> (UUR) gene in Tunisian patients with mitochondrial diseases. <i>Biochemical and Biophysical Research Communications</i> , 2007, 355, 1031-1037.	2.1	21
4	The mitochondrial ND1 m.3337G>A mutation associated to multiple mitochondrial DNA deletions in a patient with Wolfram syndrome and cardiomyopathy. <i>Biochemical and Biophysical Research Communications</i> , 2011, 411, 247-252.	2.1	19
5	A maternally inherited diabetes and deafness patient with the 12S rRNA m.1555A>G and the ND1 m.3308T>C mutations associated with multiple mitochondrial deletions. <i>Biochemical and Biophysical Research Communications</i> , 2013, 431, 670-674.	2.1	17
6	A case of Kearns-Sayre syndrome with two novel deletions (9.768 and 7.253kb) of the mtDNA associated with the common deletion in blood leukocytes, buccal mucosa and hair follicles. <i>Mitochondrion</i> , 2010, 10, 449-455.	3.4	13
7	Clinical, Molecular, and Computational Analysis in two cases with mitochondrial encephalomyopathy associated with SUCLG1 mutation in a consanguineous family. <i>Biochemical and Biophysical Research Communications</i> , 2018, 495, 1730-1737.	2.1	9
8	Screening of mitochondrial mutations in Tunisian patients with mitochondrial disorders: An overview study. <i>Mitochondrial DNA</i> , 2013, 24, 163-178.	0.6	8
9	A novel MT-CO2 m.8249G>A pathogenic variation and the MT-TW m.5521G>A mutation in patients with mitochondrial myopathy. <i>Mitochondrial DNA</i> , 2014, 25, 394-399.	0.6	8
10	Whole mitochondrial genome screening in two families with hearing loss: detection of a novel mutation in the 12S rRNA gene. <i>Bioscience Reports</i> , 2010, 30, 405-411.	2.4	7
11	Cytochrome C oxydase deficiency: SURF1 gene investigation in patients with Leigh syndrome. <i>Biochemical and Biophysical Research Communications</i> , 2018, 497, 1043-1048.	2.1	7
12	First description of a novel mitochondrial mutation in the MT-TI gene associated with multiple mitochondrial DNA deletion and depletion in family with severe dilated mitochondrial cardiomyopathy. <i>Biochemical and Biophysical Research Communications</i> , 2018, 497, 1049-1054.	2.1	6
13	A whole mitochondrial genome screening in a MELAS patient: A novel mitochondrial tRNA <sup>Val</sup> mutation. <i>Biochemical and Biophysical Research Communications</i> , 2011, 407, 747-752.	2.1	5
14	A novel MT-CO1 m.6498C>A variation associated with the m.7444G>A mutation in the mitochondrial COI/tRNASer(UCN) genes in a patient with hearing impairment, diabetes and congenital visual loss. <i>Biochemical and Biophysical Research Communications</i> , 2013, 430, 585-591.	2.1	5
15	Mutations in aARS genes revealed by targeted next-generation sequencing in patients with mitochondrial diseases. <i>Molecular Biology Reports</i> , 2020, 47, 3779-3787.	2.3	5
16	A Tunisian patient with Pearson syndrome harboring the 4.977 kb common deletion associated to two novel large-scale mitochondrial deletions. <i>Biochemical and Biophysical Research Communications</i> , 2011, 411, 381-386.	2.1	4
17	Do GSTM1 and GSTT1 polymorphisms influence the risk of developing mitochondrial diseases in a Tunisian population?. <i>Environmental Science and Pollution Research</i> , 2018, 25, 5779-5787.	5.3	4
18	Potential dysfunctional effects of synonymous variants: Insights from an exhaustive in silico analysis of the ABCB4 gene. <i>Annals of Human Genetics</i> , 2018, 82, 457-468.	0.8	0

#	ARTICLE	IF	CITATIONS
19	Mitochondrial disease patients with novel ND4 12058A→C and ND1 m.3911A→G variations: implications for a role in the phenotype following a bioinformatic investigation. Molecular Biology Reports, 2021, 48, 4373-4382.	2.3	0