The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NC_012920.1(MT-CO2):m.7724A>T

Version: 1.1

CA414793459

692767 (ClinVar)

Gene: MT-CO2 (HGNC:4513)
Condition: mitochondrial disease (MONDO:0044970)
Inheritance Mode: Mitochondrial inheritance
UUID: 430ef50b-7baa-4cee-aa37-2de13282ad00
Approved on: 2023-07-24
Published on: 2023-08-02

HGVS expressions

NC_012920.1:m.7724A>T
J01415.2:m.7724A>T
ENST00000361739.1:c.139A>T

Uncertain Significance

Met criteria codes 1

BP4

Not Met criteria codes 6

PM6 PM2 PS2 PS3 PS4 PP1

Evidence Links 0

Expert Panel

Mitochondrial Diseases VCEP

Criteria Specification Information

Criteria Specification: *ClinGen Mitochondrial Disease Nuclear and Mitochondrial Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 1_mtDNA*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Mitochondrial Diseases VCEP

The m.7724A>T variant in MT-CO2 was reviewed by the Mitochondrial Disease Nuclear and Mitochondrial Variant Curation Expert Panel on July 24, 2023. There are no individuals or families with this variant reported in the medical literature to our knowledge. There are several occurrences in population databases. This variant is present in 0.031% of individuals in GenBank MITOMAP sequences, in 0.039% of individuals in gnomAD v3.1.2 (homoplasmic in all individuals), and in 0.088% of individuals in the Helix dataset (homoplasmic in all individuals). The computational predictor APOGEE gives a consensus rating of neutral with a score of 0.3 (Min=0, Max=1), which predicts no damaging effect on gene function (BP4). There are no cybrid, single fiber, or other studies reported for this variant. In summary, this variant meets criteria to be classified as uncertain significance for primary mitochondrial disease inherited in a mitochondrial manner. This classification was approved by the NICHD/NINDS U24 ClinGen Mitochondrial Disease Variant Curation Expert Panel on July 24, 2023. Mitochondrial DNA-specific ACMG/AMP criteria applied (PMID: 32906214): BP4.

Met criteria codes

BP4	✔	The computational predictor APOGEE gives a consensus rating of neutral with a score of 0.3 (Min=0, Max=1), evidence that does not predict a damaging effect on gene function (BP4).

Not Met criteria codes

PM6	✖	There are no reported de novo occurrences of this variant to our knowledge.
PM2	✖	This variant is present in population databases (Mitomap's 61,168 sequences: AF=0.031%; Helix's 195,983 sequences: AF=0.088%; and gnomAD v3.1.2: AF=0.039%). All occurrences in these databases are homoplasmic.
PS2	✖	There are no reported de novo occurrences of this variant to our knowledge.
PS3	✖	There are no cybrids, single fiber studies, or other functional assays reported on this variant.
PS4	✖	There are no individuals with this variant reported in the medical literature to our knowledge.
PP1	✖	There are no reports of large families with this variant segregating with disease.

Curation History

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