Recombinant Mouse ACO2 / Aconitase 2 Protein (His & GST tag)

Cat.NO.: TP06734

3th Edition

Synonyms: Aco-2;Aco3;D10Wsu183e

Description:A homozygous missense mutation was identified in the ACO2 gene (c.124T>G p.Phe414Val) that segregated with HSP complicated by intellectual disability and microcephaly. Lymphoblastoid cell lines of homozygous carrier patients revealed significantly decreased activity of the mitochondrial aconitase enzyme and defective mitochondrial respiration. ACO2 encodes mitochondrial aconitase, an essential enzyme in the Krebs cycle. Recessive mutations in this gene have been previously associated with cerebellar ataxia. We found homozygous or compound heterozygous missense and frameshift mutations in the gene encoding mitochondrial aconitase (ACO2), a tricarboxylic acid cycle enzyme, catalysing interconversion of citrate into isocitrate. Unlike wild type ACO2, all mutant ACO2 proteins failed to complement the respiratory growth of a yeast aco1-deletion strain. The study shows that autosomal recessive ACO2 mutations can cause either isolated or syndromic optic neuropathy. This observation identifies ACO2 as the second gene responsible for non-syndromic autosomal recessive optic neuropathies and provides evidence for a genetic overlap between isolated and syndromic forms, giving further support to the view that optic atrophy is a hallmark of defective mitochondrial energy supply.

Form:PBS

Molecular Weight:110 kDa

Sequences: Gln 28-Gln 780

Purity:> 95% by HPLC

Concentration:

Endotoxin Level:<1.0 EU per 1 ug of protein (determined by LAL method)

Storage: Can be stored at +4°C short term (1-2 weeks). For long term storage, aliquot and store at -20°C or -70°C. Avoid repeated freezing and thawing cycles.