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

Catalog Number: PKSM040388

Note: *Centrifuge before opening to ensure complete recovery of vial contents.*

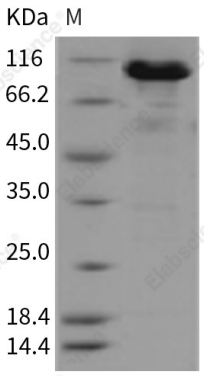
Description

Species	Mouse
Source	Baculovirus-Insect Cells-derived Mouse ACO2/Aconitase 2 protein Gln 28-Gln 780, with an N-terminal His & GST
Calculated MW	110 kDa
Observed MW	100 kDa
Accession	Q99KI0
Bio-activity	Not validated for activity

Properties

Purity	> 90 % as determined by reducing SDS-PAGE.
Endotoxin	< 1.0 EU per μg of the protein as determined by the LAL method.
Storage	Generally, lyophilized proteins are stable for up to 12 months when stored at -20 to -80°C. Reconstituted protein solution can be stored at 4-8°C for 2-7 days. Aliquots of reconstituted samples are stable at < -20°C for 3 months.
Shipping	This product is provided as lyophilized powder which is shipped with ice packs.
Formulation	Lyophilized from sterile 50mM Tris, 100mM NaCl, 10% gly, 0.5mM GSH, pH 8.0
Reconstitution	Please refer to the printed manual for detailed information.

Data



> 90 % as determined by reducing SDS-PAGE.

Background

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.

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