

Aconitase 2 Protein, Human, Recombinant (His & GST)

General Information

Synonyms:	ACONM;ICRD;aconitase 2, mitochondrial
Protein Construction:	A DNA sequence encoding the human ACO2 (Q99798) (Gln 28-Gln 780) was fused with the N-terminal polyhistidine-tagged GST tag at the N-terminus. Predicted N terminal: Met
Species:	Human
Expression Host:	Baculovirus Insect Cells
Accession:	Q99798
Molecular Weight:	110 kDa (predicted); 110 kDa (reducing conditions)

QC Testing

Biological Activity:	Activity testing is in progress. It is theoretically active, but we cannot guarantee it. If you require protein activity, we recommend choosing the eukaryotic expression version first.
Purity:	> 85 % as determined by SDS-PAGE
Endotoxin:	< 1.0 EU/μg of the protein as determined by the LAL method.
Formulation:	Lyophilized from a solution filtered through a 0.22 μm filter, containing 20 mM Tris, 500 mM NaCl, 10% glycerol, pH 8.0, 0.3 mM DTT. Typically, a mixture containing 5% to 8% trehalose, mannitol, and 0.01% Tween 80 is incorporated as a protective agent before lyophilization.

Preparation and Storage

Reconstitution:
A Certificate of Analysis (CoA) containing reconstitution instructions is included with the products. Please refer to the CoA for detailed information.

Stability & Storage:

It is recommended to store recombinant proteins at -20°C to -80°C for future use. Lyophilized powders can be stably stored for over 12 months, while liquid products can be stored for 6-12 months at -80°C. For reconstituted protein solutions, the solution can be stored at -20°C to -80°C for at least 3 months. Please avoid multiple freeze-thaw cycles and store products in aliquots.

Actual storage temperature shall be subject to the COA.

Shipping:

In general, lyophilized powders are shipped with blue ice, while solutions are shipped with dry ice.

Protein Background

A homozygous missense mutation was identified in the ACO2 gene (c.1240T>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.

Reference

Robbins AH, et al. (1989) The structure of aconitase. *Proteins*. 5 (4): 289-312.

Lauble H, et al. (1992) Crystal structures of aconitase with isocitrate and nitroisocitrate bound. *Biochemistry*. 31 (10): 2735-48.

Robbins AH, et al. (1989) Structure of activated aconitase: formation of the 4Fe-4S cluster in the crystal. *Proc Natl Acad Sci*. 86 (10): 3639-43.

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