Recombinant Protein Technical Manual

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

RPES3161

Product Data:

Product SKU: RPES3161

Size: 20µg

Species: Mouse

Expression host: Baculovirus-Insect Cells

Uniprot: Q99KI0

Protein Information:

Molecular Mass: 110 kDa

AP Molecular Mass: 100 kDa

Tag: N-His-GST

Bio-activity:

Purity: > 90 % as determined by SDS-PAGE

Endotoxin: < 1.0 EU per µg of the protein as determined by the LAL method.

Storage: 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℃ for 2-7 days. Aliquots of reconstituted samples are stable at < -20℃ 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.

Application:

Synonyms: Aco-2;Aco3;D10Wsu183e

Immunogen Information:

Sequence: Gln 28-Gln 780

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.