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

产品货号: TP06734

第三版

别名: Aco-2;Aco3;D10Wsu183e

描述: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.

配方:PBS

分子量:110 kDa

序列: Gln 28-Gln 780

纯度:> 95% by HPLC

浓度:

内毒素:<1.0 EU per 1 ug of protein (determined by LAL method)

存储: +4 ° C 保存 (1-2 周). 长期保存在-20 ° C或者-70 ° C. 避免反复冻融.