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MMADHC Human

Methylmalonic Aciduria cblD type, with Homocystinuria Human Recombinant RCP1376

Product Overview

Name	MMADHC Human
Description	
Methylmalonic Aciduria cblD type, with Homocystinuria Human Recombinant	
Accession (Primary)	Q9H3L0
Synonyms	
Chromosome 2 Open Reading Frame 25, Methylmalonic Aciduria (Cobalamin Deficiency) CblD Type With Homocystinuria, Methylmalonic Aciduria And Homocystinuria Type D Protein Mitochondrial, Protein C2orf25 Mitochondrial, CL25022, C2orf25, cblD.	
Introduction	
MMADHC is a mitochondrial protein which takes part in an early step of vitamin B1 2 metabolism. Vitamin B12 (cobalamin) is vital for regular development and existence in humans. Mutations in MMADHC can result in methylmalonic aciduria and homocystinuria type cblD, a cobalamin metabolism syndrome which is characterized by decreased levels of the coenzymes methylcobalamin and adenosylcobalamin.	
Source	
Escherichia Coli.	
Physical Appearance	
Sterile Filtered clear solution.	
Formulation	
The MMADHC solution (1mg/ml) contains 20mM Tris-HCl buffer (pH 8.0), 0.15M NaCl, 1mM DTT and 10% glycerol.	
Stability	
Store at 4°C if entire vial will be used within 2-4 weeks. Store, frozen at -20°C for longer periods of time. For long term storage it is recommended to add a carrier protein (0.1% HSA or BSA). Avoid multiple freeze-thaw cycles.	
Purity	
Greater than 90% as determined by SDS-PAGE.	
Amino acid sequence	
MGSSHHHHHH SSGLVPRGSH MGS SDESHVA AAPPDICSRT VWPDETMGPF GPQDQRFQLP GNIGFDCHLN GTASQKKSLV HKTLPDVLAE PLSSERHEFV MAQYVNEFQG NDAPVEQEIN SAETYFESAR VECAIQTCPE LLRKDFESLF PEVANGKLMI LTVTQKTKND MTVWSEEVEI EREVLLEKFI NGAKEICYAL RAEGYWADFI	

DPSSGLAFFG PYTNNTLFET DERYRHLGFS VDDLGCCKVI RHSLWGTHVV VGSIFTNATP DSHIMKKLSG N

Precautions

MMADHC Human is for research use only and not for use in diagnostic or therapeutic procedures.

Target Information: (Q9H3L0)