

Recombinant Human MMADHC protein ab162126

1 Image

Description	
Product name	Recombinant Human MMADHC protein
Expression system	Wheat germ
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MANVLCNRARLVSYLPGFCSLVKRVVNPKAFSTAGSSGS DESHVAAAPPD ICSRTVWPDETMGPFQDQRFQLPGNIGFDCHLNGTAS QKKSLVHKTL DVLAEPLSSERHEFVMAQYVNEFQGNDAPVEQEINSAET YFESARVECAI QTCPELLRKDFESLFPEVANGKLMILTVTQTKNDMTVWS EEVEIEREVL LEKFINGAKEICYALRAEGYWADFIDPSSGLAFFGPYTNNT LFETDERYR HLGFSVDDLGCCKVIRHSLWGTHVVVGSIFTNATPD SHIM KKLSGN
Amino acids	1 to 296
Tags	GST tag N-Terminus

Specifications	
Our <b>Abpromise guarantee</b> covers the use of <b>ab162126</b> in the following tested applications.	
The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.	
Applications	ELISA Western blot
Form	Liquid
Additional notes	This product was previously labelled as C2ORF25.

## Preparation and Storage

### Stability and Storage

Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles.

pH: 8.00

Constituents: 0.31% Glutathione, 0.79% Tris HCl

## General Info

### Function

Involved in cobalamin metabolism.

### Tissue specificity

Widely expressed at high levels.

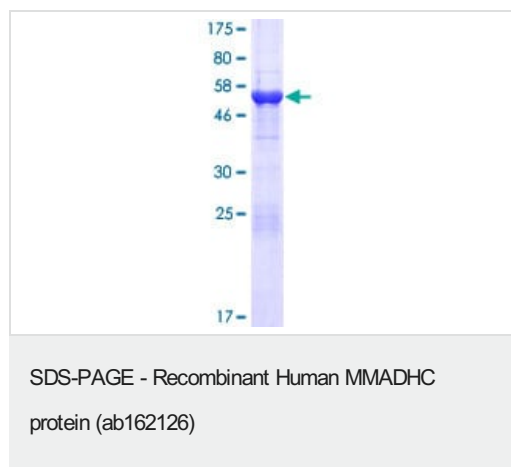
### Involvement in disease

Defects in MMADHC are the cause of methylmalonic aciduria and homocystinuria type cblD (MMAHCD) [MIM:277410]. A disorder of cobalamin metabolism characterized by decreased levels of the coenzymes adenosylcobalamin (AdoCbl) and methylcobalamin (MeCbl). Clinical features include developmental delay, hyponatremia, mental retardation, seizures, megaloblastic anemia. Some patients manifest combined methylmalonic aciduria and homocystinuria (referred to as cblD original), some have only isolated homocystinuria (cblD variant 1), and others have only methylmalonic aciduria (cblD variant 2).

### Cellular localization

Mitochondrion.

## Images



ab162126 on a 12.5% SDS-PAGE stained with Coomassie Blue.

**Please note:** All products are "FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC PROCEDURES"

## Our Abpromise to you: Quality guaranteed and expert technical support

- Replacement or refund for products not performing as stated on the datasheet
- Valid for 12 months from date of delivery
- Response to your inquiry within 24 hours
- We provide support in Chinese, English, French, German, Japanese and Spanish
- Extensive multi-media technical resources to help you
- We investigate all quality concerns to ensure our products perform to the highest standards

If the product does not perform as described on this datasheet, we will offer a refund or replacement. For full details of the Abpromise, please visit <https://www.abcam.com/abpromise> or contact our technical team.

#### **Terms and conditions**

---

- Guarantee only valid for products bought direct from Abcam or one of our authorized distributors