

## Human recombinant MTM1 protein, AF

Catalog Number: PROTQ13496

### Overview

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| Product Name | Human recombinant MTM1 protein, AF                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               |
| Description  | Myotubularin is a protein that plays a crucial role in cellular processes and is primarily associated with a rare genetic disorder called myotubular myopathy (MTM). This disorder primarily affects skeletal muscles and is characterized by severe muscle weakness and impaired motor function. Myotubularin is an enzyme that regulates the levels of a lipid molecule called phosphatidylinositol 3-phosphate (PI3P) within cells. PI3P is involved in various cellular functions, including membrane trafficking and signaling pathways. Mutations in the gene encoding myotubularin lead to a deficiency or loss of its enzymatic activity, disrupting PI3P metabolism and causing the development of MTM. |
| Size         | 5ug,20ug,100ug                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
| Tag          | His Tag (N-term)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
| Form         | Lyophilized                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      |
| Source       | Escherichia coli                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
| Formulation  | The protein was lyophilized from a 0.2 um filtered solution containing 1X PBS, pH 7.4. If you have any concerns or special requirements, please confirm with us.                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |

### Storage

Lyophilized protein should be stored at -20°C for 1 year. Upon reconstitution, store at 2°C to 8°C for up to 1 week. Further dilute in a buffer containing a carrier protein or stabilizer (e.g. 0.1% BSA, 10%FBS, 5%HSA or 5% trehalose solution), protein aliquots should be stored at -20°C or -80°C for 3-6 months. Avoid repeated freeze/thaw cycles.

### Purity

>95% as determined by SDS-PAGE.

### Molecular weight

The protein has a calculated MW of 70.75 kDa. The protein migrates as 60 kDa under reducing condition (SDS-PAGE analysis).

### Activity

Testing in process

### Endotoxin

<1 EU per 1 ug of the protein by the LAL method.

### **Amino Acid Sequence**

A DNA sequence encoding Human MTM1 Protein (#Q13496)(Met1-Phe603) was expressed with polyhistidine tag at the N-terminus.

### **Reconstitution**

Centrifuge at 3000 rpm for 5 mins before opening. It is recommended to reconstitute the lyophilized protein in sterile H<sub>2</sub>O to a concentration not less than 100 ug/mL and incubate the stock solution at room temperature for at least 20 mins to ensure sufficient re-dissolved. Do Not Vortex!

Vigorous shaking may impair the biological activity of the protein.

## Usage

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