abcam

Product datasheet

Recombinant Human Telethonin protein ab160096

1 Image

Overview

Product name	Recombinant Human Telethonin protein
Protein length	Full length protein

Description

Nature	Recombinant
Source	Wheat germ
Amino Acid Sequence	
Species	Human
Sequence	MATSELSCEVSEENCERREAFWAEWKDLTLSTRPEE GCSLHEEDTQRHET YHQQGQCQVLVQRSPWLMMRMGILGRGLQEYQLPYQ RVLPLPIFTPAKMG ATKEEREDTPIQLQELLALETALGGQCVDRQEVAEITK QLPPVVPVSKPG ALRRSLSRSMSQEAQRG
Amino acids	1 to 167
Tags	GST tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab160096** 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	Protein concentration is above or equal to 0.05 mg/ml.

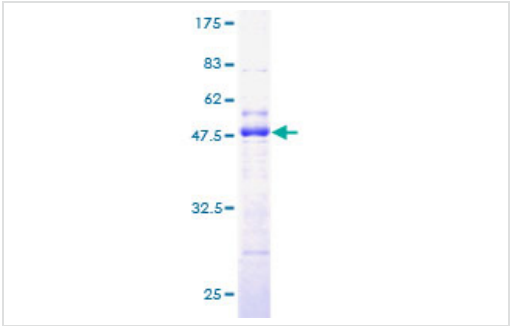
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Function	Muscle assembly regulating factor. Mediates the antiparallel assembly of titin (TTN) molecules at the sarcomeric Z-disk.
Tissue specificity	Heart and skeletal muscle.
Involvement in disease	Defects in TCAP are a cause of cardiomyopathy familial hypertrophic (CMH) [MIM:192600]; also designated FHC or HCM. Familial hypertrophic cardiomyopathy is a hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. Defects in TCAP are a cause of limb-girdle muscular dystrophy type 2G (LGMD2G) [MIM:601954]. LGMD2G is an autosomal recessive degenerative myopathy characterized by proximal and distal muscle weakness and atrophy in the limbs, dystrophic changes on muscle biopsy, and absence of telethonin. Cardiac muscle is involved in a subset of patients. Defects in TCAP are the cause of cardiomyopathy dilated type 1N (CMD1N) [MIM:607487]. Dilated cardiomyopathy is a disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death.
Cellular localization	Cytoplasm > myofibril > sarcomere.

Images



SDS-PAGE - Recombinant Human Telethonin protein (ab160096)

ab160096 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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