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Product datasheet

Recombinant Human LMBR1 protein ab163818

1 Image

Overview

Product name	Recombinant Human LMBR1 protein
Protein length	Full length protein

Description

Nature	Recombinant
Source	Wheat germ
Amino Acid Sequence	
Species	Human
Sequence	MEGQDEVSAREQHFHSQVRESTICFLLFAILYVVSYFIIT RYKRKSDEQE DEDAIVNRISLFLSTFTLAVSAGAVLLLPFSIISNEILLSF PQNYYIQWL NGSLIHGLWNLASLFSNLCLFVLMPFAFFFLESEGFAG LKKGIRARILET LVMLLLLALLILGIVWVASALIDNDAASMESLYDLWEFY LPYLYSCISLM GCLLLLLCTPVGLSRMFTVMGQLLVKPTILEDLDEQIYII TLEEEALQRR LNGLSSSVEYNIMELEQELENVKTLKTKLERRKKASAW ERNLVYPAVMVL LLIETSISVLLVACNILCLLVDETAMPKGTRGPGIGNASL STFGFVGAAL EIILIFYLMVSSVVGFYSLRFFGNFTPKKDDTTMTKIIGNC VSILVLSSA LPVMSRTLGITRFDLLGDFGRFNWLGNFYIVLSYNLLFAI VTTLCLVRKF TSAVREELFKALGLHKLHLPNTSRDSETAKPSVNGHQ KAL
Amino acids	1 to 490
Tags	GST tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab163818** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	Western blot ELISA
Form	Liquid
Additional notes	Protein concentration is above or equal to 0.05 mg/ml.

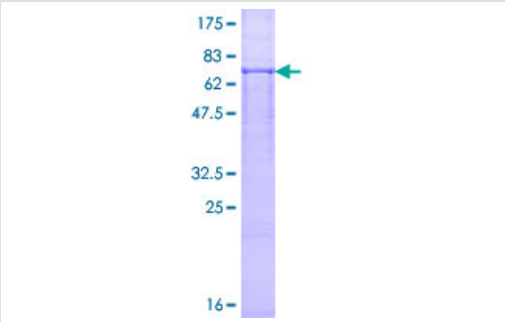
Preparation and Storage

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	Putative membrane receptor.
Tissue specificity	Widely expressed with strongest expression in heart and pancreas.
Involvement in disease	Defects in LMBR1 are associated with preaxial polydactyly type 2 (PPD2) [MIM:174500]; also known as polydactyly of triphalangeal thumb. Polydactyly consists of duplication of the distal phalanx. The thumb in PPD2 is usually opposable and possesses a normal metacarpal. The mutations do not change the normal expression of LMBR1, but alter the expression of SHH by disrupting a long-range, cis-regulatory element of that gene. Defects in LMBR1 are the cause of acheiropody (ACHP) [MIM:200500]. Acheiropody is a very rare condition characterized by bilateral congenital amputations of the hands and feet. The specific malformative phenotype consists of a complete amputation of the distal epiphysis of the humerus, amputation of the tibial diaphysis, and aplasia of the radius, ulna, fibula, and of all the bones of the hands and feet. This syndrome of autosomal recessive inheritance has only been observed in Brazil so far. Defects in LMBR1 are a cause of syndactyly type 4 (SDYT4) [MIM:186200]. SDYT4 is a very rare congenital distal limb malformation characterized by complete bilateral syndactyly (involving all digits 1 to 5). A frequent association with polydactyly (with six metacarpals and six digits) has been reported. Feet are affected occasionally. The condition is inherited as an autosomal dominant trait.
Sequence similarities	Belongs to the LIMR family.
Cellular localization	Membrane.

Images



SDS-PAGE - Recombinant Human LMBR1 protein (ab163818)

ab163818 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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