LMBR1 Antibody (Center) Blocking Peptide

PRODUCT OVERVIEW

Product Name	LMBR1, Blocking Peptide
Full Product Name	LMBR1 Antibody (Center) Blocking Peptide
Product Gene Name	LMBR1 blocking peptide
Product Synonym Gene Name	C7orf2; DIF14
Product Synonym Names	Limb region 1 protein homolog; Differentiation-related gene 14 protein; LMBR1; C7orf2; DIF14
Form/Format	Synthetic peptide was lyophilized with 100% acetonitrile and is supplied as a powder. Reconstitute with 0.1 ml DI water for a final concentration of 1 mg/ml.
Preparation and Storage	Maintain refrigerated at 2-8 degree C for up to 6 months. For long term storage store at -20 degree C.
Antibody/Peptide Pairs	LMBR1 peptide (MBS9222455) is used for blocking the activity of LMBR1 antibody (MBS9210672)
Molecular Weight	59,458 Da
OMIM	135750
Cellular Location	Membrane; Multi-pass membrane protein
Tissue Location	Widely expressed with strongest expression in heart and pancreas.
Related Product Information for LMBR1 blocking peptide	MBS9222455 is a blocking peptide targeting LMBR1. Also known as LMBR1 Antibody (Center) Blocking Peptide. Putative membrane receptor.
Other Notes	Small volumes of LMBR1 blocking peptide vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply.

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for LMBR1. It may not necessarily be applicable to this product.

NCBI GI #	74730878
NCBI GeneID	64327
NCBI Accession #	Q8WVP7.1
UniProt Primary Accession #	Q8WVP7
UniProt Secondary Accession #	Q8N3E3; Q96QZ5; Q9H5N0; Q9HAG9; Q9UDN5; Q9Y6U2; A4D242
UniProt Related Accession #	Q8WVP7
NCBI Official Full Name	Limb region 1 protein homolog
NCBI Official Synonym Full Names	limb development membrane protein 1
NCBI Official Symbol	LMBR1
NCBI Official Synonym Symbols	LSS; TPT; ZRS; ACHP; PPD2; THYP; DIF14; C7orf2
UniProt Protein Name	Limb region 1 protein homolog
UniProt Synonym Protein Names	Differentiation-related gene 14 protein
Protein Family	Limb region 1 protein

UniProt Gene Name	LMBR1
UniProt Synonym Gene Names	C7orf2; DIF14
UniProt Entry Name	LMBR1_HUMAN
NCBI Protein Information	limb region 1 protein homolog
NCBI Summary for LMBR1	This gene encodes a member of the LMBR1-like membrane protein family. Another member of this protein family has been shown to be a lipocalin transmembrane receptor. A highly conserved, cis-acting regulatory module for the sonic hedgehog gene is located within an intron of this gene. Consequently, disruption of this genic region can alter sonic hedgehog expression and affect limb patterning, but it is not known if this gene functions directly in limb development. Mutations and chromosomal deletions and rearrangements in this genic region are associated with acheiropody and preaxial polydactyly, which likely result from altered sonic hedgehog expression. [provided by RefSeq, Jul 2008]
UniProt Comments for LMBR1	LMBR1: Putative membrane receptor. Defects in LMBR1 are associated with preaxial polydactyly type 2 (PPD2); 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). 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 (SDTY4). SDTY4 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. Belongs to the LIMR family. 3 isoforms of the human protein are produced by alternative splicing. Protein type: Membrane protein, multi-pass; Membrane protein, integral; Receptor, misc. Chromosomal Location of Human Ortholog: 7q36 Disease: Acheiropody; Laurin-sandrow Syndrome; Polydactyly, Preaxial Ii; Syndactyly, Type Iv; Tibia, Hypoplasia Or Aplasia Of, With Polydactyly
Research Articles on LMBR1	**1.** A novel ZRS mutation found in the Mexican population, 402C>T, suggests that a dosage effect exists for this mutation.
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