abcam

Product datasheet

Human FHL1 peptide ab45621

Overview

Product name Human FHL1 peptide

Description

Nature Synthetic

Amino Acid Sequence

Species Human

Sequence C-NKRFVFHNEQVY

Amino acids 261 to 272

Specifications

Our Abpromise guarantee covers the use of **ab45621** in the following tested applications.

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

Applications Blocking - Blocking peptide for Anti-FHL1 antibody (ab23937)

Form Liquid

Preparation and Storage

Stability and Storage Shipped at 4°C. Upon delivery aliquot and store at -20°C. Avoid freeze / thaw cycles.

General Info

Function May have an involvement in muscle development or hypertrophy.

Tissue specificity Isoform 1 is highly expressed in skeletal muscle and to a lesser extent in heart, placenta, ovary, prostate, testis, small intestine, colon and spleen. Expression is barely detectable in brain, lung, liver, kidney, pancreas, thymus and peripheral blood leukocytes. Isoform 2 is expressed in brain, skeletal muscle and to a lesser extent in heart, colon, prostate and small intestine. Isoform 3 is expressed in testis, heart and skeletal muscle.

Involvement in disease Defects in FHL1 are the cause of X-linked dominant scapuloperoneal myopathy (SPM) [MIM:300695]. Scapuloperoneal syndrome (SPS) was initially described more than 120 years ago by Jules Broussard as 'une forme hereditaire d'atrophie musculaire progressive' beginning

	in the lower legs and affecting the shoulder region earlier and more severely than distal arm. The etiology of this condition remains unclear. Defects in FHL1 are the cause of X-linked myopathy with postural muscle atrophy (XMPMA) [MIM:300696]. Myopathies are inherited muscle disorders characterized by weakness and atrophy of voluntary skeletal muscle, and several types of myopathy also show involvement of cardiac muscle. XMPMA is a distinct form of adult-onset X-linked recessive myopathy with several features in common with other myopathies, but the presentation of a pseudoathletic phenotype, scapuloperoneal weakness, and bent spine is unique and might render the clinical phenotype distinguishable from other myopathies. Defects in FHL1 are the cause of X-linked severe early-onset reducing body myopathy (RBM) [MIM:300717]. RBM is a rare muscle disorder causing progressive muscular weakness and characteristic intracytoplasmic inclusions in myofibers. Clinical presentations of RBM have ranged from early onset fatal to childhood onset to adult onset cases. Defects in FHL1 are the cause of X-linked childhood-onset reducing body myopathy (CO-RBM) [MIM:300718]. This disorder is allelic to severe early-onset reducing body myopathy (RBM) [MIM:300717].
Sequence similarities	Contains 3 LIM zinc-binding domains.
Developmental stage	Elevated levels during postnatal muscle growth.
Cellular localization	Cytoplasm; Cytoplasm. Nucleus and Nucleus. Cytoplasm > cytosol. Predominantly nuclear in myoblasts but is cytosolic in differentiated myotubes.

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