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Rabbit Anti-Myotilin antibody

SL6479R

Product Name:	Myotilin
Chinese Name:	肌收缩蛋白MYOT抗体
Alias:	57 kDa cytoskeletal protein; LGMD 1; LGMD1; Myofibrillar titin like Ig domains protein; Myofibrillar titin-like Ig domains protein; MYOT; MYOTI_HUMAN; Myotilin; Titin immunoglobulin domain protein; TTID; TTID protein.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Pig,Cow,Horse,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=1:50-200（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	55kDa
Cellular localization:	cytoplasmicThe cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human Myotilin:121-200/498
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	Myotilin, a sarcomeric protein that is encoded by the gene mapping to human chromosome 5q31, binds to a-actinin and is localized in the Z-line of myofibrils. Myotilin is expressed in skeletal and cardiac muscle, and it co-localizes with a-actinin in the sarcomeric I-bands where it directly interacts with a-actinin. Defects in the myotilin gene are reported to cause a form of autosomal dominant limb-girdle muscular

	dystrophy (LGMD). Symptoms of adult onset LGMD are progressive weakness of the hip and shoulder girdles as well as a distinctive dysarthric pattern of speech. The muscle of affected individuals with LGMD shows degeneration of myofibers, variations in fiber size, fiber splitting, centrally located myonuclei and an enhanced number of autophagic vesicles. **Function:** Component of a complex of multiple actin cross-linking proteins. Involved in the control of myofibril assembly and stability at the Z lines in muscle cells. **Subunit:** Homodimer. Interacts with ACTA1, ACTN1, FLNA, FLNB, FLNC and MYOZ2. Interacts with the C-terminal region of MYOZ1. **Subcellular Location:** Cell membrane, sarcolemma. Cytoplasm, cytoskeleton. Cytoplasm, myofibril, sarcomere, Z line. **Tissue Specificity:** Expressed in skeletal muscle (at protein level). Expressed in skeletal muscle, heart, bone marrow and thyroid gland. **DISEASE:** Defects in MYOT are the cause of limb-girdle muscular dystrophy type 1A (LGMD1A). LGMD1A is an autosomal dominant degenerative myopathy with onset within a mean age of 28 years. LGMD1A is characterized by progressive skeletal muscle weakness of the hip and shoulder girdles, later progressing to include distal weakness, as well as a distinctive dysarthric pattern of speech. Affected muscle exhibits disorganization and streaming of the Z-line. Defects in MYOT are the cause of myopathy myofibrillar type 3 (MFM3) [MIM:609200]. A neuromuscular disorder characterized by progressive skeletal muscle weakness greater distally than proximally, tight heel cords, hyporeflexia, cardiomyopathy and peripheral neuropathy in some patients. Affected muscle exhibits disorganization and streaming of the Z-line, presence of large hyaline structures, excessive accumulation of myotilin and other ectopically expressed proteins and prominent congophilic deposits. Defects in MYOT are the cause of spheroid body myopathy (SBM) [MIM:182920]. SBM is an autosomal dominant form of myofibrillar myopathy (MFM), characterized by slowly progressing proximal muscle weakness and dysarthric nasal speech. There is no evidence of cardiomyopathy. Muscle biopsy shows spheroid bodies within the type I muscle fibers. **Similarity:** Belongs to the myotilin/palladin family. Contains 2 Ig-like C2-type (immunoglobulin-like) domains.

	SWISS: Q9UBF9 **Gene ID:** 9499 **Database links:** Entrez Gene: 9499Human Entrez Gene: 58916Mouse Entrez Gene: 291605Rat Omim: 604103Human SwissProt: Q9UBF9Human SwissProt: Q9JIF9Mouse Unigene: 84665Human Unigene: 143804Mouse Unigene: 163370Rat **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.