

Rabbit Anti-MTMR14 antibody

SL11217R

Product Name:	MTMR14
Chinese Name:	
Alias: Organism Species: Clonality: Paget Species:	C3orf29; Egg derived tyrosine phosphatase homolog; FLJ11546; FLJ22405; FLJ46453; FLJ90311; HCV NS5A transactivated protein 4 splice variant A binding protein 1; HCV NS5A-transactivated protein 4 splice variant A-binding protein 1; hJumpy; jumpy; MTMR 14; MTMR14; MTMR-14; MTMRE_HUMAN; Myotubularin related protein 14; Myotubularin-related protein 14; NS5ATP4ABP1. Rabbit Polyclonal
React Species.	WB=1.500-2000ELISA=1.500-1000IHC-P=1.400-800IHC-F=1.400-800ICC=1.100-
Applications:	500IF=1:100-500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	72kDa
Cellular localization:	cytoplasmicThe cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human MTMR14:161-260/650
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:	Myotubularin-related protein 14 (MTMR14), also known as Jumpy, is a myotubularin- related phosphoinositol-3-phosphate (PI3P) phosphatase (1). Mutations in the MTMR14 gene have been associated with centronuclear myopathy (1). MTMR14 deficiency in

mice leads to altered calcium homeostasis and muscle disorders (2). MTMR14 has also been shown to play a role in autophagy, a process that is highly regulated by phosphatidylinositides through the type III PI3K, Vps34 (3). MTMR14 was localized to autophagic isolation membranes and early autophagosomes (3). In these studies, MTMR14 inhibited autophagy and mutations of MTMR14 associated with centronuclear myopathy were also defective in autophagy inhibition. In zebrafish, MTMR14 knockdown was shown to increase the number of autophagosomes, suggesting that its activity is associated with an inhibition of autophagy (4).

Function:

Lipid phosphatase which efficiently dephosphorylates phosphatidylinositol 3-phosphate (PtdIns3P) and PtdIns(3,5)P2; inactive toward PtdIns4P, PtdIns(3,4)P2, PtdIns(4,5)P2 and PtdIns(3,4,5)P3.

Subunit:

Belongs to the protein-tyrosine phosphatase family. Non-receptor class myotubularin subfamily.

Subcellular Location:

Cytoplasm. Found in reticular structures and plasma membrane ruffles. Concentrated near the nucleus.

Tissue Specificity:

Expressed in various tissues, including heart, skeletal muscle, placenta, liver, lung, kidney and pancreas.

DISEASE:

Defects in MTMR14 may be a cause of centronuclear myopathy autosomal dominant (ADCNM) [MIM:160150]; also known as autosomal dominant myotubular myopathy. Centronuclear myopathies are congenital muscle disorders characterized by progressive muscular weakness and wasting involving mainly limb girdle, trunk, and neck muscles. It may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life. Ptosis is a frequent clinical feature. The most prominent histopathologic features include high frequency of centrally located nuclei in muscle fibers not secondary to regeneration, radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers.

Similarity:

Belongs to the protein-tyrosine phosphatase family. Non-receptor class myotubularin subfamily.

SWISS:

Q8NCE2

Gene ID:

64419

Database links:

Entrez Gene: 64419Human

Entrez Gene: 97287Mouse

Entrez Gene: 312634Rat

Omim: 611089Human

SwissProt: Q8NCE2Human

SwissProt: Q8VEL2Mouse

Unigene: 475382Human

Unigene: 197816Mouse

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

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Sample:
Lung (Mouse) Lysate at 40 ug
Primary: Anti-MTMR14 (SL11217R) at 1/1000 dilution
Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution
Predicted band size: 72 kD
Observed band size: 72 kD

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