



Rabbit Anti-MTMR2 antibody

SL11733R

Product Name:	MTMR2
Chinese Name:	肌管素相关蛋白2抗体
Alias:	CMT4B; CMT4B1; KIAA1073; MTMR2; MTMR2_HUMAN; Myotubularin related protein 2; Myotubularin-related protein 2.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Pig,Cow,Rabbit,Zebrafish,Sheep,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=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:	73kDa
Cellular localization:	cytoplasmicThe cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human MTMR2:371-450/643
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 and the myotubularin-related proteins (MTMR1-9) belong to a highly conserved family of eukaryotic phosphatases. They are protein tyrosine phosphatases that utilize inositol phospholipids, rather than phosphoproteins, as substrates. MTMR family members hydrolyze both Phosphatidylinositol 3-phosphate (PtdIns3P) and PtdIns(3,5)P2. MTMR2 interacts with MTMR5, an inactive family member that increases the enzymatic activity of MTMR2 and dictates its subcellular localization.

Mutations in MTMR2 cause autosomal recessive Charcot-Marie-Tooth type 4B1 (CMT4B1), which is characterized by reduced nerve conduction velocities, focally folded myelin sheaths and demyelination. MTMR3 and MTMR4 can either interact with each other or self associate. MTMR6 regulates the activity of the calcium-activated potassium channel 3.1. MTMR9 regulates the activity of MTMR7 and MTMR8.

Function:

Phosphatase that acts on lipids with a phosphoinositol headgroup. Has phosphatase activity towards phosphatidylinositol-3-phosphate and phosphatidylinositol-3,5-bisphosphate.

Subunit:

Homooligomer and heterooligomer. Interacts with SBF1 and SBF2.

Subcellular Location:

Cytoplasm. Membrane. Partly associated with membranes.

Post-translational modifications:

Phosphorylation at Ser-58 decreases MTMR2 localization to endocytic vesicular structures.

DISEASE:

Defects in MTMR2 are the cause of Charcot-Marie-Tooth disease type 4B1 (CMT4B1) [MIM:601382]. CMT4B1 is a recessive, severe form of Charcot-Marie-Tooth disease, the most common inherited disorder of the peripheral nervous system. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathy and primary peripheral axonal neuropathy. Demyelinating CMT neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. By convention, autosomal recessive forms of demyelinating Charcot-Marie-Tooth disease are designated CMT4.

Similarity:

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

Contains 1 GRAM domain.

Contains 1 myotubularin phosphatase domain.

SWISS:

Q13614

Gene ID:

8898

Database links:

[Entrez Gene: 536810](#)Cow

[Entrez Gene: 606918](#)Dog

[Entrez Gene: 101149089](#)Gorilla

[Entrez Gene: 100720535](#)Guinea pig

[Entrez Gene: 8898](#)Human

[Entrez Gene: 77116](#)Mouse

[Entrez Gene: 100174264](#)Orangutan

[Entrez Gene: 100518140](#)Pig

[Entrez Gene: 315422](#)Rat

[Entrez Gene: 699703](#)Rhesus monkey

[Entrez Gene: 101123404](#)Sheep

[Olim: 603557](#)Human

[SwissProt: A6QLT2](#)Cow

[SwissProt: Q13614](#)Human

[SwissProt: Q9Z2D1](#)Mouse

[SwissProt: Q5REB9](#)Orangutan

Important Note:

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