

Product name:	Myotubularin Rabbit Polyclonal Antibody
Cat number:	ABN14349
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human Myotubularin. AA range:241-290
Reactivity:	Human,Mouse
Applications:	WB 1:500-1:2000,IHC 1:50-1:300
Molecular Weight:	70kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

This gene encodes a dual-specificity phosphatase that acts on both phosphotyrosine and phosphoserine. It is required for muscle cell differentiation and mutations in this gene have been identified as being responsible for X-linked myotubular myopathy. [provided by RefSeq, Jul 2008], catalytic activity: Protein tyrosine phosphate + H₂O = protein tyrosine + phosphate., caution: The sequence shown here is derived from an Ensembl automatic analysis pipeline and should be considered as preliminary data., disease: Defects in MTM1 are the cause of X-linked centronuclear myopathy X-linked (XCNM) [MIM:310400]; also known as X-linked myotubular myopathy (XLMTM) or myotubular myopathy type 1 (MTM1). 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., function: Dual-specificity phosphatase that acts on both phosphotyrosine and phosphoserine. Could be involved in a signal transduction pathway necessary for late myogenesis, although its ubiquitous expression suggests a wider function., similarity: Belongs to the protein-tyrosine phosphatase family. Non-receptor class myotubularin subfamily., similarity: Contains 1 GRAM domain., similarity: Contains 1 myotubularin phosphatase domain.,