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Rabbit Anti-POMT1 antibody

SL5952R

Product Name:	POMT1
Chinese Name:	蛋白甘露糖基转移酶1抗体
Alias:	POMT1_HUMAN; Protein O-mannosyl-transferase 1; Dolichyl-phosphate-mannose--protein mannosyltransferase 1.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Pig,Cow,Horse,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=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:	85kDa
Cellular localization:	cytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human POMT1:651-747/747
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:	POMT1 (Protein O mannosyl transferase 1) is a multipass membrane protein that is found in the endoplasmic reticulum. POMT1 catalyses the transfer of mannosyl residues to the hydroxyl groups of serine or threonine residues. Enzymatic activity is dependent on co expression of POMT1 with POMT2. Defects in the POMT1 gene are associated with Walker-Warburg syndrome (WWS), a congential muscular dystrophy that is associated with mental retardation and is usually lethal within the first few

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	months of life. Other defects in the POMT1 gene result in limb girdle muscular dystrophy type 2K (LGMD2K), which is associated with mild mental retardation. Studies in Drosophila suggest that mutation of POMT1 alters the efficacy of synaptic transmission and a change in subunit composition of post synaptic glutamate receptors at the neuromuscular junction. Missense mutations in POMT1 have been associated with glioneuronal and glial brain tumours. **Function:** Transfers mannosyl residues to the hydroxyl group of serine or threonine residues. Coexpression of both POMT1 and POMT2 is necessary for enzyme activity, expression of either POMT1 or POMT2 alone is insufficient. **Subunit:** Interacts with POMT2 (Probable). **Subcellular Location:** Endoplasmic reticulum membrane; Multi-pass membrane protein. **Tissue Specificity:** Widely expressed. Highly expressed in testis, heart and pancreas. Detected at lower levels in kidney, skeletal muscle, brain, placenta, lung and liver. **DISEASE:** Muscular dystrophy-dystroglycanopathy congenital with mental retardation B1 (MDDGB1) [MIM:613155]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with mental retardation and mild structural brain abnormalities. Note=The disease is caused by mutations affecting the gene represented in this entry. Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. Note=The disease is caused by mutations affecting the gene represented in this entry. Muscular dystrophy-dystroglycanopathy limb-girdle C1 (MDDGC1) [MIM:609308]: An autosomal recessive degenerative myopathy associated with mild mental retardation without any obvious structural brain abnormality. An abnormal alpha-dystroglycan pattern in observed in the muscle. Note=The disease is caused by mutations affecting the gene represented in this entry. **Similarity:** Belongs to the glycosyltransferase 39 family. Contains 3 MIR domains. **SWISS:**

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	Q9Y6A1 **Gene ID:** 10585 **Database links:** Entrez Gene: 10585Human Omim: 607423Human SwissProt: Q9Y6A1Human Unigene: 522449Human **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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