

## PMGT1 rabbit pAb

Catalog No: YT6311

**Reactivity:** Human; Mouse; Rat

**Applications:** WB;IHC

Target: PMGT1

**Fields:** >>Mannose type O-glycan biosynthesis;>>Metabolic pathways

Gene Name: POMGNT1 MGAT1.2 UNQ746/PRO1475

Q8WZA1

Q91X88

Protein Name: PMGT1

Human Gene ld: 55624

**Human Swiss Prot** 

No:

Mouse Gene Id: 68273

**Mouse Swiss Prot** 

No:

**Rat Gene Id:** 362567

Rat Swiss Prot No: Q5XIN7

Immunogen: Synthesized peptide derived from human PMGT1 AA range: 171-221

**Specificity:** This antibody detects endogenous levels of PMGT1 at Human/Mouse/Rat

**Formulation :** Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% sodium azide.

Source: Polyclonal, Rabbit, IgG

**Dilution:** WB 1:500-2000;IHC 1:50-300

**Purification:** The antibody was affinity-purified from rabbit antiserum by affinity-

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chromatography using epitope-specific immunogen.

**Concentration**: 1 mg/ml

Storage Stability: -15°C to -25°C/1 year(Do not lower than -25°C)

Molecularweight: 73kD

**Background:** This gene encodes a type II transmembrane protein that resides in the Golgi

apparatus. It participates in O-mannosyl glycosylation and is specific for alpha linked terminal mannose. Mutations in this gene may be associated with muscle-eye-brain disease and several congenital muscular dystrophies. Alternatively spliced transcript variants that encode different protein isoforms have been

described. [provided by RefSeq, Feb 2014],

**Function :** catalytic activity:UDP-N-acetyl-D-glucosamine + Man-R = N-acetyl-D-

glucosamine-beta-1,2-Man-R + UDP.,cofactor:Manganese.,disease:Defects in POMGNT1 are a cause of Walker-Warburg syndrome (WWS) [MIM:236670]; also known as hydrocephalus-agyria-retinal dysplasia or HARD syndrome. WWS is an autosomal recessive disorder characterized by cobblestone lissencephaly, hydrocephalus, agyria, retinal displasia, with or without encephalocele. It is often associated with congenital muscular dystrophy and usually lethal within the first few months of life.,disease:Defects in POMGNT1 are the cause of muscle-eyebrain disease (MEB) [MIM:253280]. MEB is an autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities,

cobblestone lissencephaly and cerebellar hypoplasia. MEB patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retina

Subcellular Location:

Golgi apparatus membrane; Single-pass type II membrane protein.

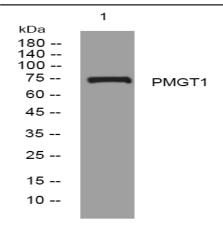
**Expression:** Constitutively expressed. An additional weaker band is also detected in spinal

cord, lymph node, and trachea. Expressed especially in astrocytes. Also

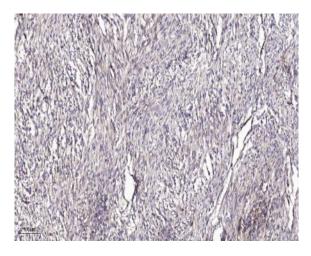
expressed in immature and mature neurons.

## **Products Images**

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Western blot analysis of lysates from 293T cells, primary antibody was diluted at 1:1000, 4° over night



Immunohistochemical analysis of paraffin-embedded human Colon cancer. 1, Antibody was diluted at 1:200(4° overnight). 2, Tris-EDTA,pH9.0 was used for antigen retrieval. 3,Secondary antibody was diluted at 1:200(room temperature, 45min).