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Dymeclin Rabbit pAb

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Catalog # AP55044

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q7RTS9
Reactivity	Human, Rat
Predicted	Mouse, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	75935
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human Dymeclin
Epitope Specificity	151-250/669
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Cytoplasmic and Golgi Apparatus
SIMILARITY	Belongs to the dymeclin family.
SUBUNIT	Interacts with GOLM1 and PPIB.
Post-translational modifications	Myristoylated in vitro; myristoylation is not essential for protein targeting to Golgi compartment.
DISEASE	Defects in DYM are the cause of Dyggve-Melchior-Clausen syndrome (DMC) [MIM:223800]. DMC is a rare autosomal recessive disorder characterized by short trunk dwarfism, microcephaly and psychomotor retardation. Electron microscopic study of cutaneous cells of affected patients shows dilated rough endoplasmic reticulum, enlarged and aberrant vacuoles and numerous vesicles. DMC is progressive. Defects in DYM are the cause of Smith-McCort dysplasia (SMC) [MIM:607326]. SMC is a rare autosomal recessive osteochondrodysplasia characterized by short limbs and trunk with barrel-shaped chest. The radiographic phenotype includes platyspondyly, generalized abnormalities of the epiphyses and metaphyses, and a distinctive lacy appearance of the iliac crest, features identical to those of Dyggve-Melchior-Clausen syndrome.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Dyggve-Melchior-Clausen syndrome (DMC), a rare autosomal recessive disorder, is characterized by microcephaly, short trunk dwarfism and sometime psychomotor retardation. Cutaneous cells of affected individuals show dilated rough endoplasmic reticulum and enlarged vacuoles. The Dyggve-Melchior-Clausen syndrome protein, also designated dymeclin, may play a role in proteoglycan metabolism and intracellular protein digestion. It is a widely expressed multi-pass membrane protein, detected primarily in chondrocytes and fetal brain tissue. Defects in dymeclin are also the cause of Smith-McCort dysplasis syndrome (SMC), which has characteristics identical to those of Dyggve-Melchior-Clausen syndrome.

Additional Information

Gene ID	54808
Other Names	Dymeclin, Dyggve-Melchior-Clausen syndrome protein, DYM
Target/Specificity	Expressed in most embryo-fetal and adult tissues. Abundant in primary chondrocytes, osteoblasts, cerebellum, kidney, lung, stomach, heart, pancreas and fetal brain. Very low or no expression in the spleen, thymus, esophagus, bladder and thyroid gland.
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

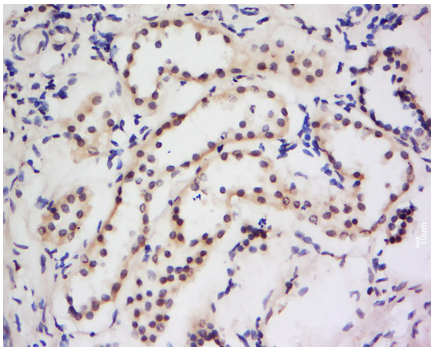
Name	DYM
Function	Necessary for correct organization of Golgi apparatus. Involved in bone development.
Cellular Location	Cytoplasm. Golgi apparatus. Membrane; Lipid-anchor. Note=Sequence analysis programs clearly predict 1 transmembrane region. However, PubMed:18996921 shows that it is not a stably anchored transmembrane protein but it weakly associates with the Golgi apparatus and shuttles between the Golgi and the cytosol
Tissue Location	Expressed in most embryo-fetal and adult tissues. Abundant in primary chondrocytes, osteoblasts, cerebellum, kidney, lung, stomach, heart, pancreas and fetal brain. Very low or no expression in the spleen, thymus, esophagus, bladder and thyroid gland

Background

Dyggve-Melchior-Clausen syndrome (DMC), a rare autosomal recessive disorder, is characterized by microcephaly, short trunk dwarfism and sometime psychomotor retardation. Cutaneous cells of affected individuals show dilated rough endoplasmic reticulum and enlarged vacuoles. The Dyggve-Melchior-Clausen syndrome protein, also designated dymeclin, may play a role in proteoglycan metabolism and intracellular protein digestion. It is a widely expressed multi-pass membrane protein, detected primarily in chondrocytes and fetal brain tissue. Defects in dymeclin are also the cause of Smith-McCort dysplasis syndrome (SMC), which has characteristics identical to those of Dyggve-Melchior-Clausen syndrome.

Images

Tissue/cell: Human kidney tissue; 4% Paraformaldehyde-fixed and paraffin-embedded; Antigen retrieval: citrate buffer (0.01M, pH 6.0), Boiling bathing for 15min; Block endogenous peroxidase by 3% Hydrogen peroxide for 30min; Blocking buffer (normal goat serum,C-0005) at 37℃ for 20 min;



Incubation: Anti- Dymeclin Polyclonal Antibody, Unconjugated(AP55044) 1:200, overnight at 4°C, followed by conjugation to the secondary antibody(SP-0023) and DAB(C-0010) staining