

# 迪格弗-梅尔基奥尔-克劳森综合征相关蛋白 抗体

产品货号： mIR13037

英文名称： Dymeclin

中文名称： 迪格弗-梅尔基奥尔-克劳森综合征相关蛋白抗体

别名： DMC; Dyggve-Melchior-Clausen syndrome protein; DYM; FLJ20071; FLJ90130; SMC; **DYM\_HUMAN**.

研究领域： 细胞生物 发育生物学 信号转导

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Dog, Cow, Horse, Sheep,

产品应用： WB=1:500-2000 ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500

(石蜡切片需做抗原修复)

not yet tested in other applications.

optimal dilutions/concentrations should be determined by the end user.

分子量： 76kDa

细胞定位： 细胞浆

性状： Lyophilized or Liquid

浓度： 1mg/ml

免疫原： KLH conjugated synthetic peptide derived from human Dymeclin:151-250/669

亚 型 : IgG

纯化方法 : affinity purified by Protein A

储 存 液 : 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件 : 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

产品介绍 : 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 dysplasia syndrome (SMC), which has characteristics identical to those of Dyggve-Melchior-Clausen syndrome.

**Function:**

Necessary for correct organization of Golgi apparatus. Involved in bone development.

**Subunit:**

Interacts with GOLM1 and PPIB.

**Subcellular Location:**

Cytoplasmic and Golgi Apparatus

**Tissue 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.

**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.

**Similarity:**

Belongs to the dymeclin family.

**SWISS:**

Q7RTS9

**Gene ID:**

54808

**Important Note:**

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

产品图片

