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Rabbit Anti-Dymeclin/PE Conjugated antibody

SL13037R-PE

Product Name	Anti-Dymeclin/PE
Chinese Name	PE 标记的迪格弗-梅尔基奥尔-克劳森综合征相关蛋白抗体
Alias	DMC; Dyggve-Melchior-Clausen syndrome protein; DYM; FLJ20071; FLJ90130; SMC; DYM_HUMAN.
Research Area	Cell biology Developmental biology Signal transduction
Immunogen Species	Rabbit
Clonality	Polyclonal
React Species	Human,Rat(predicted:Mouse,Dog,Cow,Horse,Sheep)
Applications	IF=1:100-500 not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight	76kDa
Form	Lyophilized or Liquid
Concentration	1mg/ml
immunogen	KLH conjugated synthetic peptide derived from human Dymeclin
Lsotype	IgG
Purification	affinity purified by Protein A
Storage Buffer	1M TBS(pH7.4) with 1% BSA, 3% 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 1M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
Product Detail	**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.

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.

Database links:

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Entrez Gene: 54808 Human

Omim: 607461 Human

SwissProt: Q7RTS9 Human

Unigene: 162996 Human

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