



## Rabbit Anti-PLEKHM1 antibody

SL8062R

<b>Product Name:</b>	PLEKHM1
<b>Chinese Name:</b>	石骨症相关蛋白PLEKHM1抗体
<b>Alias:</b>	162 kDa adapter protein; AP162; PH domain-containing family M member 1; PKHM1_HUMAN; Pleckstrin homology domain-containing family M member 1; PLEKHM1.
<b>Organism Species:</b>	Rabbit
<b>Clonality:</b>	Polyclonal
<b>React Species:</b>	Human,Mouse,Rat,Pig,Horse,Rabbit,
<b>Applications:</b>	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.
<b>Molecular weight:</b>	117kDa
<b>Cellular localization:</b>	cytoplasmic
<b>Form:</b>	Lyophilized or Liquid
<b>Concentration:</b>	1mg/ml
<b>immunogen:</b>	KLH conjugated synthetic peptide derived from human PLEKHM1:546-700/1056
<b>Lsotype:</b>	IgG
<b>Purification:</b>	affinity purified by Protein A
<b>Storage Buffer:</b>	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
<b>Storage:</b>	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.
<b>PubMed:</b>	<a href="#">PubMed</a>
<b>Product Detail:</b>	Involved in vesicular transport in the osteoclast (By similarity). May have a role in sialyl-lex-mediated transduction of apoptotic signals. Tissue specificity: Expressed in placenta, liver, prostate, thymus, spleen, ovary, colon, colon carcinoma and peripheral blood lymphocytes (PBL). Weakly expressed in brain, lung, kidney, and testis. No expression in heart, skeletal muscle, pancreas and small

intestine. Predominantly expressed in the breast carcinoma cell line MCF-7.  
Involvement in disease: Defects in PLEKHM1 are the cause of osteopetrosis autosomal recessive type 6 (OPTB6); also known as autosomal recessive osteopetrosis intermediate form. Osteopetrosis is a rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. The disorder occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. Autosomal recessive osteopetrosis is usually associated with normal or elevated amount of non-functional osteoclasts.

**Function:**

Involved in vesicular transport in the osteoclast (Bysimilarity). May have a role in sialyl-lex-mediated transduction of apoptotic signals.

**Subunit:**

In colon carcinoma and breast carcinoma cells, it interacts with sialyl-lex-positive protein.

**Subcellular Location:**

Cytoplasm (Potential).

**Tissue Specificity:**

Expressed in placenta, liver, prostate, thymus, spleen, ovary, colon, colon carcinoma and peripheral blood lymphocytes (PBL). Weakly expressed in brain, lung, kidney, and testis. No expression in heart, skeletal muscle, pancreas and small intestine. Predominantly expressed in the breast carcinoma cell line MCF-7.

**DISEASE:**

Defects in PLEKHM1 are the cause of osteopetrosis autosomal recessive type 6 (OPTB6) [MIM:611497]; also known as autosomal recessive osteopetrosis intermediate form. Osteopetrosis is a rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. The disorder occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. Autosomal recessive osteopetrosis is usually associated with normal or elevated amount of non-functional osteoclasts.

**Similarity:**

Contains 2 PH domains.

Contains 1 phorbol-ester/DAG-type zinc finger.

Contains 1 RUN domain.

**SWISS:**

Q9Y4G2

**Gene ID:**

9842

**Database links:**

UniProtKB/Swiss-Prot: Q9Y4G2.3

**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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