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SCCD Rabbit pAb

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Catalog # AP59084

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	Q9Y5Z9
Predicted	Rat, Pig, Human, Mouse, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	36831
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human SCCD
Epitope Specificity	2-100/338
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	Endoplasmic reticulum membrane. Cytoplasm. Nucleus. Mitochondrion.
DISEASE	Defects in UBIAD1 are the cause of crystalline corneal dystrophy of Schnyder (SCCD) [MIM:121800]. SCCD is a rare autosomal dominant disease characterized by progressive corneal opacification resulting from abnormal deposition of cholesterol and phospholipids.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene encodes a protein thought to be involved in cholesterol and phospholipid metabolism. Mutations in this gene are associated with Schnyder crystalline corneal dystrophy. [provided by RefSeq, Oct 2008]

Additional Information

Gene ID	29914
Other Names	UbiA prenyltransferase domain-containing protein 1, 2.5.1.-, 2.5.1.39, Transitional epithelial response protein 1, UBIAD1 {ECO:0000303|PubMed:20953171, ECO:0000312|HGNC:HGNC:30791}
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,ICC/IF=1:100-500,IF=1:100-500,ELISA=1:5000-10000
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	UBIAD1 {ECO:0000303|PubMed:20953171, ECO:0000312|HGNC:HGNC:30791}
Function	Prenyltransferase that mediates the formation of menaquinone- 4 (MK-4) and coenzyme Q10 (PubMed:20953171, PubMed:23374346). MK-4 is a vitamin K2 isoform present at high concentrations in the brain, kidney and pancreas, and is required for endothelial cell development (PubMed:20953171). Mediates the conversion of phylloquinone (PK) into MK-4, probably by cleaving the side chain of phylloquinone (PK) to release 2-methyl-1,4-naphthoquinone (menadione; K3) and then prenylating it with geranylgeranyl pyrophosphate (GGPP) to form MK-4 (PubMed:20953171). Also plays a role in cardiovascular development independently of MK-4 biosynthesis, by acting as a coenzyme Q10 biosynthetic enzyme: coenzyme Q10, also named ubiquinone, plays an important antioxidant role in the cardiovascular system (PubMed:23374346). Mediates biosynthesis of coenzyme Q10 in the Golgi membrane, leading to protect cardiovascular tissues from NOS3/eNOS- dependent oxidative stress (PubMed:23374346).
Cellular Location	Endoplasmic reticulum membrane; Multi-pass membrane protein. Golgi apparatus membrane; Multi-pass membrane protein. Mitochondrion membrane; Multi-pass membrane protein. Cytoplasm. Nucleus
Tissue Location	Ubiquitously expressed.

Background

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Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.