10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

ABCA12 Rabbit pAb

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Catalog # AP54667

Product Information

Primary Accession	Q86UK0
Reactivity	Human
Predicted	Rat, Pig, Dog, Mouse, Rabbit
Host	Rabbit
Clonality	Polyclonal
Calculated MW	293237
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human ABCA12
Epitope Specificity	2051-2200/2595
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	Membrane.
DISEASE	Defects in ABCA12 are the cause of ichthyosis harlequin (HI) [MIM:242500]; also known as harlequin fetus. HI is a very severe skin disorder in which the neonate is born with a thick covering of armor-like scales. The skin dries out to form hard diamond-shaped plaques separated by fissures, resembling 'armor plating'. The normal facial features are severely affected, with distortion of the lips (eclabion), eyelids (ectropion), ears, and nostrils. Affected babies are often born prematurely and rarely survive the perinatal period. Defects in ABCA12 are the cause of ichthyosis lamellar type 2 (LI2) [MIM:601277]; also known as ichthyosis congenita IIB (ICR2B). LI is a non-bullous ichthyosis, a skin disorder characterized by abnormal cornification of the epidermis. It is one the most severe forms of ichthyoses apparent at birth and persisting throughout life. LI patients are born encased in a tight, shiny, translucent covering called collodion membrane. Over the first weeks of life, the collodion membrane is gradually replaced by generalized large, dark brown, plate-like scales with minimal to no erythroderma. Tautness of facial skin commonly results in ectropion, eclabium and scarring alopecia of the scalp. Common complications are severe heat intolerance and recurrent ear infections.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	The ATP-binding cassette (ABC) transporters, or traffic ATPases, constitute an expansive family of proteins accountable for the transport of a wide variety of substrates across cell membranes in both prokaryotic and eukaryotic cells. They also aid in the regulation of lipid transport and membrane trafficking. ABCA12 (ATP-Binding Cassette, Subfamily A, Member 12) contains two transmembrane (TM) domains, each with six membrane-spanning segments, and two nucleotide-binding domains (NBDs), which are located in the cytoplasm. ABCA12 is expressed in normal human keratinocytes (RT-PCR reveals expression in placenta, testis, fetal brain, and skin) and is upregulated during keratinization. Immunoelectron microscopy reveals that the ABCA12

protein is located in lamellar granules in the upper epidermal keratinocytes of human skin. The ABCA12 gene, which synthesizes a 2,595-amino acid protein, may produce an alternative splice variant with an in-frame deletion leading to truncation of 79 amino acids.

Additional Information

Gene ID	26154
Other Names	Glucosylceramide transporter ABCA12, 7.6.2.1, ATP-binding cassette sub-family A member 12, ATP-binding cassette transporter 12, ATP-binding cassette 12, ABCA12 (HGNC:14637), ABC12
Dilution	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	ABCA12 (HGNC:14637)
Synonyms	ABC12
Function	Transports lipids such as glucosylceramides from the outer to the inner leaflet of lamellar granules (LGs) membrane, whereby the lipids are finally transported to the keratinocyte periphery via the trans-Golgi network and LGs and released to the apical surface of the granular keratinocytes to form lipid lamellae in the stratum corneum of the epidermis, which is essential for skin barrier function (PubMed:16007253, PubMed:20869849). In the meantime, participates in the transport of the lamellar granules-associated proteolytic enzymes, in turn regulates desquamation and keratinocyte differentiation (PubMed:19179616). Furthermore, is essential for the regulation of cellular cholesterol homeostasis by regulating ABCA1-dependent cholesterol efflux from macrophages through interaction with NR1H2 and ABCA1 (By similarity). Plays pleiotropic roles in regulating glucose stimulated insulin secretion from beta cells, regulating the morphology and fusion of insulin granules, lipid raft abundance and the actin cytoskeleton (By similarity). Also involved in lung surfactant biogenesis (By similarity).
Cellular Location	Cytoplasmic vesicle, secretory vesicle membrane; Multi-pass membrane protein. Golgi apparatus membrane. Note=Localizes in the limiting membrane of the lamellar granules (LGs) (PubMed:17927575). Trafficks from the Golgi apparatus to the lamellar granules (LGs) at the cell periphery in the uppermost granular layer keratinocytes where ABCA12-positive LGs fuse with the keratinocyte-cell membrane to secrete their lipid content to the extracellular space of the stratum corneum (PubMed:16007253, PubMed:17927575). Co-localizes through the Golgi apparatus to the cell periphery with glucosylceramide (PubMed:17927575)
Tissue Location	Mainly expressed in the stomach, placenta, testis and fetal brain (PubMed:12697999). Expressed in the upper epidermal layers, mainly the granular layers, of skin (PubMed:16007253, PubMed:17591952, PubMed:17927575). Expressed throughout the normal interfollicular epidermis with prominent expression in the stratum granulosum

(PubMed:19179616). Expressed in alpha and beta cells of pancreatic islets (PubMed:32072744).

Background

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