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Rabbit Anti-ABCA12 antibody

SL11906R

Product Name	ABCA12
Chinese Name	腺苷三磷酸结合盒转运体 12 抗体
Alias	ABC transporter A family member 12; ABC transporter ABCA.12; ABC12; ABCA12; ABCAC_HUMAN; AtABCA12; ATH16; ATP binding cassette 12; ATP binding cassette sub family A (ABC1) member 12; ATP binding cassette sub family A member 12; ATP binding cassette transporter 12; ATP-binding cassette 12; ATP-binding cassette sub-family A member 12; ATP-binding cassette transporter 12; Ichthyosis congenita II lamellar ichthyosis B; ICR2B; LI2; Putative ABC2 homolog 16; ABCAC_HUMAN.
Research Area	Tumour Cell biology immunology Signal transduction The new supersedes the old The cell membrane 蛋白
Immunogen Species	Rabbit
Clonality	Polyclonal
React Species	Human, (predicted: Mouse, Rat, Dog, Pig, Rabbit,)
Applications	ELISA=1:5000-10000 not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Theoretical molecular weight	293kDa
Cellular localization	The cell membrane
Form	Liquid
Concentration	1mg/ml
immunogen	KLH conjugated synthetic peptide derived from human ABCA12: 2051-2200/2595
Lsotype	IgG
Purification	affinity purified by Protein A
Buffer Solution	1M TBS(pH7.4) with 1% BSA, 3% Proclin300 and 50% Glycerol.
Storage	Shipped at 4℃. Store at -20 °C for one year. Avoid repeated freeze/thaw cycles.

Attention

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

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Product Detail

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.

Function:
Probable transporter involved in lipid homeostasis.

Subcellular Location:
Membrane.

Tissue Specificity:
Mainly expressed in the stomach, placenta, testis and fetal brain.

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.

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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.

Similarity:
Belongs to the ABC transporter superfamily. ABCA family.
Contains 2 ABC transporter domains.

SWISS:
Q86UK0

Gene ID:
26154

Database links:

Entrez Gene: 26154 Human

Entrez Gene: 74591 Mouse

Entrez Gene: 301482 Rat

Omim: 607800 Human

SwissProt: Q86UK0 Human

Unigene: 134585 Human