abcam

Product datasheet

Recombinant Human ABCA4 protein ab114660

1 图像

描述

产品名称	重组人ABCA4蛋白
表达系统	Wheat germ
Accession	P78363
蛋白长度	Protein fragment
无动物成分	No
性质	Recombinant
种属	Human
序列	PKDDLLPDLNPVEQFFQGNFPGSVQRERHYNMLQFQVSSSSLARIFQLLL SHKDSLLIEEYSVTQTTLDQVFVNFAKQQTESHDLPLHPRAAGASRQAQD
预测分子量	37 kDa including tags
氨基酸	2174 to 2273

技术指标

Our **Abpromise guarantee** covers the use of **ab114660** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

应用	Western blot SDS-PAGE ELISA
形式	Liquid

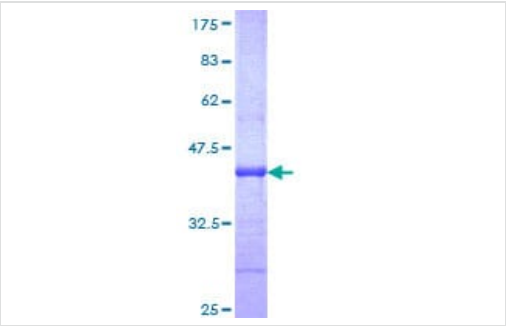
制备和贮存

稳定性和存储	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.3% Glutathione, 0.79% Tris HCl

常规信息

功能	In the visual cycle, acts as an inward-directed retinoid flipase, retinoid substrates imported by ABCA4 from the extracellular or intradiscal (rod) membrane surfaces to the cytoplasmic membrane surface are all-trans-retinaldehyde (ATR) and N-retinyl-phosphatidyl-ethanolamine (NR-PE). Once transported to the cytoplasmic surface, ATR is reduced to vitamin A by trans-retinol dehydrogenase (tRDH) and then transferred to the retinal pigment epithelium (RPE) where it is converted to 11-cis-retinal. May play a role in photoresponse, removing ATR/NR-PE from the extracellular photoreceptor surfaces during bleach recovery.
组织特异性	Retinal-specific. Seems to be exclusively found in the rims of rod photoreceptor cells.
疾病相关	Defects in ABCA4 are the cause of Stargardt disease type 1 (STGD1) [MIM:248200]. STGD is one of the most frequent causes of macular degeneration in childhood. It is characterized by macular dystrophy with juvenile-onset, rapidly progressive course, alterations of the peripheral retina, and subretinal deposition of lipofuscin-like material. STGD1 inheritance is autosomal recessive. Defects in ABCA4 are the cause of fundus flavimaculatus (FFM) [MIM:248200]. FFM is an autosomal recessive retinal disorder very similar to Stargardt disease. In contrast to Stargardt disease, FFM is characterized by later onset and slowly progressive course. Defects in ABCA4 may be a cause of age-related macular degeneration type 2 (ARMD2) [MIM:153800]. ARMD is a multifactorial eye disease and the most common cause of irreversible vision loss in the developed world. In most patients, the disease is manifest as ophthalmoscopically visible yellowish accumulations of protein and lipid (known as drusen) that lie beneath the retinal pigment epithelium and within an elastin-containing structure known as Bruch membrane. Defects in ABCA4 are the cause of cone-rod dystrophy type 3 (CORD3) [MIM:604116]. CORDs are inherited retinal dystrophies belonging to the group of pigmentary retinopathies. CORDs are characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. Defects in ABCA4 are the cause of retinitis pigmentosa type 19 (RP19) [MIM:601718]. RP leads to degeneration of retinal photoreceptor cells. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. RP19 is characterized by choroidal atrophy. Inheritance is autosomal recessive.
序列相似性	Belongs to the ABC transporter superfamily. ABCA family. Contains 2 ABC transporter domains.
细胞定位	Membrane. Localized to outer segment disk edges of rods and cones, with around one million copies/photoreceptor.

图片



SDS-PAGE - Recombinant Human ABCA4 protein (ab114660)

SDS-PAGE analysis of ab114660 on a 12.5% gel stained with Coomassie Blue.

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