abcam

Product datasheet

Recombinant Human ABCA4 protein ab114660

画像数 1

製品の詳細

製品名	Recombinant Human ABCA4 protein
発現系	Wheat germ
アクセッション番号	P78363
タンパク質長	Protein fragment
Animal free	No
由来	Recombinant
生物種	Human
配列	PKDDLLPDLNPVEQFFQGNFPGSVQRERHYNMLQFQVSSSSLARIFQLLL SHKDSLLIEEYSVTQTTLDQVFVNFAKQQTESHDLPLHPRAAGASRQAQD
予測される分子量	37 kDa including tags
領域	2174 to 2273

特性

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.

アプリケーション	Western blot SDS-PAGE ELISA
製品の状態	Liquid

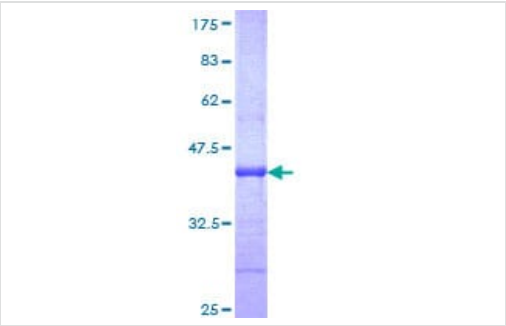
前処理および保存

保存方法および安定性	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

関連情報

機能	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.
組織特異性	Retinal-specific. Seems to be exclusively found in the rims of rod photoreceptor cells.
関連疾患	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.
配列類似性	Belongs to the ABC transporter superfamily. ABCA family. Contains 2 ABC transporter domains.
細胞内局在	Membrane. Localized to outer segment disk edges of rods and cones, with around one million copies/photoreceptor.

画像



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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