abcam

Product datasheet

Recombinant Human CRB1 protein ab161634

1 Image

Description

Product name	Recombinant Human CRB1 protein
Expression system	Wheat germ
Protein length	Protein fragment
Animal free	No
Nature	Recombinant
Species	Human
Sequence	FCNKNNTRCLSNSCQNNSTCKDFSKDNDCSCSDTAN NLDKDCDNMKDPCF SNPCQGSATCVNTPGERSFLCKCPPGYSGTICETTIGS CGKNSCQHGGIC HQDPIYPVC
Amino acids	26 to 134
Tags	GST tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of **ab161634** in the following tested applications.

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

Applications	ELISA Western blot
Form	Liquid
Additional notes	Protein concentration is above or equal to 0.05 mg/ml.

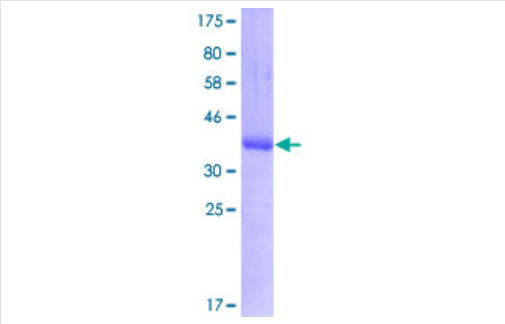
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Function	Plays a role in photoreceptor morphogenesis in the retina. May maintain cell polarization and adhesion.
Tissue specificity	Preferential expression in retina, also expressed in brain, testis, fetal brain and fetal eye.
Involvement in disease	Note=CRB1 mutations have been found in various retinal dystrophies, chronic and disabling disorders of visual function. They predominantly involve the posterior portion of the ocular fundus, due to degeneration in the sensory layer of the retina, retinal pigment epithelium, Bruch membrane, choroid, or a combination of these tissues. Onset of inherited retinal dystrophies is painless, bilateral and typically progressive. Most people experience gradual peripheral vision loss or tunnel vision, and difficulties with poor illumination and night vision. Central vision is usually unaffected, so the person may still be able to read. However, it can also deteriorate to cause total blindness. Examples of retinal dystrophies are retinitis pigmentosa, Leber congenital amaurosis, cone-rod dystrophy among others. Defects in CRB1 are the cause of retinitis pigmentosa type 12 (RP12) [MIM:600105]. A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells, followed by secondary loss of cone photoreceptors. 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. RP12 is an autosomal recessive severe form oFTen manifesting in early childhood. Patients experiment progressive visual field loss with severe visual impairment before the age of twenty. Some patients have a preserved paraarteriolar retinal pigment epithelium (PPRPE) and hypermetropia. Defects in CRB1 are the cause of Leber congenital amaurosis type 8 (LCA8) [MIM:613835]. LCA designates a clinically and genetically heterogeneous group of childhood retinal degenerations, generally inherited in an autosomal recessive manner. Affected infants have little or no retinal photoreceptor function as tested by electroretinography. LCA represents the most common genetic cause of congenital visual impairment in infants and children. Defects in CRB1 are the cause of pigmented paravenous chorioretinal atrophy (PPCRA) [MIM:172870]. PPCRA is an unusual retinal degeneration characterized by accumulation of pigmentation along retinal veins. PPCRA is dominantly inherited, but exhibited variable expressivity. Males are more likely to exhibit a severe phenotype, whereas females may remain virtually asymptomatic even in later years. The PPCRA phenotype is associated with a mutation in CRB1 gene which is likely to affect the structure of the CRB1 protein.
Sequence similarities	Belongs to the Crumbs protein family. Contains 19 EGF-like domains. Contains 3 laminin G-like domains.
Post-translational modifications	Extensively glycosylated.
Cellular localization	Secreted and Apical cell membrane. Distributed at the apical membrane of all retinal epithelial cells. Located in the apical membrane of the adherens junction in outer limiting membrane (OLM) of the retina.

Images



SDS-PAGE - Recombinant Human CRB1 protein (ab161634)

ab161634 on a 12.5% SDS-PAGE stained with Coomassie Blue.

Please note: All products are "FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC PROCEDURES"

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