abcam

Product datasheet

Recombinant Human RS1 protein ab159464

1 Image

Description

Product name Recombinant Human RS1 protein

Expression system Wheat germ

Protein length Full length protein

Animal free No

Nature Recombinant

Species Human

Sequence MSRKIEGFLLLLLFGYEATLGLSSTEDEGEDPWYQKACKC

DCQGGPNALW

SAGATSLDCIPECPYHKPLGFESGEVTPDQITCSNPEQYV

GWYSSWTANK

ARLNSQGFGCAWLSKFQDSSQWLQIDLKEIKVISGILTQGR

CDIDEWMTK

YSVQYRTDERLNWIYYKDQTGNNRVFYGNSDRTSTVQNLL

RPPIISRFIR LIPLGWHVRIAIRMELLECVSKCA

Amino acids 1 to 224

Tags GST tag N-Terminus

Specifications

Our Abpromise guarantee covers the use of ab159464 in the following tested applications.

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

Applications Western blot

ELISA

Form Liquid

Additional notes

Preparation and Storage

Stability and Storage Shipped on dry ice. Upon delivery aliquot and store at -80°C. Avoid freeze / thaw cycles.

pH: 8.00

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General Info

Function May be active in cell adhesion processes during retinal development.

Tissue specificity Restricted to the retina (at protein level). At the mRNA level, detected only within the

photoreceptor cell layer, most prominently within the inner segments of the photoreceptors. Undetectable in the inner plexiform layers and the inner nuclear layer. At the protein level, found in the inner segment of the photoreceptors, the inner nuclear layer, the inner plexiform layer and the ganglion cell layer. At the macula, expressed in both the outer and inner nuclear layers and in the

inner plexiform layer (at protein level).

Involvement in disease Defects in RS1 are the cause of retinoschisis juvenile X-linked type 1 (XLRS1) [MIM:312700]. A

vitreo-retinal dystrophy characterized by macular pathology and by splitting of the superficial layer of the retina. Macular changes are present in almost all cases. In the fundi, radially oriented intraretinal foveomacular cysts are seen in a spoke-wheel configuration, with the absence of foveal reflex in most cases. In addition, approximately half of cases have bilateral peripheral retinoschisis in the inferotemporal part of the retina. Aside from the typical fundus appearance, strabismus, nystagmus, axial hyperopia, defective color vision and foveal ectopy can be present. The most important complications are vitreous hemorrhage, retinal detachment, and neovascular

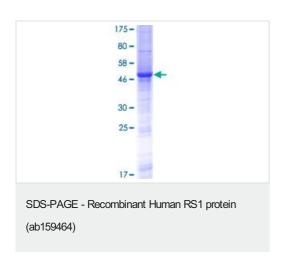
glaucoma.

Sequence similarities Contains 1 F5/8 type C domain.

Developmental stage Up-regulated during the differentiation of a retinoblastoma cell line.

Cellular localization Secreted.

Images



ab159464 on a 12.5% SDS-PAGE stained with Coomassie Blue.

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