

Product datasheet

Human RPE65 peptide ab99880

1 Image

Description

Product name	Human RPE65 peptide
Purity	> 70 % HPLC. 70 - 90% by HPLC
Accession	<u>Q16518</u>
Animal free	No
Nature	Synthetic
Species	Human

Specifications

Our **Abpromise guarantee** covers the use of **ab99880** in the following tested applications.

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

Applications	Blocking
Form	Liquid
Additional notes	<ul style="list-style-type: none"> - First try to dissolve a small amount of peptide in either water or buffer. The more charged residues on a peptide, the more soluble it is in aqueous solutions. - If the peptide doesn't dissolve try an organic solvent e.g. DMSO, then dilute using water or buffer. - Consider that any solvent used must be compatible with your assay. If a peptide does not dissolve and you need to recover it, lyophilise to remove the solvent. - Gentle warming and sonication can effectively aid peptide solubilisation. If the solution is cloudy or has gelled the peptide may be in suspension rather than solubilised. - Peptides containing cysteine are easily oxidised, so should be prepared in solution just prior to use.

Preparation and Storage

Stability and Storage	<p>Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.</p> <p>Information available upon request.</p>
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General Info

Function

Plays important roles in the production of 11-cis retinal and in visual pigment regeneration. The soluble form binds vitamin A (all-trans-retinol), making it available for LRAT processing to all-trans-retinyl ester. The membrane form, palmitoylated by LRAT, binds all-trans-retinyl esters, making them available for IMH (isomerohydrolase) processing to all-cis-retinol. The soluble form is regenerated by transferring its palmitoyl groups onto 11-cis-retinol, a reaction catalyzed by LRAT. The enzymatic activity is linearly dependent of the expression levels and membrane association.

Tissue specificity

Retinal pigment epithelium specific.

Involvement in disease

Defects in RPE65 are the cause of Leber congenital amaurosis type 2 (LCA2) [MIM:204100]. 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 RPE65 are the cause of retinitis pigmentosa type 20 (RP20) [MIM:613794]. 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. RP20 inheritance is autosomal dominant.

Sequence similarities

Belongs to the carotenoid oxygenase family.

Post-translational modifications

Palmitoylation by LRAT regulates ligand binding specificity; the palmitoylated form (membrane form) specifically binds all-trans-retinyl-palmitate, while the soluble unpalmitoylated form binds all-trans-retinol (vitamin A).

Cellular localization

Cytoplasm. Cell membrane. Attached to the membrane by a lipid anchor when palmitoylated (membrane form), soluble when unpalmitoylated.

Images

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Human RPE65 peptide (ab99880)

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