



Rabbit Anti-RPE65 antibody

SL9575R

Product Name:	RPE65
Chinese Name:	视网膜色素epithelial cells特异性蛋白65抗体
Alias:	All-trans-retinyl-palmitate hydrolase; LCA 2; LCA2; Leber congenital amaurosis; mRPE 65; mRPE65; p63; rd 12; rd12; Retinal pigment epithelium specific 61 kDa protein; Retinal pigment epithelium specific 65 kDa protein; Retinal pigment epithelium specific protein; Retinal pigment epithelium specific protein 65kDa; Retinal pigment epithelium-specific 65 kDa protein; Retinitis pigmentosa 20; Retinoid isomerohydrolase; RP 20; RP20; RPE 65; RPE65; RPE65_HUMAN; sRPE 65; sRPE65.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Dog,Cow,Horse,Rabbit,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	59kDa
Cellular localization:	cytoplasmicThe cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human RPE65:21-120/533
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	The retinal pigment epithelium (RPE) is a monolayer simple epithelium in proximity to

the outer surface of the retinal photoreceptor cells. Retinal pigment epithelium-specific protein (RPE65) is a 65 kDa protein belonging to the β -carotene dioxygenase family. This protein is important in 11-cis retinal production as well as in visual pigment regeneration. RPE65 is attached to the membrane by a lipid anchor when palmitoylated (membrane form) and soluble when unpalmitoylated. The soluble form of the protein binds vitamin A. Defects in RPE65 causes autosomal dominant retinitis pigmentosa and/or Leber congenital amaurosis type 2.

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.

Subunit:

Interacts with MYO7A; this mediates light-dependent intracellular transport of RPE65.

Subcellular Location:

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

Tissue Specificity:

Retinal pigment epithelium specific.

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).

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.

Similarity:

Belongs to the carotenoid oxygenase family.

SWISS:

Q16518

Gene ID:
6121

Database links:

[Entrez Gene: 6121](#)Human

[Entrez Gene: 19892](#)Mouse

[Entrez Gene: 89826](#)Rat

[Oimim: 180069](#)Human

[SwissProt: Q9YGX2](#)Chicken

[SwissProt: Q28175](#)Cow

[SwissProt: Q16518](#)Human

[SwissProt: Q91ZQ5](#)Mouse

[SwissProt: O70276](#)Rat

[Unigene: 2133](#)Human

[Unigene: 131708](#)Mouse

[Unigene: 76724](#)Rat

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.