



Human RPE65 peptide (DAG-P1919)

This product is for research use only and is not intended for diagnostic use.

PRODUCT INFORMATION

Antigen Description	This gene encodes a protein which is located in the retinal pigment epithelium and is involved in the production of 11-cis retinal and in visual pigment regeneration. There are two forms of this protein, a soluble form called sRPE65, and a palmitoylated, membrane-bound form known as mRPE65. mRPE65 serves as the palmitoyl donor for lecithin retinol acyl transferase (LRAT), the enzyme that catalyzes the vitamin A to all trans retinol step of the chromophore regeneration process. Both mRPE65 and sRPE65 also serve as regulatory proteins, with the ratio and concentrations of these molecules playing a role in the inhibition of 11-cis retinal synthesis. Mutations in this gene have been associated with Leber congenital amaurosis type 2 (LCA2) and retinitis pigmentosa. [provided by RefSeq, Jul 2008]
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Specificity	Retinal pigment epithelium specific.
Purity	70 - 90% by HPLC.
Conjugate	Unconjugated
Sequence Similarities	Belongs to the carotenoid oxygenase family.
Format	Liquid
Preservative	None
Storage	Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles. Information available upon request.

GENE INFORMATION

Gene Name	RPE65 retinal pigment epithelium-specific protein 65kDa [Homo sapiens (human)]
Official Symbol	RPE65

Synonyms	RPE65; retinal pigment epithelium-specific protein 65kDa; LCA2; RP20; rd12; mRPE65; sRPE65; retinoid isomerohydrolase; p63; retinol isomerase; RBP-binding membrane protein; all-trans-retinyl-palmitate hydrolase; retinitis pigmentosa 20 (autosomal recessive); retinal pigment epithelium-specific 65 kDa protein;
Entrez Gene ID	6121
mRNA Refseq	NM_000329.2
Protein Refseq	NP_000320.1
UniProt ID	Q16518
Chromosome Location	1p31
Pathway	Disease, organism-specific biosystem; Diseases associated with visual transduction, organism-specific biosystem; Retinol metabolism, organism-specific biosystem; Retinol metabolism, conserved biosystem; Signal Transduction, organism-specific biosystem; The canonical retinoid cycle in rods (twilight vision), organism-specific biosystem; Visual phototransduction, organism-specific biosystem; Visual signal transduction: Cones, organism-specific biosystem; Visual signal transduction: Rods, organism-
Function	all-trans-retinyl-ester hydrolase, 11-cis retinol forming activity; all-trans-retinyl-palmitate hydrolase, 11-cis retinol forming activity; metal ion binding; retinal isomerase activity;