Recombinant Human RPE65, MYC/DDK-tagged

Cat. No. RPE65-246H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human RPE65, fused with C-terminal MYC/DDK, was expressed in HEK293 cells.
Species	Human
Source	HEK293
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.
Molecular Mass	60.8 kDa
Purity	> 80% as determined by SDS-PAGE and Coomassie blue staining
Concentration	>50 ug/mL as determined by microplate BCA method
Storage Buffer	25 mM Tris.HCl, pH 7.3, 100 mM glycine, 10% glycerol.

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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; retinal pigment epithelium specific protein (65kD) , RP20; retinoid isomerohydrolase; all trans retinyl palmitate hydrolase; LCA2; rd12; retinol isomerase; p63; RBP-binding membrane protein; all-trans-retinyl-palmitate hydrolase; retinitis pigmentosa 20 (autosomal recessive); retinal pigment epithelium-specific 65 kDa protein; RP20; mRPE65; sRPE65
Gene ID	6121
mRNA Refseq	NM_000329
Protein Refseq	NP_000320
MIM	180069
UniProt ID	Q16518
Chromosome Location	1p31
Pathway	Diseases associated with visual transduction; Retinol metabolism; Visual signal transduction: Rods
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

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