

## Recombinant Human RPE65, MYC/DDK-tagged

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

### SPECIFICATION

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

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## GENE INFORMATION

<b>Gene Name</b>	RPE65 retinal pigment epithelium-specific protein 65kDa [ Homo sapiens (human) ]
<b>Official Symbol</b>	RPE65
<b>Synonyms</b>	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
<b>Gene ID</b>	6121
<b>mRNA Refseq</b>	NM_000329
<b>Protein Refseq</b>	NP_000320
<b>MIM</b>	180069
<b>UniProt ID</b>	Q16518
<b>Chromosome Location</b>	1p31
<b>Pathway</b>	Diseases associated with visual transduction; Retinol metabolism; Visual signal transduction: Rods
<b>Function</b>	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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