



## **Human ABCA4 blocking peptide (CDBP0257)**

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

## PRODUCT INFORMATION

Product Overview	Blocking/Immunizing peptide for anti-ABCA4 (aa1336-49) antibody
Antigen Description	The membrane-associated protein encoded by this gene is a member of the superfamily of ATP-binding cassette (ABC) transporters. ABC proteins transport various molecules across extra- and intracellular membranes. ABC genes are divided into seven distinct subfamilies (ABC1, MDR/TAP, MRP, ALD, OABP, GCN20, White). This protein is a member of the ABC1 subfamily. Members of the ABC1 subfamily comprise the only major ABC subfamily found exclusively in multicellular eukaryotes. This protein is a retina-specific ABC transporter with N-retinylidene-PE as a substrate. It is expressed exclusively in retina photoreceptor cells, indicating the gene product mediates transport of an essental molecule across the photoreceptor cell membrane. Mutations in this gene are found in patients diagnosed with Stargardt disease, a form of juvenile-onset macular degeneration. Mutations in this gene are also associated with retinitis pigmentosa-19, cone-rod dystrophy type 3, early-onset severe retinal dystrophy, fundus flavimaculatus, and macular degeneration age-related 2.
Species	Human
Conjugate	Unconjugated
Applications	Apuri, BL, ELISA
Format	Lyophilized powder
Size	100 μg
Preservative	None
Storage	Shipped at ambient temperature, store at -20°C.

## **GENE INFORMATION**

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Gene Name	ABCA4 ATP-binding cassette, sub-family A (ABC1), member 4 [ Homo sapiens ]
Official Symbol	ABCA4
Synonyms	ABCA4; ATP-binding cassette, sub-family A (ABC1), member 4; ABCR, ATP binding cassette transporter, retinal specific, RP19, STGD, STGD1; retinal-specific ATP-binding cassette transporter; ARMD2; FFM; Stargardt disease; RIM protein; RIM ABC transporter; photoreceptor rim protein; stargardt disease protein; retina-specific ABC transporter; ATP binding cassette transporter; ATP-binding transporter, retina-specific; ATP-binding cassette sub-family A member 4; ATP-binding cassette transporter, retinal-specific; RMP; ABCR; RP19; STGD; ABC10; CORD3; STGD1; FLJ17534; DKFZp781N1972;
Entrez Gene ID	24
mRNA Refseq	<u>NM 000350</u>
Protein Refseq	<u>NP_000341</u>
UniProt ID	P78363
Chromosome Location	1p22
Pathway	ABC transporters, organism-specific biosystem; ABC transporters, conserved biosystem; ABC-family proteins mediated transport, organism-specific biosystem; Transmembrane transport of small molecules, organism-specific biosystem;
Function	ATP binding; ATPase activity; ATPase activity, coupled to transmembrane movement of substances; nucleotide binding; phospholipid-translocating ATPase activity; transporter activity;