

Recombinant Human ABCA4 Protein, GST-Tagged

Cat. No. ABCA4-033H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human ABCA4 partial ORF (NP_000341, 2174 a.a. - 2273 a.a.) recombinant protein with GST-tag at N-terminal.
Species	Human
Source	Wheat Germ
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 essential 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. [provided by RefSeq, Jul 2008]
Molecular Mass	36.74 kDa

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AA Sequence	PKDDLPLDLPVEQFFQGNFPGSVQRRERHYNMLQFQVSSSSLARIFQLLLSHKDSLL IEEYSVTQTTLQVFNFAKQQTESHDLPLHPRAAGASRQAQD
Applications	Enzyme-linked Immunoabsorbent Assay Western Blot (Recombinant protein) Antibody Production Protein Array
Notes	Best use within three months from the date of receipt of this protein.
Storage	Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.
Storage Buffer	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.
GENE INFORMATION	
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;
Gene ID	24

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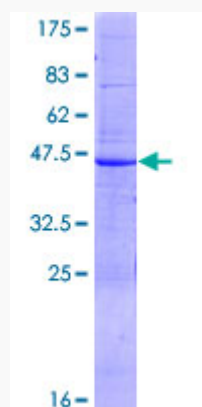
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mRNA Refseq NM_000350

Protein Refseq NP_000341

MIM 601691

UniProt ID P78363



12.5% SDS-PAGE Stained with Coomassie Blue.

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