

## ABCA4

**Reactivity:** Human Mouse Rat

**Tested applications:** WB

**Recommended Dilution:** WB 1:1000 - 1:2000

**Calculated MW:** 256kDa

**Observed MW:** Refer to figures

**Immunogen:**

Recombinant protein of human ABCA4

**Storage Buffer:**

Store at -20. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 50% glycerol, pH7.3.

**Synonym:**

FFM; RMP; ABCR; RP19; STGD; ABC10; ARMD2; CORD3; STGD1;

**Catalog #:** A10556

**Antibody Type:**

Polyclonal Antibody

**Species:** Rabbit

**Gene ID:** 24

**Isotype:** IgG

**Swiss Prot:** P78363

**Purity:** Affinity purification

For research use only.

**Background:**

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.

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