

ABCA4 (H-235): sc-20967

BACKGROUND

The ATP-binding cassette (ABC) superfamily is comprised of transmembrane proteins involved in energy-dependent transport of a variety of substrates across membranes. ABCA4 (also designated ABCR, photoreceptor RIM or RMP) is a photoreceptor specific ATP-binding cassette (ABC) transporter. ABCA4 is exclusively expressed within photoreceptor cells, indicating that ABCA4 mediates the transport of an essential molecule either into or out of photoreceptor cells. Mutations in the gene encoding ABCA4 are responsible for autosomal recessive Stargardt disease (STGD), an early onset macular degeneration, and some forms of autosomal recessive cone-rod dystrophy and autosomal recessive retinitis pigmentosa. In addition, heterozygosity for ABCA4 mutations may possess a risk factor for age-related macular degeneration. ABCA4 is most closely related to the mouse and human ABC1 and ABC2 and maps to human chromosome 1p22.1.

REFERENCES

- Hyde, S.C., Emsley, P., Hartshorn, M.J., Mimmack, M.M., Gileadi, U., Pearce, S.R., Gallagher, M.P., Gill, D.R., Hubbard, R.E. and Higgins, C.F. 1990. Structural model of ATP-binding proteins associated with cystic fibrosis, multidrug resistance and bacterial transport. *Nature* 346: 362-365.
- Dean, M. and Allikmets, R. 1995. Evolution of ATP-binding cassette transporter genes. *Curr. Opin. Genet. Dev.* 5: 779-785.
- Allikmets, R., Shroyer, N.F., Singh, N., Seddon, J.M., Lewis, R.A., Bernstein, P.S., Peiffer, A., Zabriskie, N.A., Li, Y., Hutchinson, A., Dean, M., Lupski, J.R. and Leppert, M. 1997. Mutation of the Stargardt disease gene (ABCR) in age-related macular degeneration. *Science* 277: 1805-1807.
- Schwiebert, E.M. 1999. ABC transporter-facilitated ATP conductive transport. *Am. J. Physiol.* 276: 1-8.
- Sun, H., Smallwood, P.M. and Nathans, J. 2000. Biochemical defects in ABCR protein variants associated with human retinopathies. *Nat. Genet.* 26: 242-246.
- Yatsenko, A.N., Shroyer, N.F., Lewis, R.A. and Lupski, J.R. 2001. Late-onset Stargardt disease is associated with missense mutations that map outside known functional regions of ABCR (ABCA4). *Hum. Genet.* 108: 346-355.
- Paloma, E., Martinez-Mir, A., Vilageliu, L., Gonzalez-Duarte, R. and Balcells, S. 2001. Spectrum of ABCA (ABCR) gene mutations in Spanish patients with autosomal recessive macular dystrophies. *Hum. Mutat.* 17: 504-510.

CHROMOSOMAL LOCATION

Genetic locus: ABCA4 (human) mapping to 1p22.1; Abca4 (mouse) mapping to 3 G1.

SOURCE

ABCA4 (H-235) is a rabbit polyclonal antibody raised against amino acids 1273-1508 of ABCA4 of human origin.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

ABCA4 (H-235) is recommended for detection of ABCA4 of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for ABCA4 siRNA (h): sc-41140, ABCA4 siRNA (m): sc-41141, ABCA4 shRNA Plasmid (h): sc-41140-SH, ABCA4 shRNA Plasmid (m): sc-41141-SH, ABCA4 shRNA (h) Lentiviral Particles: sc-41140-V and ABCA4 shRNA (m) Lentiviral Particles: sc-41141-V.

Molecular Weight of ABCA4: 220 kDa.

Positive Controls: mouse eye extract: sc-364241.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

For research use only, not for use in diagnostic procedures.

PROTOCOLS

See our web site at www.scbt.com or our catalog for detailed protocols and support products.



Try **ABCA4 (3F4): sc-65672**, our highly recommended monoclonal alternative to ABCA4 (H-235). Also, for AC, HRP, FITC, PE, Alexa Fluor® 488 and Alexa Fluor® 647 conjugates, see **ABCA4 (3F4): sc-65672**.