

AIPL1 (5-RY34): sc-134253

BACKGROUND

The inherited blindness associated protein, aryl hydrocarbon receptor interacting protein-like 1 (AIPL1), interacts with the cell cycle regulator protein NUB1. AIPL1 is crucial for protein folding and stabilization, as well as for protein trafficking. It localizes to the nucleus or cytoplasm and is highly expressed in the pineal gland and the retina. In the retina, AIPL1 is expressed in both developing cone and rod photoreceptors, but it is restricted to rod photoreceptors in the adult human retina. Defects in the gene encoding for AIPL1 can cause Leber congenital amaurosis type IV, an early-onset, inherited autosomal recessive disorder that results in childhood blindness.

REFERENCES

1. van der Spuy, J. and Cheetham, M.E. 2004. The Leber congenital amaurosis protein AIPL1 modulates the nuclear translocation of NUB1 and suppresses inclusion formation by NUB1 fragments. *J. Biol. Chem.* 279: 48038-48047.
2. van der Spuy, J. and Cheetham, M.E. 2004. Role of AIP and its homologue the blindness-associated protein AIPL1 in regulating client protein nuclear translocation. *Biochem. Soc. Trans.* 32: 643-645.
3. Dyer, M.A., et al. 2004. Retinal degeneration in AIPL1-deficient mice: a new genetic model of Leber congenital amaurosis. *Brain Res. Mol. Brain Res.* 132: 208-220.
4. Allikmets, R. 2004. Leber congenital amaurosis: a genetic paradigm. *Ophthalmic Genet.* 25: 67-79.
5. Silva, E., et al. 2004. A missense mutation in GUCY2D acts as a genetic modifier in RPE65-related Leber congenital amaurosis. *Ophthalmic Genet.* 25: 205-217.
6. Gallon, V.A., et al. 2004. Purification, characterisation and intracellular localisation of aryl hydrocarbon interacting protein-like 1 (AIPL1) and effects of mutations associated with inherited retinal dystrophies. *Biochim. Biophys. Acta.* 1690: 141-149.
7. Liu, X., et al. 2004. AIPL1, the protein that is defective in Leber congenital amaurosis, is essential for the biosynthesis of retinal rod cGMP phosphodiesterase. *Proc. Natl. Acad. Sci. USA* 101: 13903-13908.
8. Hanein, S., et al. 2005. Leber congenital amaurosis: comprehensive survey of genetic heterogeneity. A clinical definition update. *J. Fr. Ophthalmol.* 28: 98-105.
9. Galvin, J.A., et al. 2005. Clinical phenotypes in carriers of Leber congenital amaurosis mutations. *Ophthalmology* 112: 349-356.

CHROMOSOMAL LOCATION

Genetic locus: AIPL1 (human) mapping to 17p13.2.

SOURCE

AIPL1 (5-RY34) is a mouse monoclonal antibody raised against recombinant AIPL1 protein of human origin.

RESEARCH USE

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

PRODUCT

Each vial contains 100 µg IgG_{2a} kappa light chain in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

APPLICATIONS

AIPL1 (5-RY34) is recommended for detection of AIPL1 of 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)] and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Suitable for use as control antibody for AIPL1 siRNA (h): sc-60062, AIPL1 shRNA Plasmid (h): sc-60062-SH and AIPL1 shRNA (h) Lentiviral Particles: sc-60062-V.

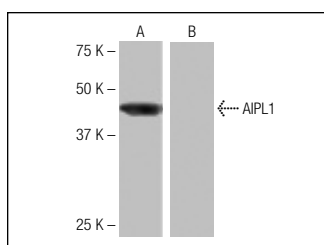
Molecular Weight of AIPL1: 43 kDa.

Positive Controls: AIPL1 transfected 293T whole cell lysate.

RECOMMENDED SUPPORT REAGENTS

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgGκ BP-HRP: sc-516102 or m-IgGκ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), 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).

DATA



AIPL1 (5-RY34): sc-134253. Western blot analysis of AIPL1 expression in human AIPL1 transfected (A) and non-transfected (B) 293T whole cell lysates.

STORAGE

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

PROTOCOLS

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