



RP2 (h2): 293T Lysate: sc-371779

BACKGROUND

RP2 (retinitis pigmentosa 2), also known as TBCCD2, is a 350 amino acid protein that localizes to the cytoplasmic side of the cell membrane and belongs to the TBCC family. Expressed ubiquitously, RP2 functions to stimulate the GTPase activity of tubulin and is thought to act as a guanine nucleotide dissociation inhibitor for ARL3 (ADP-ribosylation factor-like 3), preventing the GTP-bound form of ARL3 from dissociating. Via its ability to stimulate tubulin activity, RP2 plays an important role in retinal development. RP2 contains one C-CAP/cofactor C-like domain and can be myristoylated or palmitoylated, both of which are thought to be required for proper membrane targeting. Defects in the gene encoding RP2 are the cause of retinitis pigmentosa type 2 (RP2), a disorder characterized by the degeneration of photoreceptor cells, resulting in night vision blindness and an eventual loss of both peripheral and central vision.

REFERENCES

1. Thiselton, D.L., Hampson, R.M., Nayudu, M., Van Maldergem, L., Wolf, M.L., Saha, B.K., Bhattacharya, S.S. and Hardcastle, A.J. 1996. Mapping the RP2 locus for X-linked retinitis pigmentosa on proximal Xp: a genetically defined 5-cM critical region and exclusion of candidate genes by physical mapping. *Genome Res.* 6: 1093-1102.
2. Schwahn, U., Lenzner, S., Dong, J., Feil, S., Hinzmann, B., van Duijnhoven, G., Kirschner, R., Hemberger, M., Bergen, A.A., Rosenberg, T., Pinckers, A.J., Fundele, R., Rosenthal, A., Cremers, F.P., Ropers, H.H. and Berger, W. 1998. Positional cloning of the gene for X-linked retinitis pigmentosa 2. *Nat. Genet.* 19: 327-332.
3. Rosenberg, T., Schwahn, U., Feil, S. and Berger, W. 1999. Genotype-phenotype correlation in X-linked retinitis pigmentosa 2 (RP2). *Ophthalmic Genet.* 20: 161-172.
4. Chapple, J.P., Hardcastle, A.J., Grayson, C., Spackman, L.A., Willison, K.R. and Cheetham, M.E. 2000. Mutations in the N-terminus of the X-linked retinitis pigmentosa protein RP2 interfere with the normal targeting of the protein to the plasma membrane. *Hum. Mol. Genet.* 9: 1919-1926.
5. Thiselton, D.L., Zito, I., Plant, C., Jay, M., Hodgson, S.V., Bird, A.C., Bhattacharya, S.S. and Hardcastle, A.J. 2000. Novel frameshift mutations in the RP2 gene and polymorphic variants. *Hum. Mutat.* 15: 580.
6. Breuer, D.K., Yashar, B.M., Filippova, E., Hiriyan, S., Lyons, R.H., Mears, A.J., Asaye, B., Acar, C., Vervoort, R., Wright, A.F., Musarella, M.A., Wheeler, P., MacDonald, I., Iannaccone, A., Birch, D., et al. 2002. A comprehensive mutation analysis of RP2 and RPGR in a North American cohort of families with X-linked retinitis pigmentosa. *Am. J. Hum. Genet.* 70: 1545-1554.
7. Bartolini, F., Bhamidipati, A., Thomas, S., Schwahn, U., Lewis, S.A. and Cowan, N.J. 2002. Functional overlap between retinitis pigmentosa 2 protein and the tubulin-specific chaperone cofactor C. *J. Biol. Chem.* 277: 14629-14634.
8. Jin, Z.B., Liu, X.Q., Hayakawa, M., Murakami, A. and Nao-i, N. 2006. Mutational analysis of RPGR and RP2 genes in Japanese patients with retinitis pigmentosa: identification of four mutations. *Mol. Vis.* 12: 1167-1174.

CHROMOSOMAL LOCATION

Genetic locus: RP2 (human) mapping to Xp11.23.

PRODUCT

RP2 (h2): 293T Lysate represents a lysate of human RP2 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

RP2 (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive RP2 antibodies. Recommended use: 10-20 µl per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. 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 for detailed protocols and support products.