

RPGRIP1 siRNA (h): sc-92374

BACKGROUND

RPGRIP1 (retinitis pigmentosa GTPase regulator interacting protein 1), also known as LCA6, RGI1, RGRIP, CORD13, RPGRIP or RPGRIP1d, is a 1,286 amino acid protein that belongs to the RPGRIP1 family and localizes to the cilium. Expressed in retina, RPGRIP1 colocalizes with SEC16S in the outer segment of rod photoreceptors and cone outer segments. RPGRIP1 forms homodimers and elongated homopolymers, and exists six alternatively spliced isoforms. RPGRIP1 is required for SEC16S function and is essential for normal disk morphogenesis. Mutations in the gene encoding RPGRIP1 are the cause of Leber congenital amaurosis type 6 (LCA6) and cone-rod dystrophy type 13 (CORD13). LCA is considered the most common genetic cause of congenital visual impairment in infants and children. CORD13 is an inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination and initial loss of cone photoreceptors followed by rod degeneration.

REFERENCES

1. Boylan, J.P., et al. 2000. Identification of a novel protein interacting with RPGR. *Hum. Mol. Genet.* 9: 2085-2093.
2. Roepman, R., et al. 2000. The retinitis pigmentosa GTPase regulator (RPGR) interacts with novel transport-like proteins in the outer segments of rod photoreceptors. *Hum. Mol. Genet.* 9: 2095-2105.
3. Gerber, S., et al. 2001. Complete exon-intron structure of the RPGR-interacting protein (RPGRIP1) gene allows the identification of mutations underlying Leber congenital amaurosis. *Eur. J. Hum. Genet.* 9: 561-571.
4. Mavlyutov, T.A., et al. 2002. Species-specific subcellular localization of RPGR and RPGRIP1 isoforms: implications for the phenotypic variability of congenital retinopathies among species. *Hum. Mol. Genet.* 11: 1899-1907.
5. Hameed, A., et al. 2003. Evidence of RPGRIP1 gene mutations associated with recessive cone-rod dystrophy. *J. Med. Genet.* 40: 616-619.
6. Pawlyk, B.S., et al. 2010. Replacement gene therapy with a human RPGRIP1 sequence slows photoreceptor degeneration in a murine model of Leber congenital amaurosis. *Hum. Gene Ther.* 21: 993-1004.
7. Fernández-Martínez, L., et al. 2011. Evidence for RPGRIP1 gene as risk factor for primary open angle glaucoma. *Eur. J. Hum. Genet.* 19: 445-451.
8. Kuznetsova, T., et al. 2011. Structural organization and expression pattern of the canine RPGRIP1 isoforms in retinal tissue. *Invest. Ophthalmol. Vis. Sci.* 52: 2989-2998.
9. Busse, C., et al. 2011. Ophthalmic and cone derived electrodiagnostic findings in outbred Miniature Long-haired Dachshunds homozygous for a RPGRIP1 mutation. *Vet. Ophthalmol.* 14: 146-152.

CHROMOSOMAL LOCATION

Genetic locus: RPGRIP1 (human) mapping to 14q11.2.

PROTOCOLS

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

PRODUCT

RPGRIP1 siRNA (h) is a pool of 3 target-specific 19-25 nt siRNAs designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10 μ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see RPGRIP1 shRNA Plasmid (h): sc-92374-SH and RPGRIP1 shRNA (h) Lentiviral Particles: sc-92374-V as alternate gene silencing products.

For independent verification of RPGRIP1 (h) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-92374A, sc-92374B and sc-92374C.

STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20° C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20° C, avoid contact with RNAses and repeated freeze thaw cycles.

Resuspend lyophilized siRNA duplex in 330 μ l of the RNase-free water provided. Resuspension of the siRNA duplex in 330 μ l of RNase-free water makes a 10 μ M solution in a 10 μ M Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

APPLICATIONS

RPGRIP1 siRNA (h) is recommended for the inhibition of RPGRIP1 expression in human cells.

SUPPORT REAGENTS

For optimal siRNA transfection efficiency, Santa Cruz Biotechnology's siRNA Transfection Reagent: sc-29528 (0.3 ml), siRNA Transfection Medium: sc-36868 (20 ml) and siRNA Dilution Buffer: sc-29527 (1.5 ml) are recommended. Control siRNAs or Fluorescein Conjugated Control siRNAs are available as 10 μ M in 66 μ l. Each contain a scrambled sequence that will not lead to the specific degradation of any known cellular mRNA. Fluorescein Conjugated Control siRNAs include: sc-36869, sc-44239, sc-44240 and sc-44241. Control siRNAs include: sc-37007, sc-44230, sc-44231, sc-44232, sc-44233, sc-44234, sc-44235, sc-44236, sc-44237 and sc-44238.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor RPGRIP1 gene expression knockdown using RT-PCR Primer: RPGRIP1 (h)-PR: sc-92374-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

RESEARCH USE

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