

## LCA5L siRNA (m): sc-146668

### BACKGROUND

Leber congenital amaurosis (LCA) is one of the most common causes of hereditary blindness or severe visual impairment in infants. Mutations in several genes with diverse functions mapping to two loci have been implicated in LCA causation. These proteins are involved in processes such as photoreceptor development and maintenance, phototransduction, vitamin A metabolism and protein trafficking. LCA5, also known as Lebercilin, is a ciliary protein that is widely expressed during development and localizes to the connecting cilia of photoreceptors and to the microtubules, centrioles and primary cilia of cultured mammalian cells. The Leber congenital amaurosis 5-like protein (LCA5L) is a 670 amino acid protein that belongs to the LCA5 family.

### REFERENCES

1. Mohamed, M.D., Topping, N.C., Jafri, H., Raashed, Y., McKibbin, M.A. and Inglehearn, C.F. 2003. Progression of phenotype in Leber's congenital amaurosis with a mutation at the LCA5 locus. *Br. J. Ophthalmol.* 87: 473-475.
2. Gerber, S., Hanein, S., Perrault, I., Delphin, N., Aboussair, N., Leowski, C., Dufier, J.L., Roche, O., Munnich, A., Kaplan, J. and Rozet, J.M. 2007. Mutations in LCA5 are an uncommon cause of Leber congenital amaurosis (LCA) type II. *Hum. Mutat.* 28: 1245.
3. den Hollander, A.I., Koenekoop, R.K., Mohamed, M.D., Arts, H.H., Boldt, K., Towns, K.V., Sedmak, T., Beer, M., Nagel-Wolfrum, K., McKibbin, M., Dharmaraj, S., Lopez, I., Ivings, L., Williams, G.A., Springell, K., et al. 2007. Mutations in LCA5, encoding the ciliary protein lebercilin, cause Leber congenital amaurosis. *Nat. Genet.* 39: 889-895.
4. Ramprasad, V.L., Soumitra, N., Nancarrow, D., Sen, P., McKibbin, M., Williams, G.A., Arokiasamy, T., Lakshmipathy, P., Inglehearn, C.F. and Kumaramanickavel, G. 2008. Identification of a novel splice-site mutation in the Lebercilin (LCA5) gene causing Leber congenital amaurosis. *Mol. Vis.* 14: 481-486.
5. den Hollander, A.I., Roepman, R., Koenekoop, R.K. and Cremers, F.P. 2008. Leber congenital amaurosis: genes, proteins and disease mechanisms. *Prog. Retin. Eye Res.* 27: 391-419.
6. Jacobson, S.G., Aleman, T.S., Cideciyan, A.V., Sumaroka, A., Schwartz, S.B., Windsor, E.A., Swider, M., Herrera, W. and Stone, E.M. 2009. Leber congenital amaurosis caused by Lebercilin (LCA5) mutation: retained photoreceptors adjacent to retinal disorganization. *Mol. Vis.* 15: 1098-1106.
7. Seong, M.W., Kim, S.Y., Yu, Y.S., Hwang, J.M., Kim, J.Y. and Park, S.S. 2009. LCA5, a rare genetic cause of leber congenital amaurosis in Koreans. *Ophthalmic Genet.* 30: 54-55.

### CHROMOSOMAL LOCATION

Genetic locus: Lca5l (mouse) mapping to 16 C4.

### PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) for detailed protocols and support products.

### PRODUCT

LCA5L siRNA (m) is a target-specific 19-25 nt siRNA designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10  $\mu$ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see LCA5L shRNA Plasmid (m): sc-146668-SH and LCA5L shRNA (m) Lentiviral Particles: sc-146668-V as alternate gene silencing products.

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

### APPLICATIONS

LCA5L siRNA (m) is recommended for the inhibition of LCA5L expression in mouse 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  $\mu$ M in 66  $\mu$ 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 LCA5L gene expression knockdown using RT-PCR Primer: LCA5L (m)-PR: sc-146668-PR (20  $\mu$ 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.