# LMBR1 siRNA (h): sc-89636



The Power to Question

# **BACKGROUND**

LMBR1 (limb region 1), also known as TPT, ACHP, PPD2 or DIF14, is a 490 amino acid multi-pass membrane protein that is widely expressed with strongest expression in heart and pancreas. Belonging to the LIMR family, LMBR1 shares 95% sequence identity with the mouse protein and may play crucial role in the evolution of limb and skeletal system. LMBR1 is critical for expression of sonic hedgehog (Shh) in the developing posterior limb bud mesenchyme. Mutations in the gene encoding LMBR1 is the cause of several rare conditions such as acheiropody (ACHP) and syndactyly type 4 (SDYT4). ACHP is an autosomal recessive inherited disorder characterized by bilateral congenital amputations of the hands and feet. SDYT4, an autosomal dominant trait, is a very rare congenital distal limb malformation characterized by complete bilateral syndactyly, which involves all five digits. LMBR1 exists as three alternatively spliced isoforms.

# **REFERENCES**

- Clark, R.M., Marker, P.C., Roessler, E., Dutra, A., Schimenti, J.C., Muenke, M. and Kingsley, D.M. 2001. Reciprocal mouse and human limb phenotypes caused by gain- and loss-of-function mutations affecting Lmbr1. Genetics 159: 715-726.
- Sagai, T., Masuya, H., Tamura, M., Shimizu, K., Yada, Y., Wakana, S., Gondo, Y., Noda, T. and Shiroishi, T. 2004. Phylogenetic conservation of a limbspecific, cis-acting regulator of Sonic hedgehog (Shh). Mamm. Genome 15: 23-34.
- Sagai, T., Hosoya, M., Mizushina, Y., Tamura, M. and Shiroishi, T. 2005. Elimination of a long-range *cis*-regulatory module causes complete loss of limb-specific Shh expression and truncation of the mouse limb. Development 132: 797-803.
- Gurnett, C.A., Bowcock, A.M., Dietz, F.R., Morcuende, J.A., Murray, J.C. and Dobbs, M.B. 2007. Two novel point mutations in the long-range Shh enhancer in three families with triphalangeal thumb and preaxial polydactyly. Am. J. Med. Genet. A 143A: 27-32.
- Sato, D., Liang, D., Wu, L., Pan, Q., Xia, K., Dai, H., Wang, H., Nishimura, G., Yoshiura, K., Xia, J. and Niikawa, N. 2007. A syndactyly type IV locus maps to 7q36. J. Hum. Genet. 52: 561-564.
- He, F., Wu, D.D., Kong, Q.P. and Zhang, Y.P. 2008. Intriguing balancing selection on the intron 5 region of LMBR1 in human population. PLoS ONE 3: e2948.
- Semerci, C.N., Demirkan, F., Ozdemir, M., Biskin, E., Akin, B., Bagci, H. and Akarsu, N.A. 2009. Homozygous feature of isolated triphalangeal thumbpreaxial polydactyly linked to 7q36: no phenotypic difference between homozygotes and heterozygotes. Clin. Genet. 76: 85-90.
- 8. Li, H., Wang, C.Y., Wang, J.X., Wu, G.S., Yu, P., Yan, X.Y., Chen, Y.G., Zhao, L.H. and Zhang, Y.P. 2009. Mutation analysis of a large Chinese pedigree with congenital preaxial polydactyly. Eur. J. Hum. Genet. 17: 604-610.

# **PROTOCOLS**

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

#### **CHROMOSOMAL LOCATION**

Genetic locus: LMBR1 (human) mapping to 7q36.3.

## **PRODUCT**

LMBR1 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  $\mu\text{M}$  solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see LMBR1 shRNA Plasmid (h): sc-89636-SH and LMBR1 shRNA (h) Lentiviral Particles: sc-89636-V as alternate gene silencing products.

For independent verification of LMBR1 (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-89636A, sc-89636B and sc-89636C.

#### STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20 $^{\circ}$  C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20 $^{\circ}$  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**

LMBR1 siRNA (h) is recommended for the inhibition of LMBR1 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 µM in 66 µl. 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 LMBR1 gene expression knockdown using RT-PCR Primer: LMBR1 (h)-PR: sc-89636-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.

Santa Cruz Biotechnology, Inc. 1.800.457.3801 831.457.3801 Fax 831.457.3801 Europe +00800 4573 8000 49 6221 4503 0 www.scbt.com