

Dymeclin siRNA (h): sc-60558

BACKGROUND

Dyggve-Melchior-Clausen syndrome (DMC), a rare autosomal recessive disorder, is characterized by microcephaly, short trunk dwarfism and sometime psychomotor retardation. Cutaneous cells of affected individuals show dilated rough endoplasmic reticulum and enlarged vacuoles. The Dyggve-Melchior-Clausen syndrome protein, also designated Dymeclin, may play a role in proteoglycan metabolism and intracellular protein digestion. It is a widely expressed multi-pass membrane protein, detected primarily in chondrocytes and fetal brain tissue. Defects in Dymeclin are also the cause of Smith-McCort dysplasia syndrome (SMC), which has characteristics identical to those of Dyggve-Melchior-Clausen syndrome.

REFERENCES

1. El Ghouzi, V., et al. 2003. Mutations in a novel gene Dymeclin (FLJ20071) are responsible for Dyggve-Melchior-Clausen syndrome. *Hum. Mol. Genet.* 12: 357-364.
2. Paupe, V., et al. 2004. Recent advances in Dyggve-Melchior-Clausen syndrome. *Mol. Genet. Metab.* 83: 51-59.
3. Kinning, E., et al. 2005. Genomic duplic in an autosomal recessive disorder. *J. Med. Genet.* 42: e70.
4. Pogue, R., et al. 2005. Probable identity-by-descent for a mutation in the Dyggve-Melchior-Clausen/Smith-McCort dysplasia (Dymeclin) gene among patients from Guam, Chile, Argentina, and Spain. *Am. J. Med. Genet. A* 138: 75-78.
5. Geneviève, D., et al. 2005. Exclusion of the Dymeclin and PAPSS2 genes in a novel form of spondyloepimetaphyseal dysplasia and mental retardation. *Eur. J. Hum. Genet.* 13: 541-546.

CHROMOSOMAL LOCATION

Genetic locus: DYM (human) mapping to 18q21.1.

PRODUCT

Dymeclin 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 Dymeclin shRNA Plasmid (h): sc-60558-SH and Dymeclin shRNA (h) Lentiviral Particles: sc-60558-V as alternate gene silencing products.

For independent verification of Dymeclin (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-60558A, sc-60558B and sc-60558C.

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.

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

Dymeclin siRNA (h) is recommended for the inhibition of Dymeclin 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 Dymeclin gene expression knockdown using RT-PCR Primer: Dymeclin (h)-PR: sc-60558-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.