

Dymeclin (N-15): sc-49942

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., Dagonneau, N., Kinning, E., Thauvin-Robinet, C., Chemaitilly, W., Prost-Squarcioni, C., Al-Gazali, L.I., Verloes, A., Le Merrer, M., Munnich, A., Trembath, R.C. and Cormier-Daire, V. 2003. Mutations in a novel gene Dymeclin (FLJ20071) are responsible for Dyggve-Melchior-Clausen syndrome. *Hum. Mol. Genet.* 12: 357-364.
2. Paupe, V., Gilbert, T., Le Merrer, M., Munnich, A., Cormier-Daire, V. and El Ghouzi, V. 2004. Recent advances in Dyggve-Melchior-Clausen syndrome. *Mol. Genet. Metab.* 83: 51-59.
3. Kinning, E., Tufarelli, C., Winship, W.S., Aldred, M.A. and Trembath, R.C. 2005. Genomic duplications in an autosomal recessive disorder. *J. Med. Genet.* 42: e70.
4. Pogue, R., Ehteshami, N., Repetto, G.M., Carrero-Valenzuela, R., de Casella, C.B., de Pons, S.P., Martínez-Frías, M.L., Heuertz, S., Cormier-Daire, V. and Cohn, D.H. 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. Part A* 138: 75-78.
5. Geneviève, D., Heron, D., El Ghouzi, V., Prost-Squarcioni, C., Le Merrer, M., Jacquette, A., Sanlaville, D., Pinton, F., Villeneuve, N., Kalifa, G., Munnich, A. and Cormier-Daire, V. 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; Dym (mouse) mapping to 18 E3.

SOURCE

Dymeclin (N-15) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of Dymeclin of human origin.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-49942 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

Dymeclin (N-15) is recommended for detection of Dymeclin of mouse, rat and human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000).

Dymeclin (N-15) is also recommended for detection of Dymeclin in additional species, including equine, canine, bovine and avian.

Suitable for use as control antibody for Dymeclin siRNA (h): sc-60558, Dymeclin siRNA (m): sc-60559, Dymeclin shRNA Plasmid (h): sc-60558-SH, Dymeclin shRNA Plasmid (m): sc-60559-SH, Dymeclin shRNA (h) Lentiviral Particles: sc-60558-V and Dymeclin shRNA (m) Lentiviral Particles: sc-60559-V.

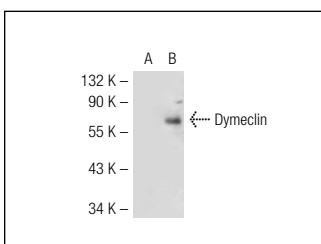
Molecular Weight of Dymeclin: 75 kDa.

Positive Controls: Dymeclin (h2): 293T Lysate: sc-116965, HeLa whole cell lysate: sc-2200 or mouse brain extract: sc-2253.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

DATA



Dymeclin (N-15): sc-49942. Western blot analysis of Dymeclin expression in non-transfected: sc-117752 (A) and human Dymeclin transfected: sc-116965 (B) 293T whole cell lysates.

STORAGE

Store at 4° C, **DO NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

RESEARCH USE

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