



Dymeclin (h2): 293T Lysate: sc-116965

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., Dagoneau, 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 duplic 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. 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.

PRODUCT

Dymeclin (h2): 293T Lysate represents a lysate of human Dymeclin transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

PROTOCOLS

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

APPLICATIONS

Dymeclin (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive Dymeclin antibodies. Recommended use: 10-20 µl per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

RESEARCH USE

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