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emerin (h): 293T Lysate: sc-128526

BACKGROUND

Emerin is believed to be a member of the nuclear lamina associated protein family. It is ubiquitously expressed and localized to the nuclear membrane in normal cells. Mutations of the gene that encodes emerin result in the X-linked recessive disease Emery-Dreifuss muscular dystrophy (EDMD), which is characterized by slowly progressing contractures, skeletal muscle wasting and cardiomyopathy. Research has demonstrated that the lack of emerin expression is one cause of EDMD. Emerin is involved in the association of the nuclear membrane with the lamina, and is localized specifically to desmosomes and fascia adherentes in the heart. This may account for conduction defects in patients with EDMD.

REFERENCES

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2. Bione, S., Small, K., Aksmanovic, V.M., D'Urso, M., Ciccodicola, A., Merlini, L., Morandi, L., Kress, W., Yates, J.R., Warren, S.T., et al. 1995. Identification of new mutations in the Emery-Dreifuss muscular dystrophy gene and evidence for genetic heterogeneity of the disease. *Hum. Mol. Genet.* 4: 1859-1863.
3. Cartegni, L., di Barletta, M.R., Barresi, R., Squarzone, S., Sabatelli, P., Maraldi, N., Mora, M., Di Blasi, C., Cornelio, F., Merlini, L., Villa, A., Cobiauchi, F. and Toniolo, D. 1997. Heart-specific localization of emerin: new insights into Emery-Dreifuss muscular dystrophy. *Hum. Mol. Genet.* 6: 2257-2264.
4. Kubo, S., Tsukahara, T. and Arahata, K. 1997. Emery-Dreifuss muscular dystrophy. *Nippon Rinsho* 55: 3186-3189.
5. Small, K. and Warren, S.T. 1998. Emerin deletions occurring on both Xq28 inversion backgrounds. *Hum. Mol. Genet.* 7: 135-139.

CHROMOSOMAL LOCATION

Genetic locus: EMD (human) mapping to Xq28.

PRODUCT

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

APPLICATIONS

emerin (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive emerin 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.

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.

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.