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



The Power to Question

## **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-Dreyfuss 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 fasciae adherentes in the heart. This may account for conduction defects in patients with EDMD.

## **REFERENCES**

- Bione, S., Maestrini, E., Rivella, S., Mancini, M., Regis, S., Romeo, G. and Toniolo, D. 1994. Identification of a novel X-linked gene responsible for Emery-Dreifuss muscular dystrophy. Nat. Genet. 8: 323-327.
- 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.
- Cartegni, L., di Barletta, M.R., Barresi, R., Squarzoni, S., Sabatelli, P., Maraldi, N., Mora, M., Di Blasi, C., Cornelio, F., Merlini, L., Villa, A., Cobianchi, F. and Toniolo, D. 1997. Heart-specific localization of emerin: new insights into Emery-Dreifuss muscular dystrophy. Hum. Mol. Genet. 6: 2257-2264.
- Kubo, S., Tsukahara, T. and Arahata, K. 1997. Emery-Dreifuss muscular dystrophy. Nippon Rinsho 55: 3186-3189.
- 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.

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