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SANTA CRUZ BIOTECHNOLOGY, INC.

Dyskerin (h4): 293T Lysate: sc-170686

BACKGROUND

Dyskerin (NAP57) associates with the chaperone protein Nopp140 and forms a small ribonucleoprotein particle with GAR1 (NOLA1), NHP2 (NOLA2) and Nop10 for the isomerization of uridine to pseudouridine. GAR1, NHP2 and Dyskerin localize to the dense fibrillar component of the nucleolus and in nuclear Cajal bodies. The Dyskerin gene maps to chromosome Xq28. Missense mutations in the Dyskerin gene interfere with normal nuclear localization of Dyskerin and cause Dyskeratosis congenita (DKC). DKC is a rare, X-linked bone marrow disorder characterized by cutaneous hyperpigmentation, dystrophy of the nails, atrophy of the testicles and leukoplakia of the oral mucosa. The GAR1 gene maps to chromosome 4q25 and encodes a 28 kDa protein. The NHP2 gene maps to chromosome 5q35.3 and encodes a 155 amino acid protein.

REFERENCES

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2. Heiss, N.S., Girod, A., Salowsky, R., Wiemann, S., Pepperkok, R. and Poustka, A. 1999. Dyskerin localizes to the nucleolus and its mislocalization is unlikely to play a role in the pathogenesis of dyskeratosis congenita. Hum. Mol. Genet. 8: 2515-2524.
3. Dragon, F., Pogacic, V. and Filipowicz, W. 2000. *In vitro* assembly of human H/ACA small nucleolar RNPs reveals unique features of U17 and telomerase RNAs. Mol. Cell. Biol. 20: 3037-3048.
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5. Wang, C., Query, C.C. and Meier, U.T. 2002. Immunopurified small nucleolar ribonucleoprotein particles pseudouridylate rRNA independently of their association with phosphorylated Nopp140. Mol. Cell. Biol. 22: 8457-8466.
6. LocusLink Report (LocusID: 54433). http://www.ncbi.nlm.nih.gov/LocusLink/

CHROMOSOMAL LOCATION

Genetic locus: DKC1 (human) mapping to Xq28.

PRODUCT

Dyskerin (h4): 293T Lysate represents a lysate of human Dyskerin transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

Dyskerin (h4): 293T Lysate is suitable as a Western Blotting positive control for human reactive Dyskerin 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.