

BUD13 (h): 293T Lysate: sc-174350

BACKGROUND

BUD13 is a 619 amino acid protein that belongs to the CWC26 family and exists as 2 alternatively spliced isoforms that are encoded by a gene located on human chromosome 11q23.3. Chromosome 11 comprises approximately 4% of human genomic DNA and is considered a gene and disease association dense chromosome. The chromosome 11 encoded Atm gene is important for regulation of cell cycle arrest and apoptosis following double strand DNA breaks. Atm mutation leads to the disorder known as ataxia-telangiectasia. The blood disorders Sickle cell anemia and thalassemia are caused by HBB gene mutations, while Wilms' tumors, WAGR syndrome and Denys-Drash syndrome are associated with mutations of the WT1 gene. Jervell and Lange-Nielsen syndrome, Jacobsen syndrome, Niemann-Pick disease, hereditary angioedema and Smith-Lemli-Opitz syndrome are also associated with defects in chromosome 11-encoded genes.

REFERENCES

1. Fabiani, J.E., Avigliano, A., Dupont, J.C. and Fabiana, J.E. 2000. Hereditary angioedema. Long-term follow-up of 88 patients. Experience of the Argentine allergy and immunology institute. *Allergol. Immunopathol.* 28: 267-271.
2. Jira, P.E., Waterham, H.R., Wanders, R.J., Smeitink, J.A., Sengers, R.C. and Wevers, R.A. 2003. Smith-lemli-opitz syndrome and the DHCR7 gene. *Ann. Hum. Genet.* 67: 269-280.
3. Schuchman, E.H. 2007. The pathogenesis and treatment of acid sphingomyelinase-deficient Niemann-Pick disease. *J. Inherit. Metab. Dis.* 30: 654-663.
4. Siem, G., Früh, A., Leren, T.P., Heimdal, K., Teig, E. and Harris, S. 2008. Jervell and Lange-Nielsen syndrome in Norwegian children: aspects around cochlear implantation, hearing, and balance. *Ear Hear.* 29: 261-269.
5. Bhuiyan, Z.A., Momenah, T.S., Amin, A.S., Al-Khadra, A.S., Alders, M., Wilde, A.A. and Mannens, M.M. 2008. An intronic mutation leading to incomplete skipping of exon-2 in KCNQ1 rescues hearing in Jervell and Lange-Nielsen syndrome. *Prog. Biophys. Mol. Biol.* 98: 319-327.
6. Coldren, C.D., Lai, Z., Shragg, P., Rossi, E., Glidewell, S.C., Zuffardi, O., Mattina, T., Ivy, D.D., Curfs, L.M., Mattson, S.N., Riley, E.P., Treier, M. and Grossfeld, P.D. 2009. Chromosomal microarray mapping suggests a role for BSX and Neurogranin in neurocognitive and behavioral defects in the 11q terminal deletion disorder (Jacobsen syndrome). *Neurogenetics* 10: 89-95.

CHROMOSOMAL LOCATION

Genetic locus: BUD13 (human) mapping to 11q23.3.

PRODUCT

BUD13 (h): 293T Lysate represents a lysate of human BUD13 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.

APPLICATIONS

BUD13 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive BUD13 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.

PROTOCOLS

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