SANTA CRUZ BIOTECHNOLOGY, INC.

DGCR14 (h2): 293T Lysate: sc-128453

BACKGROUND

DGCR14 (DiGeorge syndrome critical region 14, ES2 protein) is a 476 amino acid nuclear protein that belongs to the DGCR14 family. DGCR14 is believed to play a part in the etiology of the velocardiofacial/DiGeorge syndrome (VCFS/DGS), a developmental disorder characterized by structural and functional palate anomalies, conotruncal cardiac malformations, immunodeficiency, hypocalcemia and typical facial anomalies. Most cases result from a deletion of chromosome 22q11.2 (DiGeorge syndrome chromosome region, or DGCR). This protein localizes to the nucleus and co-purifies with C complex spliceosomes.

REFERENCES

1. Rizzu, P., Lindsay, E.A., Taylor, C., O'Donnell, H., Levy, A., Scambler, P. and Baldini, A. 1996. Cloning and comparative mapping of a gene from the commonly deleted region of DiGeorge and Velocardiofacial syndromes conserved in *C. elegans*. Mamm. Genome 7: 639-643.
2. Gong, W., Emanuel, B.S., Galili, N., Kim, D.H., Roe, B., Driscoll, D.A. and Budarf, M.L. 1997. Structural and mutational analysis of a conserved gene (DGSI) from the minimal DiGeorge syndrome critical region. Hum. Mol. Genet. 6: 267-276.
3. Chieffo, C., Garvey, N., Gong, W., Roe, B., Zhang, G., Silver, L., Emanuel, B.S. and Budarf, M.L. 1997. Isolation and characterization of a gene from the DiGeorge chromosomal region homologous to the mouse Tbx1 gene. Genomics 43: 267-277.
4. Lindsay, E.A., Harvey, E.L., Scambler, P.J. and Baldini, A. 1998. ES2, a gene deleted in DiGeorge syndrome, encodes a nuclear protein and is expressed during early mouse development, where it shares an expression domain with a Goosecoid-like gene. Hum. Mol. Genet. 7: 629-635.
5. Wakamiya, M., Lindsay, E.A., Rivera-Perez, J.A., Baldini, A. and Behringer, R.R. 1998. Functional analysis of Gscl in the pathogenesis of the DiGeorge and velocardiofacial syndromes. Hum. Mol. Genet. 7: 1835-1840.
6. Hoogendoorn, B., Coleman, S.L., Guy, C.A., Smith, S.K., O'Donovan, M.C. and Buckland, P.R. 2004. Functional analysis of polymorphisms in the promoter regions of genes on 22q11. Hum. Mutat. 24: 35-42.
7. Wang, H., Duan, S., Du, J., Li, X., Xu, Y., Zhang, Z., Wang, Y., Huang, G., Feng, G. and He, L. 2006. Transmission disequilibrium test provides evidence of association between promoter polymorphisms in 22q11 gene DGCR14 and schizophrenia. J. Neural Transm. 113: 1551-1561.

CHROMOSOMAL LOCATION

Genetic locus: DGCR14 (human) mapping to 22q11.21.

PRODUCT

DGCR14 (h2): 293T Lysate represents a lysate of human DGCR14 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

DGCR14 (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive DGCR14 antibodies.

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.