



DAX-1 (h): 293 Lysate: sc-158427

BACKGROUND

Adrenal hypoplasia congenita (AHC) is an X-linked disorder characterized by primary adrenal insufficiency. The disorder, which is lethal if untreated, results in adrenal insufficiency early in infancy and is characterized by low serum concentration of glucocorticoids, mineralocorticoids and androgens and failure to respond to ACTH. AHC has been mapped to chromosome Xp21 at the same or close to an X-linked locus involved in sex determination, DSS (for dosage-sensitive sex reversal). The gene corresponding to DSS and AHC (designated DAX-1 for DSS-AHC critical region on the X chromosome, gene 1) has been cloned and shown to be deleted in AHC deletion patients and mutated in AHC non-deletion patients. The carboxy terminal 250 amino acids of the DAX-1-encoded protein, DAX-1, exhibits approximately 50% continuous similarity to the ligand-binding domain of the members of the nuclear hormone receptor superfamily while the amino terminal domain contains a putative DNA-binding motif. DAX-1 binds to retinoic acid responsive elements and down regulates retinoic acid receptor-mediated transcriptional activation.

REFERENCES

1. Walker, A.P., et al. 1993. Isolation of the human Xp21 glycerol kinase gene by positional cloning. *Hum. Molec. Genet.* 2: 107-114.
2. Worley, K.C., et al. 1993. Yeast artificial chromosome cloning in the glycerol kinase and adrenal hypoplasia congenita region of Xp21. *Genomics* 16: 407-416.
3. Bardoni, B., et al. 1994. A dosage sensitive locus at chromosome Xp21 is involved in male to female sex reversal. *Nature Genet.* 7: 497-501.
4. Zanaria, E., et al. 1994. An unusual member of the nuclear hormone receptor superfamily responsible for X-linked adrenal hypoplasia congenita. *Nature* 372: 635-641.
5. Muscatelli, F., et al. 1994. Mutations in the DAX-1 gene give rise to both X-linked adrenal hypoplasia congenita and hypogonadotropic hypogonadism. *Nature* 372: 672-676.

CHROMOSOMAL LOCATION

Genetic locus: NROB1 (human) mapping to Xp21.2.

PRODUCT

DAX-1 (h): 293 Lysate represents a lysate of human DAX-1 transfected 293 cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

DAX-1 (h): 293 Lysate is suitable as a Western Blotting positive control for human reactive DAX-1 antibodies. Recommended use: 10-20 µl per lane.

Control 293 Lysate: sc-110760 is available as a Western Blotting negative control lysate derived from non-transfected 293 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.