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FAM46D (h2): 293T Lysate: sc-175037

BACKGROUND

The X and Y chromosomes are the human sex chromosomes. Chromosome X consists of about 153 million base pairs and nearly 1,000 genes. The combination of an X and Y chromosome lead to normal male development while two copies of X lead to normal female development. There are a number of conditions related to an unusual number and combination of sex chromosomes being inherited. More than one copy of the X chromosome with a Y chromosome causes Klinefelter's syndrome. A single copy of X alone leads to Turner's syndrome. More than two copies of the X chromosome, in the absence of a Y chromosome, is known as Triple X syndrome. Color blindness, hemophilia, and Duchenne muscular dystrophy are well known X chromosome-linked conditions which affect males more frequently as males carry a single X chromosome. The FAM46D gene product has been provisionally designated FAM46D pending further characterization.

REFERENCES

1. Givens, J.R., Wilroy, R.S., Summitt, R.L., Andersen, R.N., Wiser, W.L. and Fish, S.A. 1975. Features of Turner's syndrome in women with polycystic ovaries. *Obstet. Gynecol.* 45: 619-624.
2. Bernardino-Sgherri, J., Flagiello, D. and Dutrillaux, B. 2002. Overall DNA methylation and chromatin structure of normal and abnormal X chromosomes. *Cytogenet. Genome Res.* 99: 85-91.
3. Ozcelik, T. 2002. Uncovering the complex mysteries of mosaicism. *Nature* 417: 588.
4. Muntoni, F., Torelli, S. and Ferlini, A. 2003. Dystrophin and mutations: one gene, several proteins, multiple phenotypes. *Lancet Neurol.* 2: 731-740.
5. Deeb, S.S. 2005. The molecular basis of variation in human color vision. *Clin. Genet.* 67: 369-377.
6. Bojesen, A., Kristensen, K., Birkebaek, N.H., Fedder, J., Mosekilde, L., Bennett, P., Laurberg, P., Frystyk, J., Flyvbjerg, A., Christiansen, J.S. and Gravholt, C.H. 2006. The metabolic syndrome is frequent in Klinefelter's syndrome and is associated with abdominal obesity and hypogonadism. *Diabetes Care* 29: 1591-1598.
7. Hayashi, T., Kubo, A., Takeuchi, T., Gekka, T., Goto-Omoto, S. and Kitahara, K. 2006. Novel form of a single X-linked visual pigment gene in a unique dichromatic color-vision defect. *Vis. Neurosci.* 23: 411-417.
8. Augui, S., Fillion, G.J., Huart, S., Nora, E., Guggiari, M., Maresca, M., Stewart, A.F. and Heard, E. 2007. Sensing X chromosome pairs before X inactivation via a novel X-pairing region of the Xic. *Science* 318: 1632-1636.
9. Rolle, U., Linse, B., Glasow, S., Sandig, K.R., Richter, T. and Till, H. 2007. Duodenal atresia in an infant with triple-X syndrome: a new associated malformation in 47,XXX. *Birth Defects Res. Part A Clin. Mol. Teratol.* 79: 612-613.

CHROMOSOMAL LOCATION

Genetic locus: FAM46D (human) mapping to Xq21.1.

RESEARCH USE

For research use only, not for use in diagnostic procedures.

PRODUCT

FAM46D (h2): 293T Lysate represents a lysate of human FAM46D transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

APPLICATIONS

FAM46D (h2): 293T Lysate is suitable as a Western Blotting positive control for human reactive FAM46D 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.

PROTOCOLS

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