



FAM47A (K-18): sc-246721

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 FAM47A gene product has been provisionally designated FAM47A pending further characterization.

REFERENCES

- Givens, J.R., Wilroy, R.S., Summitt, R.L., Andersen, R.N., Wisner, W.L. and Fish, S.A. 1975. Features of Turner's syndrome in women with polycystic ovaries. *Obstet. Gynecol.* 45: 619-624.
- Bernardino-Sgheri, 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.
- Ozcelik, T. 2002. Uncovering the complex mysteries of mosaicism. *Nature* 417: 588.
- Muntoni, F., Torelli, S. and Ferlini, A. 2003. Dystrophin and mutations: one gene, several proteins, multiple phenotypes. *Lancet Neurol.* 2: 731-740.
- Deeb, S.S. 2005. The molecular basis of variation in human color vision. *Clin. Genet.* 67: 369-377.
- 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.
- 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.
- 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.
- 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: FAM47A (human) mapping to Xp21.1.

SOURCE

FAM47A (K-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of FAM47A of human origin.

PRODUCT

Each vial contains 200 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-246721 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

FAM47A (K-18) is recommended for detection of FAM47A of human origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000); non cross-reactive with FAM47B, FAM47C or FAM47E.

Suitable for use as control antibody for FAM47A siRNA (h): sc-91251, FAM47A shRNA Plasmid (h): sc-91251-SH and FAM47A shRNA (h) Lentiviral Particles: sc-91251-V.

Molecular Weight of FAM47A: 91 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat IgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat IgG-HRP: sc-2033 (dilution range: 1:2000-1:5000), Cruz Marker™ Molecular Weight Standards: sc-2035, TBS Blotto A Blocking Reagent: sc-2333 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use donkey anti-goat IgG-FITC: sc-2024 (dilution range: 1:100-1:400) or donkey anti-goat IgG-TR: sc-2783 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

STORAGE

Store at 4° C, ****DO NOT FREEZE****. Stable for one year from the date of shipment. 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 or our catalog for detailed protocols and support products.