FAM46D (M-12): sc-139251



The Power to Question

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 unsual number and combinations 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 2 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

- Givens, J.R., et al. 1975. Features of Turner's syndrome in women with polycystic ovaries. Obstet. Gynecol. 45: 619-624.
- Bernardino-Sgherri, J., et al. 2002. Overall DNA methylation and chromatin structure of normal and abnormal X chromosomes. Cytogenet. Genome Res. 99: 85-91.
- 3. Ozçelik, T. 2002. Uncovering the complex mysteries of mosaicism. Nature 417: 588.
- Muntoni, F., et al. 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., et al. 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., et al. 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., et al. 2007. Sensing X chromosome pairs before X inactivation via a novel X-pairing region of the Xic. Science 318: 1632-1636.
- Rolle, U., et al. 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 (mouse) mapping to X A1.1.

SOURCE

FAM46D (M-12) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the C-terminus of FAM46D of mouse origin.

STORAGE

Store at 4° C, **D0 NOT FREEZE**. Stable for one year from the date of shipment. Non-hazardous. No MSDS required.

PRODUCT

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

Blocking peptide available for competition studies, sc-139251 P, (100 μ g peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

FAM46D (M-12) is recommended for detection of FAM46D of mouse and rat 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 FAM46A, FAM46B or FAM46C.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use donkey anti-goat lgG-HRP: sc-2020 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible donkey anti-goat lgG-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 lgG-FITC: sc-2024 (dilution range: 1:100-1:400) with UltraCruz™ Mounting Medium: sc-24941.

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.

Santa Cruz Biotechnology, Inc. 1.800.457.3801 831.457.3800 fax 831.457.3801 **Europe** +00800 4573 8000 49 6221 4503 0 **www.scbt.com**