

# FAM48B1 (P-16): sc-246733

## BACKGROUND

FAM38B1 is a 823 amino acid protein that is expressed by a gene that maps to human chromosome X. 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 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.

## REFERENCES

1. Givens, J.R., et al. 1975. Features of Turner's syndrome in women with polycystic ovaries. *Obstet. Gynecol.* 45: 619-624.
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3. Ozelik, T. 2002. Uncovering the complex mysteries of mosaicism. *Nature* 417: 588.
4. Muntoni, F., et al. 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., 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.
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## CHROMOSOMAL LOCATION

Genetic locus: SUPT20HL1 (human) mapping to Xp22.11.

## SOURCE

FAM48B1 (P-16) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of FAM48B1 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-246733 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

FAM48B1 (P-16) is recommended for detection of FAM48B1 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).

Molecular Weight of FAM48B1: 89 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](http://www.scbt.com) or our catalog for detailed protocols and support products.