

# FAM136A/B (D-17): sc-246575

## BACKGROUND

The second largest human chromosome, 2 consists of 237 million bases encoding over 1,400 genes and making up approximately 8% of the human genome. A number of genetic diseases are linked to genes on chromosome 2. Harlequin ichthyosis, a rare and morbid skin deformity, is associated with mutations in the ABCA12 gene. The lipid metabolic disorder sitosterolemia is associated with ABCG5 and ABCG8. An extremely rare recessive genetic disorder, Alström syndrome is due to mutations in the ALMS1 gene. Interestingly, chromosome 2 contains what appears to be a vestigial second centromere and vestigial telomeres which gives credence to the hypothesis that human chromosome 2 is the result of an ancient fusion of two ancestral chromosomes seen in modern form today in apes. The FAM136A gene product has been provisionally designated FAM136A pending further characterization.

## REFERENCES

1. Ijdo, J.W., et al. 1991. Origin of human chromosome 2: an ancestral telomere-telomere fusion. *Proc. Natl. Acad. Sci. USA* 88: 9051-9055.
2. Avarello, R., et al. 1992. Evidence for an ancestral alphoid domain on the long arm of human chromosome 2. *Hum. Genet.* 89: 247-249.
3. Hillier, L.W., et al. 2005. Generation and annotation of the DNA sequences of human chromosomes 2 and 4. *Nature* 434: 724-731.
4. Thomas, A.C., et al. 2006. ABCA12 is the major harlequin ichthyosis gene. *J. Invest. Dermatol.* 126: 2408-2413.
5. Akiyama, M., et al. 2007. Compound heterozygous ABCA12 mutations including a novel nonsense mutation underlie harlequin ichthyosis. *Dermatology* 215: 155-159.

## CHROMOSOMAL LOCATION

Genetic locus: FAM136A (human) mapping to 2p13.3, FAM136BP (human) mapping to 6p25.2.

## SOURCE

FAM136A/B (D-17) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of FAM136A 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-246575 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## 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.

## APPLICATIONS

FAM136A/B (D-17) is recommended for detection of FAM136A and FAM136B of human origin and FAM136A of rat origin by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000), immunoprecipitation [1-2 µg per 100-500 µg of total protein (1 ml of cell lysate)], 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).

FAM136A/B (D-17) is also recommended for detection of FAM136A and FAM136B additional species, including equine, canine and porcine.

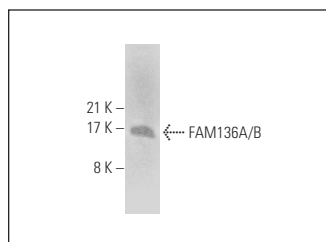
Molecular Weight of FAM136A/B: 16 kDa.

Positive Controls: Jurkat whole cell lysate: sc-2204.

## 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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) 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.

## DATA



FAM136A/B (D-17): sc-246575. Western blot analysis of FAM136A/B expression in Jurkat whole cell lysate.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) or our catalog for detailed protocols and support products.