

# 4930417G10Rik (D-17): sc-246839

## 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 FLJ30851 gene product has been provisionally designated FLJ30851 pending further characterization.

## REFERENCES

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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.
6. Marshall, J.D., et al. 2007. Alström syndrome. *Eur. J. Hum. Genet.* 15: 1193-1202.
7. Marshall, J.D., et al. 2007. Spectrum of ALMS1 variants and evaluation of genotype-phenotype correlations in Alström syndrome. *Hum. Mutat.* 28: 1114-1123.
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## CHROMOSOMAL LOCATION

Genetic locus: Fam228a (mouse) mapping to 12 A1.1.

## SOURCE

4930417G10Rik (D-17) is an affinity purified goat polyclonal antibody raised against a peptide mapping near the N-terminus of 4930417G10Rik of mouse 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-246839 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

4930417G10Rik (D-17) is recommended for detection of 4930417G10Rik of mouse origin and the corresponding rat homolog 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).

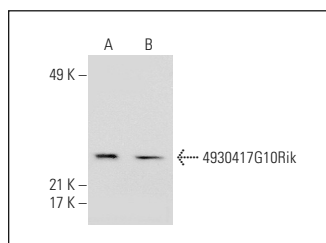
Suitable for use as control antibody for 4930417G10Rik siRNA (m): sc-140040, 4930417G10Rik shRNA Plasmid (m): sc-140040-SH and 4930417G10Rik shRNA (m) Lentiviral Particles: sc-140040-V.

Positive Controls: KNRK whole cell lysate: sc-2214 or RAW 264.7 whole cell lysate: sc-2211.

## 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.

## DATA



4930417G10Rik (D-17): sc-246839. Western blot analysis of 4930417G10Rik expression in KNRK (A) and RAW 264.7 (B) whole cell lysates.

## 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.