

FER1L5 (C-12): sc-139352

BACKGROUND

Chromosome 2, the second largest human chromosome, consists of 237 million bases encoding over 1,400 genes and makes 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 FER1L5 gene product has been provisionally designated FER1L5 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.
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.
8. Tabas, I. 2007. A two-carbon switch to sterol-induced autophagic death. *Autophagy* 3: 38-41.
9. Wang, D.Q. 2007. Regulation of intestinal cholesterol absorption. *Annu. Rev. Physiol.* 69: 221-248.

CHROMOSOMAL LOCATION

Genetic locus: FER1L5 (human) mapping to 2q11.2.

SOURCE

FER1L5 (C-12) is an affinity purified rabbit polyclonal antibody raised against a peptide mapping at the C-terminus of FER1L5 of human origin.

PRODUCT

Each vial contains 100 µg IgG in 1.0 ml of PBS with < 0.1% sodium azide and 0.1% gelatin.

Blocking peptide available for competition studies, sc-139352 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

FER1L5 (C-12) is recommended for detection of FER1L5 of human origin by Western Blotting (starting dilution 1:100, dilution range 1:50-1:500), immunofluorescence (starting dilution 1:25, dilution range 1:25-1:250) and solid phase ELISA (starting dilution 1:30, dilution range 1:30-1:3000); non cross-reactive with FER1L6.

Suitable for use as control antibody for FER1L5 siRNA (h): sc-94386, FER1L5 shRNA Plasmid (h): sc-94386-SH and FER1L5 shRNA (h) Lentiviral Particles: sc-94386-V.

Molecular Weight of FER1L5 isoform 1: 242 kDa.

Molecular Weight of FER1L5 isoform 2: 93 kDa.

Molecular Weight of FER1L5 isoform 3: 16 kDa.

RECOMMENDED SECONDARY REAGENTS

To ensure optimal results, the following support (secondary) reagents are recommended: 1) Western Blotting: use goat anti-rabbit IgG-HRP: sc-2004 (dilution range: 1:2000-1:100,000) or Cruz Marker™ compatible goat anti-rabbit IgG-HRP: sc-2030 (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 goat anti-rabbit IgG-FITC: sc-2012 (dilution range: 1:100-1:400) or goat anti-rabbit IgG-TR: sc-2780 (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.