SANTA CRUZ BIOTECHNOLOGY, INC.

DFNA5 (A-18): sc-79231

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The Power to Question

BACKGROUND

DFNA5 (deafness, autosomal dominant 5), also known as ICERE-1, is a 496 amino acid protein that is expressed in cochlea tissue, as well as in placenta, brain, heart, liver, lung and pancreas as 2 alternatively spliced isoforms, designated short and long. Defects in the gene encoding DFNA5 are the cause of non-syndromic sensorineural deafness autosomal dominant type 5 (DFNA5), a form of sensorineural hearing loss that results from damage to one of various structures that receive sound information in the brain. The gene encoding DFNA5 maps to human chromosome 7, which houses over 1,000 genes and comprises nearly 5% of the human genome. Defects in some of the genes localized to chromosome 7 have been linked to osteogenesis imperfecta, Williams-Beuren syndrome, Pendred syndrome, lissencephaly, citrullinemia and Shwachman-Diamond syndrome.

REFERENCES

1. Van Laer, L., et al. 1997. Refined mapping of a gene for autosomal dominant progressive sensorineural hearing loss (DFNA5) to a 2-cM region, and exclusion of a candidate gene that is expressed in the cochlea. Eur. J. Hum. Genet. 5: 397-405.
2. Van Laer, L., et al. 1998. Nonsyndromic hearing impairment is associated with a mutation in DFNA5. Nat. Genet. 20: 194-197.
3. Van Laer, L., et al. 2002. Is DFNA5 a susceptibility gene for age-related hearing impairment? Eur. J. Hum. Genet. 10: 883-886.
4. Gregan, J., et al. 2003. A yeast model for the study of human DFNA5, a gene mutated in nonsyndromic hearing impairment. Biochim. Biophys. Acta 1638: 179-186.
5. Masuda, Y., et al. 2006. The potential role of DFNA5, a hearing impairment gene, in p53-mediated cellular response to DNA damage. J. Hum. Genet. 51: 652-664.
6. Van Laer, L., et al. 2007. A novel DFNA5 mutation does not cause hearing loss in an Iranian family. J. Hum. Genet. 52: 549-552.
7. Kim, M.S., et al. 2008. Methylation of the DFNA5 increases risk of lymph node metastasis in human breast cancer. Biochem. Biophys. Res. Commun. 370: 38-43.
8. Kim, M.S., et al. 2008. Aberrant promoter methylation and tumor suppressive activity of the DFNA5 gene in colorectal carcinoma. Oncogene 27: 3624-3634.
9. Online Mendelian Inheritance in Man, OMIM™. 2009. Johns Hopkins University, Baltimore, MD. MIM Number: 608798. World Wide Web URL: http://www.ncbi.nlm.nih.gov/omim/

CHROMOSOMAL LOCATION

Genetic locus: DFNA5 (human) mapping to 7p15.3.

SOURCE

DFNA5 (A-18) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of DFNA5 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-79231 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

APPLICATIONS

DFNA5 (A-18) is recommended for detection of DFNA5 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).

Suitable for use as control antibody for DFNA5 siRNA (h): sc-77135, DFNA5 shRNA Plasmid (h): sc-77135-SH and DFNA5 shRNA (h) Lentiviral Particles: sc-77135-V.

Molecular Weight of DFNA5 long: 55 kDa.

Molecular Weight of DFNA5 short: 11 kDa.

Positive Controls: Hep G2 cell lysate: sc-2227, human kidney extract: sc-363764 or HeLa whole cell lysate: sc-2200.

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 or our catalog for detailed protocols and support products.