SANTA CRUZ BIOTECHNOLOGY, INC.

DFNA5 (L-24): sc-133509

SANTA CRUZ BIOTECHNOLOGY

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

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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.

CHROMOSOMAL LOCATION

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

SOURCE

DFNA5 (L-24) is a Protein A purified rabbit polyclonal antibody raised against synthetic DFNA5 peptide of human origin.

PRODUCT

Each vial contains 100 µg IgG in 1.0 ml PBS with < 0.1% sodium azide, 0.1% gelatin and < 0.02% sucrose.

APPLICATIONS

DFNA5 (L-24) is recommended for detection of DFNA5 of human 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), immunohistochemistry (including paraffin-embedded sections) (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.

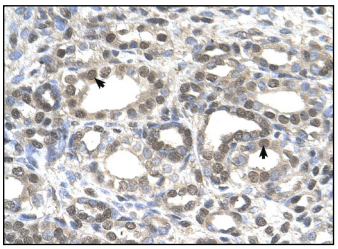
Molecular Weight of DFNA5 short: 11 kDa.

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

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) Immunoprecipitation: use Protein A/G PLUS-Agarose: sc-2003 (0.5 ml agarose/2.0 ml). 3) 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. 4) Immunohistochemistry: use ImmunoCruz™: sc-2051 or ABC: sc-2018 rabbit IgG Staining Systems.

DATA



DFNA5 (L-24): sc-133509. Immunoperoxidase staining of formalin-fixed, paraffin-embedded human kidney tissue showing nuclear and cytoplasmic localization.

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.