



DFNA5 siRNA (h): sc-77135

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 two 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. Grogan, 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.

CHROMOSOMAL LOCATION

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

PRODUCT

DFNA5 siRNA (h) is a pool of 3 target-specific 19-25 nt siRNAs designed to knock down gene expression. Each vial contains 3.3 nmol of lyophilized siRNA, sufficient for a 10 μ M solution once resuspended using protocol below. Suitable for 50-100 transfections. Also see DFNA5 shRNA Plasmid (h): sc-77135-SH and DFNA5 shRNA (h) Lentiviral Particles: sc-77135-V as alternate gene silencing products.

For independent verification of DFNA5 (h) gene silencing results, we also provide the individual siRNA duplex components. Each is available as 3.3 nmol of lyophilized siRNA. These include: sc-77135A, sc-77135B and sc-77135C.

STORAGE AND RESUSPENSION

Store lyophilized siRNA duplex at -20° C with desiccant. Stable for at least one year from the date of shipment. Once resuspended, store at -20° C, avoid contact with RNases and repeated freeze thaw cycles.

Resuspend lyophilized siRNA duplex in 330 μ l of the RNase-free water provided. Resuspension of the siRNA duplex in 330 μ l of RNase-free water makes a 10 μ M solution in a 10 μ M Tris-HCl, pH 8.0, 20 mM NaCl, 1 mM EDTA buffered solution.

APPLICATIONS

DFNA5 siRNA (h) is recommended for the inhibition of DFNA5 expression in human cells.

SUPPORT REAGENTS

For optimal siRNA transfection efficiency, Santa Cruz Biotechnology's siRNA Transfection Reagent: sc-29528 (0.3 ml), siRNA Transfection Medium: sc-36868 (20 ml) and siRNA Dilution Buffer: sc-29527 (1.5 ml) are recommended. Control siRNAs or Fluorescein Conjugated Control siRNAs are available as 10 μ M in 66 μ l. Each contain a scrambled sequence that will not lead to the specific degradation of any known cellular mRNA. Fluorescein Conjugated Control siRNAs include: sc-36869, sc-44239, sc-44240 and sc-44241. Control siRNAs include: sc-37007, sc-44230, sc-44231, sc-44232, sc-44233, sc-44234, sc-44235, sc-44236, sc-44237 and sc-44238.

GENE EXPRESSION MONITORING

DFNA5 (G-9): sc-393162 is recommended as a control antibody for monitoring of DFNA5 gene expression knockdown by Western Blotting (starting dilution 1:200, dilution range 1:100-1:1000) or immunofluorescence (starting dilution 1:50, dilution range 1:50-1:500).

To ensure optimal results, the following support reagents are recommended: 1) Western Blotting: use m-IgG κ BP-HRP: sc-516102 or m-IgG κ BP-HRP (Cruz Marker): sc-516102-CM (dilution range: 1:1000-1:10000), Cruz Marker™ Molecular Weight Standards: sc-2035, UltraCruz® Blocking Reagent: sc-516214 and Western Blotting Luminol Reagent: sc-2048. 2) Immunofluorescence: use m-IgG κ BP-FITC: sc-516140 or m-IgG κ BP-PE: sc-516141 (dilution range: 1:50-1:200) with UltraCruz® Mounting Medium: sc-24941 or UltraCruz® Hard-set Mounting Medium: sc-359850.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor DFNA5 gene expression knockdown using RT-PCR Primer: DFNA5 (h)-PR: sc-77135-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.

SELECT PRODUCT CITATIONS

1. Xu, W., et al. 2020. Apaf-1 pyroptosome senses mitochondrial permeability transition. *Cell Metab.* E-published.

RESEARCH USE

For research use only, not for use in diagnostic procedures.