

DFNB59 siRNA (h): sc-106723

BACKGROUND

DFNB59 (deafness, autosomal recessive 59), also known as Pejvakin or PJKV, is a 352 amino acid belonging to the gasdermin family, which is a family exclusive to vertebrates. Encoded by a gene that maps to human chromosome 2q31.2, DFNB59 is essential for the proper function of auditory pathway neurons and outer hair cell function. DFNB59 defects may cause non-syndromic sensorineural deafness autosomal recessive type 59, a form of sensorineural hearing impairment characterized by absent or severely abnormal auditory brainstem response but normal otoacoustic emissions (auditory neuropathy or auditory dyssynchrony). DFNB contains a nuclear localization signal, a zinc-binding motif and consists of seven exons spanning 9.8 kb of genomic sequence. DFNB59 shares significant similarity with DFNA5, indicating that these genes share a common origin.

REFERENCES

1. Delmaghani, S., et al. 2006. Mutations in the gene encoding pejvakin, a newly identified protein of the afferent auditory pathway, cause DFNB59 auditory neuropathy. *Nat. Genet.* 38: 770-778.
2. Hashemzadeh Chaleshtori, M., et al. 2007. Novel mutations in the pejvakin gene are associated with autosomal recessive non-syndromic hearing loss in Iranian families. *Clin. Genet.* 72: 261-263.
3. Tamura, M., et al. 2007. Members of a novel gene family, Gsdm, are expressed exclusively in the epithelium of the skin and gastrointestinal tract in a highly tissue-specific manner. *Genomics* 89: 618-629.
4. Ebermann, I., et al. 2007. Truncating mutation of the DFNB59 gene causes cochlear hearing impairment and central vestibular dysfunction. *Hum. Mutat.* 28: 571-577.
5. Collin, R.W., et al. 2007. Involvement of DFNB59 mutations in autosomal recessive nonsyndromic hearing impairment. *Hum. Mutat.* 28: 718-723.
6. Schwander, M., et al. 2007. A forward genetics screen in mice identifies recessive deafness traits and reveals that pejvakin is essential for outer hair cell function. *J. Neurosci.* 27: 2163-2175.
7. Xu, S., et al. 2008. Sequence analysis of DFNB59 gene in a Chinese family with dominantly inherited auditory neuropathy. *Lin Chung Er Bi Yan Hou Tou Jing Wai Ke Za Zhi* 22: 880-882.
8. Mahdiah, N., et al. 2010. Genetic causes of nonsyndromic hearing loss in Iran in comparison with other populations. *J. Hum. Genet.* 55: 639-648.

CHROMOSOMAL LOCATION

Genetic locus: DFNB59 (human) mapping to 2q31.2.

RESEARCH USE

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

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.

PRODUCT

DFNB59 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 DFNB59 shRNA Plasmid (h): sc-106723-SH and DFNB59 shRNA (h) Lentiviral Particles: sc-106723-V as alternate gene silencing products.

For independent verification of DFNB59 (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-106723A, sc-106723B and sc-106723C.

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

DFNB59 siRNA (h) is recommended for the inhibition of DFNB59 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.

RT-PCR REAGENTS

Semi-quantitative RT-PCR may be performed to monitor DFNB59 gene expression knockdown using RT-PCR Primer: DFNB59 (h)-PR: sc-106723-PR (20 μ l). Annealing temperature for the primers should be 55-60° C and the extension temperature should be 68-72° C.