

# DFNB59 (Y-15): sc-167621

## BACKGROUND

DFNB59 (deafness, autosomal recessive 59), also known as Pejvakin or PJVK, 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 dys-synchrony). DFNB contains a nuclear localization signal, a zinc-binding motif and consists of 7 exons spanning 9.8 kb of genomic sequence. DFNB59 shares significant similarity with DFNA5, indicating that these genes share a common origin.

## REFERENCES

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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. Mahdieh, N., Rabbani, B., Wiley, S., Akbari, M.T. and Zeinali, S. 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; Dfnb59 (mouse) mapping to 2 C3.

## SOURCE

DFNB59 (Y-15) is an affinity purified goat polyclonal antibody raised against a peptide mapping within an internal region of DFNB59 of human origin.

## RESEARCH USE

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

## 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-167621 P, (100 µg peptide in 0.5 ml PBS containing < 0.1% sodium azide and 0.2% BSA).

## APPLICATIONS

DFNB59 (Y-15) is recommended for detection of DFNB59 of mouse, rat and 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).

DFNB59 (Y-15) is also recommended for detection of DFNB59 in additional species, including equine, bovine, porcine and canine.

Suitable for use as control antibody for DFNB59 siRNA (h): sc-106723, DFNB59 siRNA (m): sc-143018, DFNB59 shRNA Plasmid (h): sc-106723-SH, DFNB59 shRNA Plasmid (m): sc-143018-SH, DFNB59 shRNA (h) Lentiviral Particles: sc-106723-V and DFNB59 shRNA (m) Lentiviral Particles: sc-143018-V.

Molecular Weight of DFNB59: 40 kDa.

## 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.

## PROTOCOLS

See our web site at [www.scbt.com](http://www.scbt.com) or our catalog for detailed protocols and support products.