Resource Summary Report

Generated by SPARC SAWG on Aug 9, 2026

Connexin-deafness

RRID:SCR_006531
Type: Tool

Proper Citation

Connexin-deafness (RRID:SCR_006531)

Resource Information

URL: http://davinci.crg.es/deafness/

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Description: Database and data set of known mutations in connexins related to deafness with associated information including published work and classification scheme. Users may submit new mutations. A large number of subjects are affected by hearing impairment. In developed countries deafness has an important genetic origin and at least 60% of the cases are inherited. The pattern of inheritance can be dominant, recessive, X-linked and mitochondrial. Many genes are involved in the different types of deafness (syndromic and non-syndromic). Non-syndromic hereditary deafness is mainly (80%) due to recessive genes (or mutations). It is believed that more than one hundred genes could be involved in hearing impairment. Several of these genes have been identified recently by positional cloning or positional candidate gene approaches. Despite the fact that more than 20 loci have been described for non-syndromic autosomal recessive deafness (DFNB), a single locus, DFNB1, accounts for a high proportion of the cases, with variability depending on the population. The gene involved in this type of deafness is GJB2, which encodes the gap junction protein connexin 26(Cx26). NEW Recent data indicates that DFNB1 can also be due to a deletion of 342Kb involving GJB6, a gene that is very close to GJB2. This deletion has been reported to cause deafness both in the homozygous status and in heterozygosity with a GJB2 point mutation in trans (see big deletions affecting connexin genes...). Connexins are transmembrane proteins that form channels allowing rapid transport of ions or small molecules between cells. There are two types of connexins, alpha and beta, named GJA or GJB followed by a number. Connexins are expressed in many different tissues. Other connexin genes are also involved in deafness. These are GJB1 (Cx32), which is also responsible for X-linked Charcot-Marie-Tooth disease type I; GJB3 (Cx31), involved in both deafness or a skin disease, erythrokeratodermia variabilis, depending on the location of the mutation; GJB6 (Cx30), which has been related to a dominant type of deafness in an Italian family and NEW GJA1 (Cx43), which has recently been shown to be involved in recessive deafness.

Abbreviations: Connexin-deafness

Synonyms: The Connexins-deafness, Connexins and deafness, Connexins-deafness, Connexins-deafness homepage, Connexin-deafness homepage, Connexins and deafness Homepage

Resource Type: data or information resource, data set, database, service resource, storage service resource, data repository

Keywords: connexin, deletion, insertion, gjb1, gjb2, gjb3, gjb6, mutation, nonsyndromic deafness, polymorphism, syndromic deafness, hereditary deafness, gene, dominant, recessive, hearing, FASEB list

Related Condition: Deafness, Hearing impairment

Availability: Acknowledgement requested

Resource Name: Connexin-deafness

Resource ID: SCR_006531

Alternate IDs: nif-0000-10449

Record Creation Time: 20220129T080236+0000

Record Last Update: 20260808T185839+0000

Ratings and Alerts

No rating or validation information has been found for Connexin-deafness.

No alerts have been found for Connexin-deafness.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 46 mentions in open access literature.

Listed below are recent publications. The full list is available at SPARC SAWG .

Zhao LJ, et al. (2022) Novel m.4268T>C mutation in the mitochondrial tRNAIle gene is associated with hearing loss in two Chinese families. World journal of clinical cases, 10(1), 205.

Fukunaga I, et al. (2021) Generation of two iPSC lines from siblings of a homozygous patient with hearing loss and a heterozygous carrier with normal hearing carrying p.G45E/Y136X mutation in GJB2. Stem cell research, 53, 102290.

Fukunaga I, et al. (2020) Generation of two induced pluripotent stem cell lines from PBMCs of siblings carrying c.235delC mutation in the GJB2 gene associated with

sensorineural hearing loss. Stem cell research, 47, 101910.

Usami SI, et al. (2020) Cochlear Implantation From the Perspective of Genetic Background. Anatomical record (Hoboken, N.J. : 2007), 303(3), 563.

Fukunaga I, et al. (2020) Generation of the induced pluripotent stem cell (hiPSC) line (JUFMDOi004-A) from a patient with hearing loss carrying GJB2 (p.V37I) mutation. Stem cell research, 43, 101674.

Sindura KP, et al. (2019) An Immunological Perspective to Non-syndromic Sensorineural Hearing Loss. Frontiers in immunology, 10, 2848.

Wu X, et al. (2019) Structure and Function of Cochlear Gap Junctions and Implications for the Translation of Cochlear Gene Therapies. Frontiers in cellular neuroscience, 13, 529.

Amritkumar P, et al. (2018) Role of DFNB1 mutations in hereditary hearing loss among assortative mating hearing impaired families from South India. BMC medical genetics, 19(1), 105.

Batissoco AC, et al. (2018) A Cell Junctional Protein Network Associated with Connexin-26. International journal of molecular sciences, 19(9).

Naseri M, et al. (2018) Genetic Linkage Analysis of DFNB4, DFNB28, DFNB93 Loci in Autosomal Recessive Non-syndromic Hearing Loss: Evidence for Digenic Inheritance in GJB2 and GJB3 Mutations. Iranian journal of public health, 47(1), 95.

Del Castillo FJ, et al. (2017) DFNB1 Non-syndromic Hearing Impairment: Diversity of Mutations and Associated Phenotypes. Frontiers in molecular neuroscience, 10, 428.

Pandya A, et al. (2016) Genetic hearing loss: the journey of discovery to destination - how close are we to therapy? Molecular genetics & genomic medicine, 4(6), 583.

Shi X, et al. (2016) Polymorphism of the 86th amino acid in CX26 protein and hereditary deafness. Journal of otology, 11(2), 84.

Maxwell KN, et al. (2016) Evaluation of ACMG-Guideline-Based Variant Classification of Cancer Susceptibility and Non-Cancer-Associated Genes in Families Affected by Breast Cancer. American journal of human genetics, 98(5), 801.

Rodriguez-Paris J, et al. (2016) Comparative functional characterization of novel non-syndromic GJB2 gene variant p.Gly45Arg and lethal syndromic variant p.Gly45Glu. PeerJ, 4, e2494.

Strain GM, et al. (2015) The Genetics of Deafness in Domestic Animals. Frontiers in veterinary science, 2, 29.

Chang Q, et al. (2015) Timed conditional null of connexin26 in mice reveals temporary requirements of connexin26 in key cochlear developmental events before the onset of hearing. Neurobiology of disease, 73, 418.

Taniguchi M, et al. (2015) Carrier frequency of the GJB2 mutations that cause hereditary hearing loss in the Japanese population. Journal of human genetics, 60(10), 613.

Zheng J, et al. (2015) GJB2 Mutation Spectrum and Genotype-Phenotype Correlation in 1067 Han Chinese Subjects with Non-Syndromic Hearing Loss. PloS one, 10(6), e0128691.

Wei Q, et al. (2014) A novel compound heterozygous mutation in the GJB2 gene causing non-syndromic hearing loss in a family. International journal of molecular medicine, 33(2), 310.