GJB2 cDNA Clone – Gap Junction Protein, Beta 2, 26kDa (Human)

PRODUCT OVERVIEW

Product Name	GJB2, cDNA Clone
Full Product Name	GJB2 cDNA Clone
Product Gene Name	GJB2 cdna clone
Sequence	atggattggg gcacgctgca gacgatcctg gggggtgtga acaaacactc caccagcatt ggaaagatct ggctcaccgt cctcttcatt tttcgcatta tgatcctcgt tgtggctgca aaggaggtgt ggggagatga gcaggccgac tttgtctgca acaccctgca gccaggctgc aagaacgtgt gctacgatca ctacttcccc atctcccaca tccggctatg ggccctgcag ctgatcttcg tgtccacgcc agcgctccta gtggccatgc acgtggccta ccggagacat gagaagaaga ggaagttcat caaggggggag ataaagagtg aatttaagga catcgaggag atcaaaaccc agaaggtccg catcgaaggc tccctgtggt ggacctacac aagcagcatc ttcttccggg tcatcttcga agccgccttc atgtacgtct tctatgtcat gtacgacggc ttctccatgc agcggctggt gaagtgcaac gcctggcctt gtcccaacac tgtggactgc tttgtgtccc ggcccacgga gaagactgtc ttcacagtgt tcatgattgc agtgtctgga atttgcatcc tgctgaatgt cactgaattg tgttatttgc taattagata ttgttctggg aagtcaaaaa agccagttta a
Molecular Weight	26,215 Da
OMIM	121011
Vector	pENTR223.1 or pUC
Clone Sequence Report	Provided with product shipment
ISO Certification	Manufactured in an ISO 9001:2015 Certified Laboratory.
Related Product Information for GJB2 cdna clone	MBS1267284 is a cDNA clone targeting GJB2.
Other Notes	Small volumes of GJB2 cdna clone vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply.

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for GJB2. It may not necessarily be applicable to this product.

NCBI GI #	16877591
NCBI GeneID	2706
NCBI Accession #	BC017048
UniProt Secondary Accession #	Q508A5; Q508A6; Q5YLL0; Q5YLL1; Q5YLL4; Q6IPV5; Q86U88; Q96AK0; Q9H536; Q9NNY4
UniProt Related Accession #	P29033
NCBI Official Full Name	Homo sapiens gap junction protein, beta 2, 26kDa, mRNA
NCBI Official Synonym Full Names	gap junction protein beta 2
NCBI Official Symbol	GJB2
NCBI Official Synonym Symbols	HID; KID; PPK; CX26; DFNA3; DFNB1; NSRD1; DFNA3A; DFNB1A
UniProt Protein Name	Gap junction beta-2 protein
UniProt Synonym Protein Names	Connexin-26; Cx26
Protein Family	Gap junction beta-2 protein
UniProt Gene Name	GJB2

UniProt Synonym Gene Names	Cx26
UniProt Entry Name	CXB2_HUMAN
NCBI Protein Information	gap junction beta-2 protein
NCBI Summary for GJB2	This gene encodes a member of the gap junction protein family. The gap junctions were first characterized by electron microscopy as regionally specialized structures on plasma membranes of contacting adherent cells. These structures were shown to consist of cell-to-cell channels that facilitate the transfer of ions and small molecules between cells. The gap junction proteins, also known as connexins, purified from fractions of enriched gap junctions from different tissues differ. According to sequence similarities at the nucleotide and amino acid levels, the gap junction proteins are divided into two categories, alpha and beta. Mutations in this gene are responsible for as much as 50% of pre-lingual, recessive deafness. [provided by RefSeq, Oct 2008]
UniProt Comments for GJB2	GJB2: One gap junction consists of a cluster of closely packed pairs of transmembrane channels, the connexons, through which materials of low MW diffuse from one cell to a neighboring cell. Defects in GJB2 are the cause of deafness autosomal recessive type 1A (DFNB1A). DFNB1A is a form of sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. Defects in GJB2 are the cause of deafness autosomal dominant type 3A (DFNA3A). Defects in GJB2 are a cause of Vohwinkel syndrome (VS). VS is an autosomal dominant disease characterized by hyperkeratosis, constriction on finger and toes and congenital deafness. Defects in GJB2 are a cause of palmoplantar keratoderma with deafness (PPKDFN). PPKDFN is an autosomal dominant disorder characterized by the association of palmoplantar hyperkeratosis with progressive, bilateral, high-frequency, sensorineural deafness. Defects in GJB2 are a cause of keratitis-ichthyosis- deafness syndrome (KID syndrome); an autosomal dominant form of ectodermal dysplasia. Ectodermal dysplasias (EDs) constitute a heterogeneous group of developmental disorders affecting tissues of ectodermal origin. EDs are characterized by abnormal development of two or more ectodermal structures such as hair, teeth, nails and sweat glands, with or without any additional clinical sign. Each combination of clinical features represents a different type of ectodermal dysplasia. KID syndrome is characterized by the association of hyperkeratotic skin lesions with vascularizing keratitis and profound sensorineural hearing loss. Clinical features include deafness, ichthyosis, photobia, absent or decreased eyebrows, sparse or absent scalp hair, decreased sweating and dysplastic finger and toenails. Defects in GJB2 are the cause of Bart-Pumphrey syndrome (BPS). BPS is an autosomal dominant disorder characterized by sensorineural hearing loss, palmoplantar keratoderma, knuckle pads, and leukonychia, It shows considerable phenotypic variability. Defects in GJB2 are the cause of ichthyosis hystrix-like with deafness syndrome (HID syndrome). HID syndrome is an autosomal-dominant inherited keratinizing disorder characterized by sensorineural deafness and spiky hyperkeratosis affecting the entire skin. HID syndrome is considered to differ from the similar KID syndrome in the extent and time of occurrence of skin symptoms and the severity of the associated keratitis. Belongs to the connexin family. Beta-type (group I) subfamily. Protein type: Membrane protein, multi-pass; Membrane protein, integral; Motility/polarity/chemotaxis; Cell adhesion Chromosomal Location of Human Ortholog: 13q11-q12 Cellular Component: ER-Golgi intermediate compartment; plasma membrane Biological Process: gap junction assembly; transport Disease: Deafness, Autosomal Dominant 3a; Deafness, Autosomal Recessive 1a; Deafness, Congenital, With Keratopachydermia And Constrictions Of Fingers And Toes; Deafness, X-linked 2; Ichthyosis, Hystrix-like, With Deafness; Keratitis-ichthyosis-deafness Syndrome, Autosomal Dominant; Keratoderma, Palmoplantar, With Deafness; Knuckle Pads, Leukonychia, And Sensorineural Deafness
Research Articles on GJB2	**1.** The detection rate of the single GJB2 mutation among the 160 SNHL subjects in the present study (6.25%) was higher than 2.58% in normal hearing controls in Korean. The DFNB1 was clearly excluded as a molecular etiology in four (40%) subjects
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