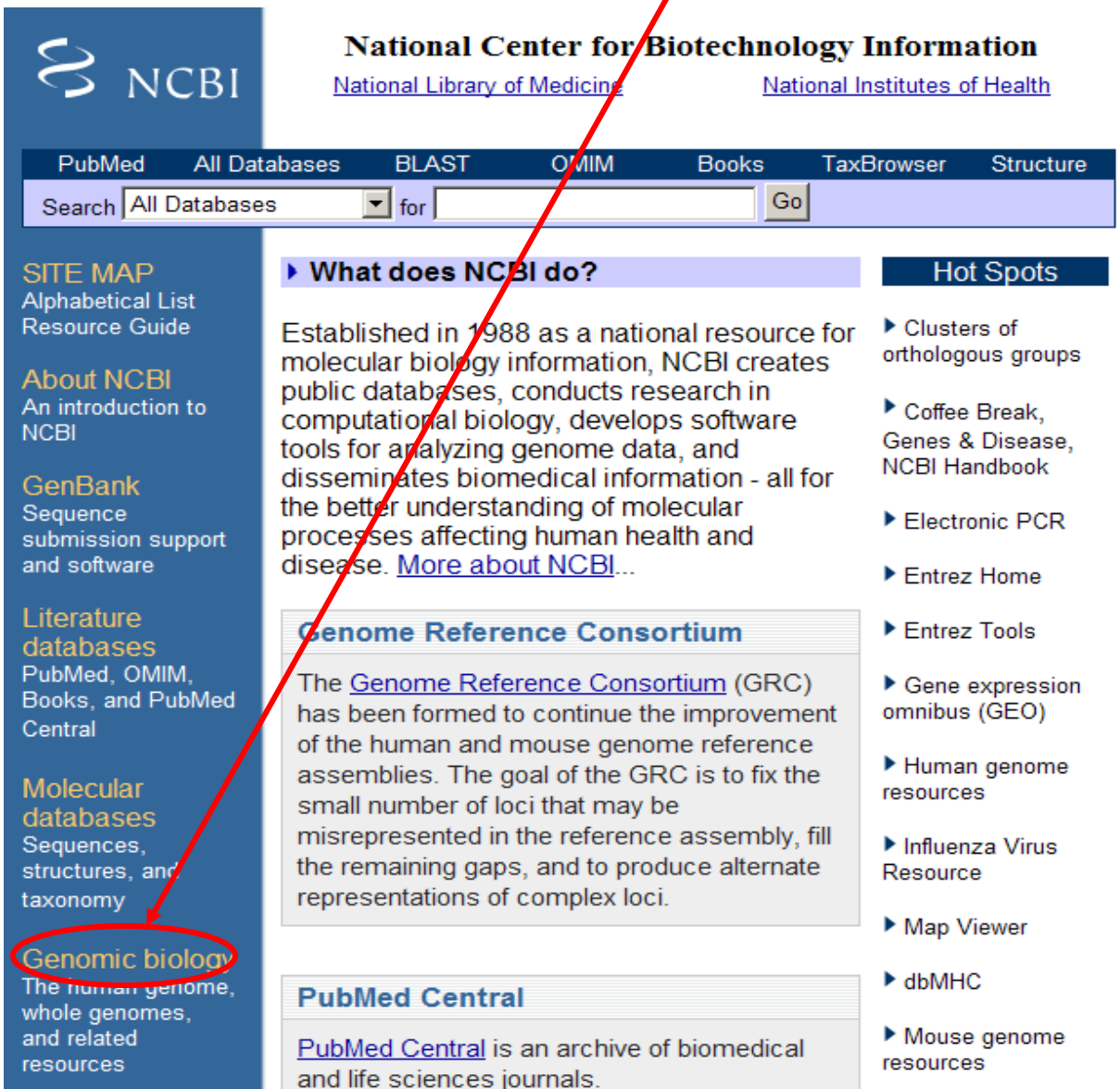
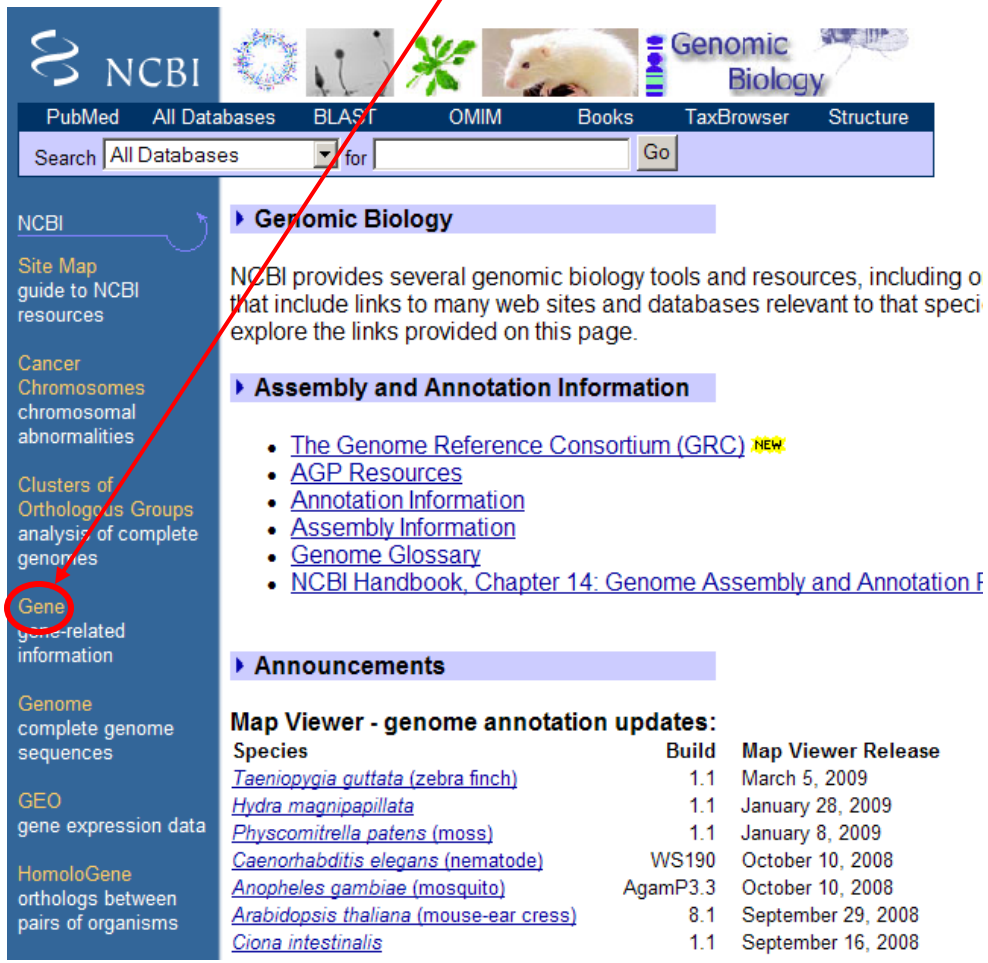
Using Entrez GENE

March 23-24, 2009

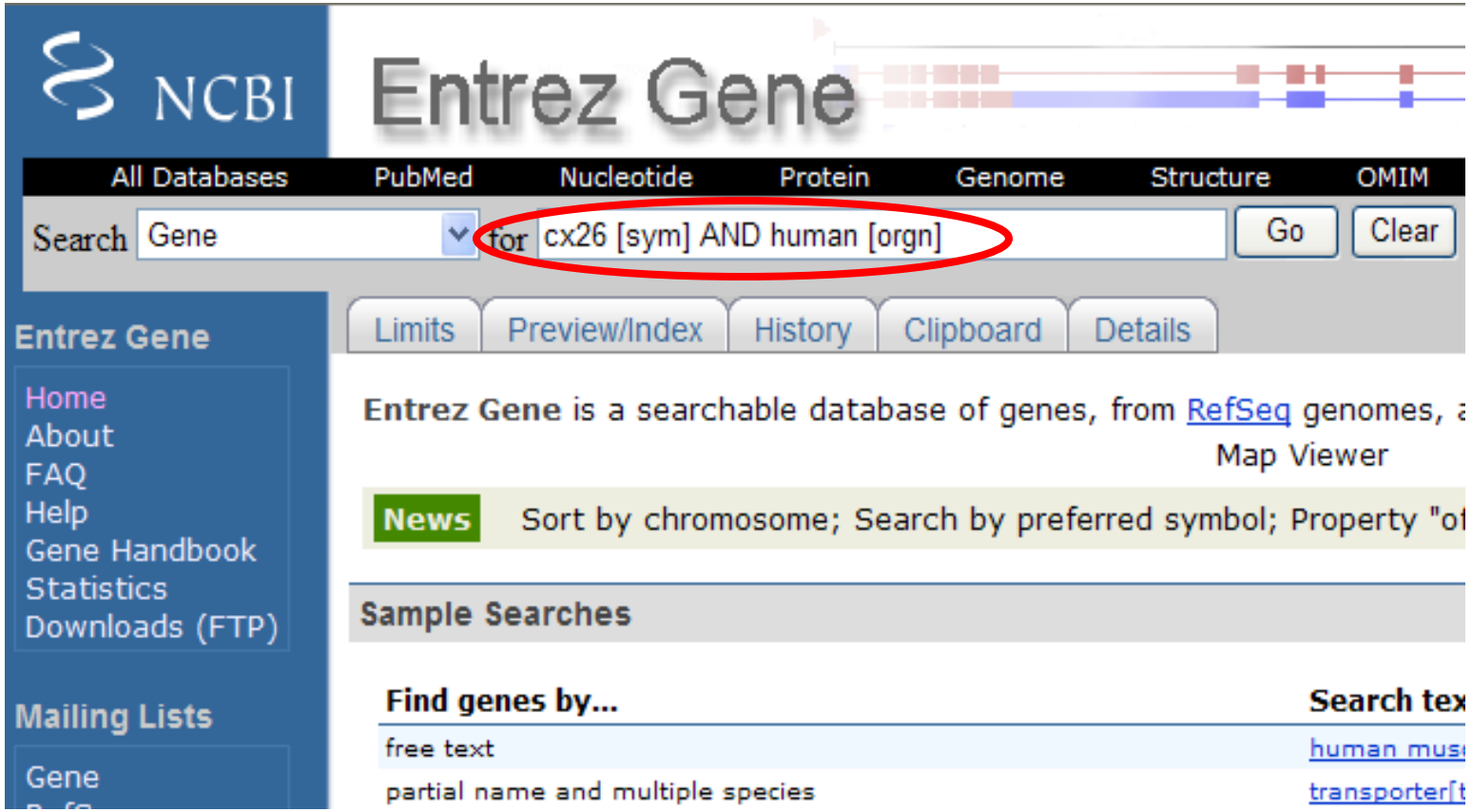
To access Entrez GENE: Click on “Genomic Biology” category



Click on “Gene”

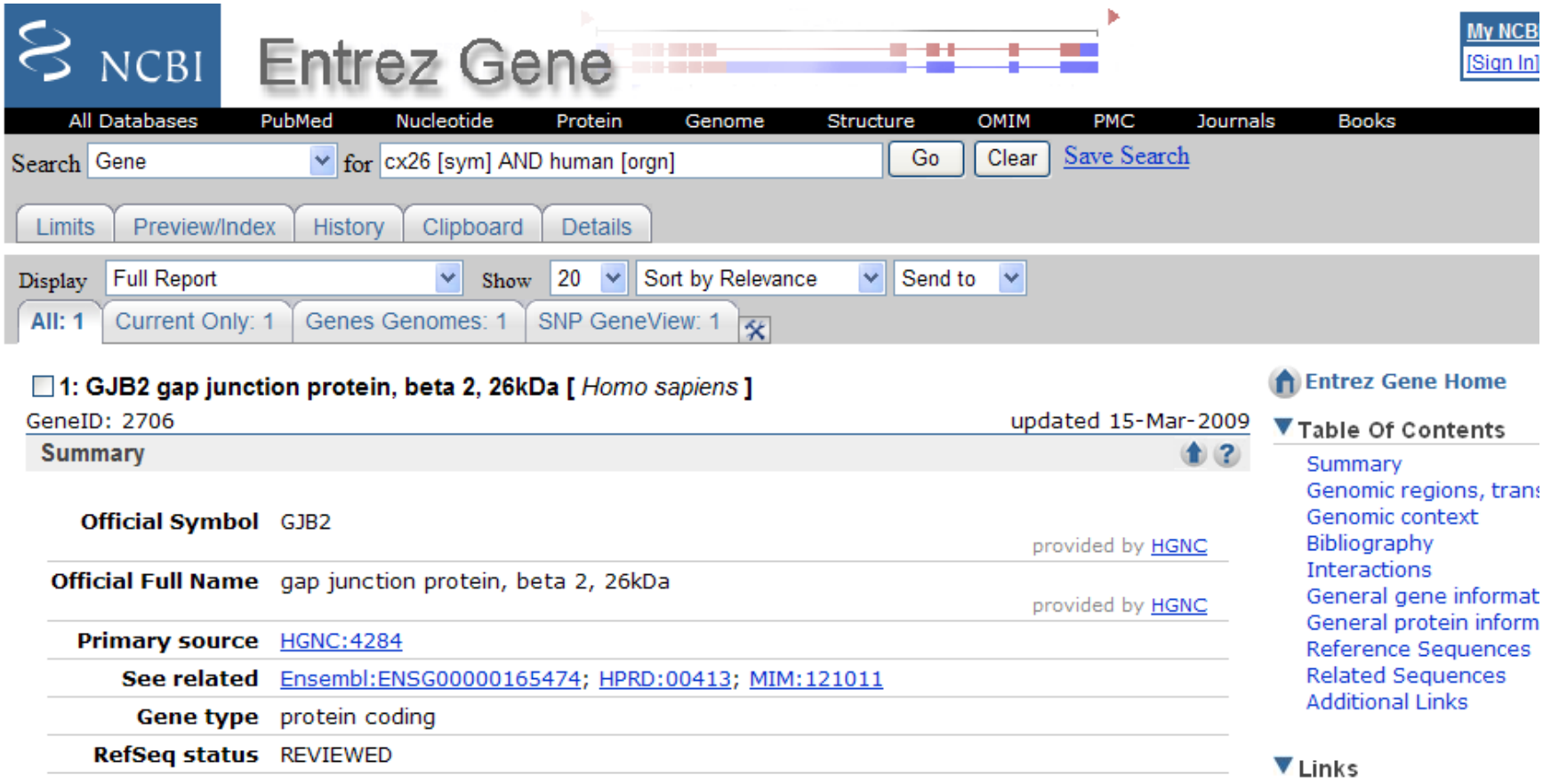


You will use Entrez Gene to answer question #3 on the datasheet. The following screenshots provide clues on where to look for the answers.
You cCan search by name of disease, gene name, gene symbol. Remember to limit your search to human [orgn]



What chromosome is your disease gene found on?
What is the map location for this gene?

Scroll down to the section labeled “Genomic Context.”



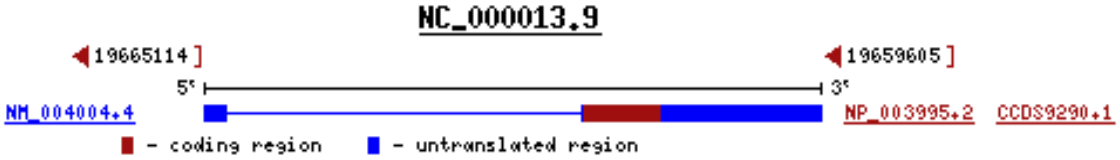
Also known as HID; KID; PPK; CX26; DFNA3; DFNB1; NSRD1; DFNA3A; DFNB1A; GJB2

Summary 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]

Genomic regions, transcripts, and products

(minus strand) Go to reference sequence details

Try our new Sequence Viewer



Genomic context

chromosome: 13; **Location:** 13q11-q12

See GJB2 in MapViewer

How many nucleotides is this gene's coding region?
How many nucleotides are in the gene's transcript?

Click on the NM link. Click on GenBank. This takes you to the mRNA record in Gen Bank.

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]

Genomic regions, transcripts, and products

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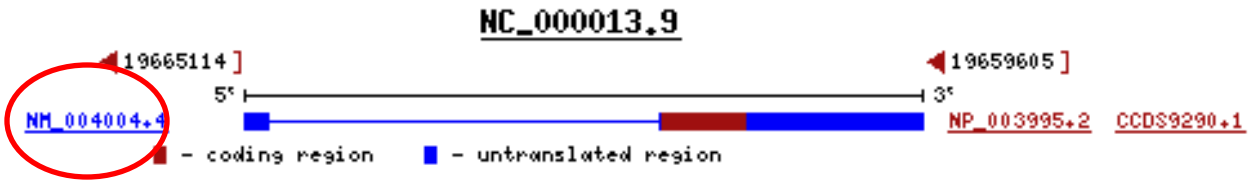
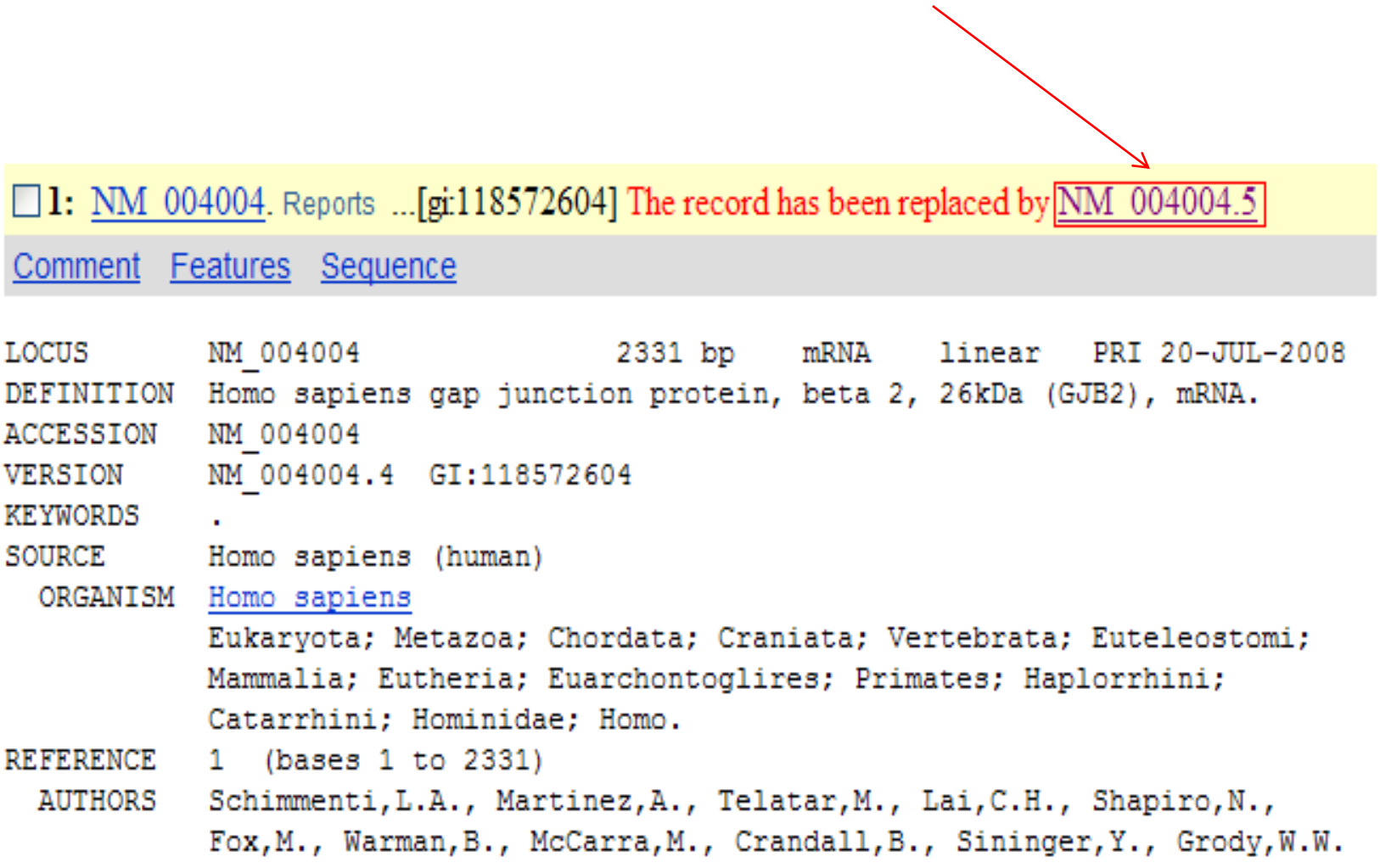


Genomic context

chromosome: 13; **Location:** 13q11-q12

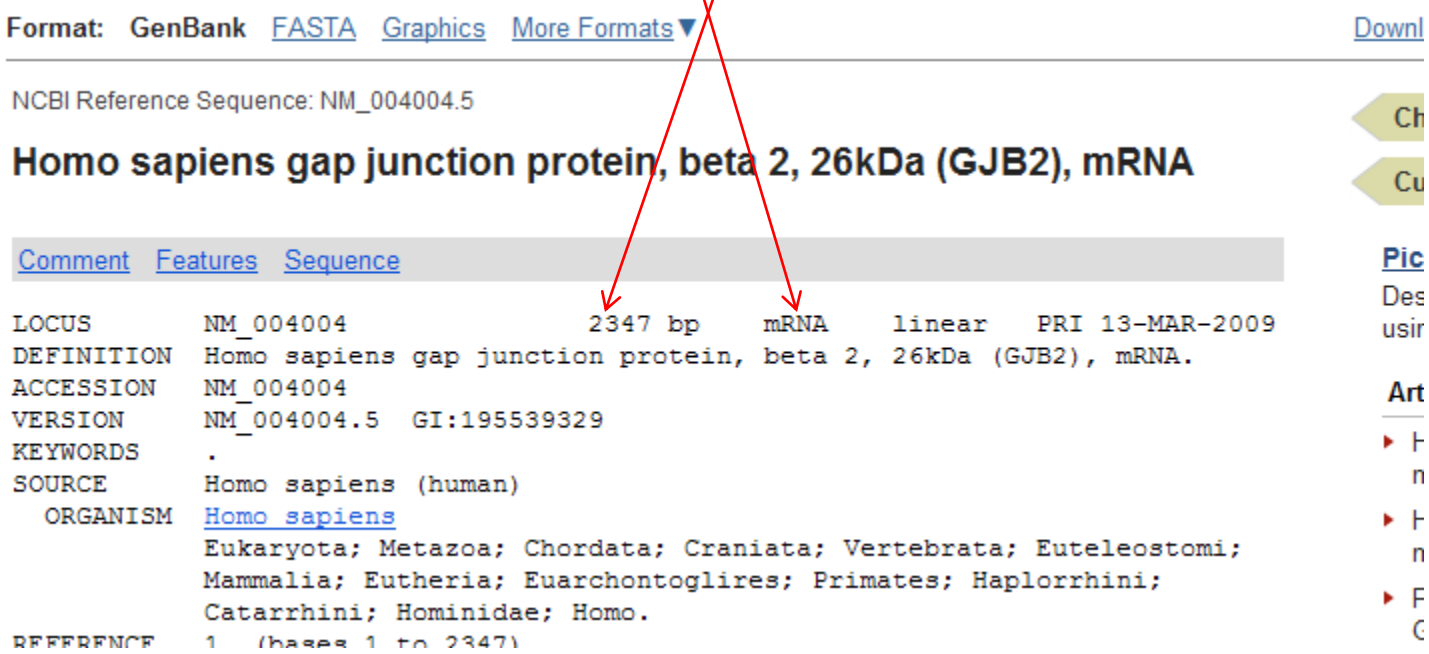
See GJB2 in MapViewer

If your mRNA has a replacement record, click on that link.



This is the GenBank record for the mRNA.

How many nucleotides are in the gene's transcript?



Format: GenBank FASTA Graphics More Formats▼ Downl

NCBI Reference Sequence: NM_004004.5

Homo sapiens gap junction protein, beta 2, 26kDa (GJB2), mRNA

Comment Features Sequence

```
LOCUS       NM_004004               2347 bp    mRNA    linear   PRI 13-MAR-2009
DEFINITION  Homo sapiens gap junction protein, beta 2, 26kDa (GJB2), mRNA.
ACCESSION   NM_004004
VERSION     NM_004004.5  GI:195539329
KEYWORDS    .
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
            Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
            Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
            Catarrhini; Hominidae; Homo.
REFERENCE   1  (bases 1 to 2347)
```

Now, scroll to the “Features” section of the record. Look for the section labeled “CDS”. CDS stands for coding sequence---the region of nucleotides that corresponds with the sequence of amino acids in a protein (location includes start and stop codons).

QUESTION: How many nucleotides is this gene’s coding region? Subtract the smaller number from the larger number, so 896-216 = 680 nucleotides in the coding region.

Use the back arrow to return to the record in EntrezGene

```
exon            194..2334
                /gene="GJB2"
                /gene_synonym="CX26; DFNA3; DFNA3A; DFNB1; DFNB1A; HID;
                KID; NSRD1; PPK"
                /inference="alignment:Splign"
                /number=2
CDS             216..896
                /gene="GJB2"
                /gene_synonym="CX26; DFNA3; DFNA3A; DFNB1; DFNB1A; HID;
                KID; NSRD1; PPK"
                /note="connexin 26; gap junction protein, beta 2, 26kD
                (connexin 26)"
                /codon_start=1
                /product="gap junction protein beta 2"
                /protein_id="NP_003995.2"
                /db_xref="GI:42558283"
                /db_xref="CCDS:CCDS9290.1"
                /db_xref="GeneID:2706"
                /db_xref="HGNC:4284"
                /db_xref="HPRD:00413"
                /db_xref="MIM:121011"
                /translation="MDWGTLQTILGGVNKHSTSIGKIWLTVLFIFRIMILVVAAKEVW
                GDEQADFVCNTLQPGCKNVCYDHYFPISHIRLWALQLIFVSTPALLVAMHVAYRRHEK
                KRKFIKGEIKSEFKDIEEIKTQKVRIEGSLWWTYTSSIFFRVIFEAAFMYVFYVMYDG
                FSMQRLVKCNAWPCPNTVDCFVSRPTEKTVFTVFMIAVSGICILLNVTELCYLLIRYC
                SGKSKKPV"
```

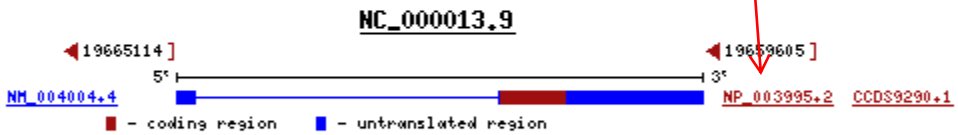
How many amino acids are in the gene's peptide? To find the answer, click on the NP link and choose "GENPEPT"

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]

Genomic regions, transcripts, and products

(minus strand) Go to reference sequence details

Try our new Sequence Viewer

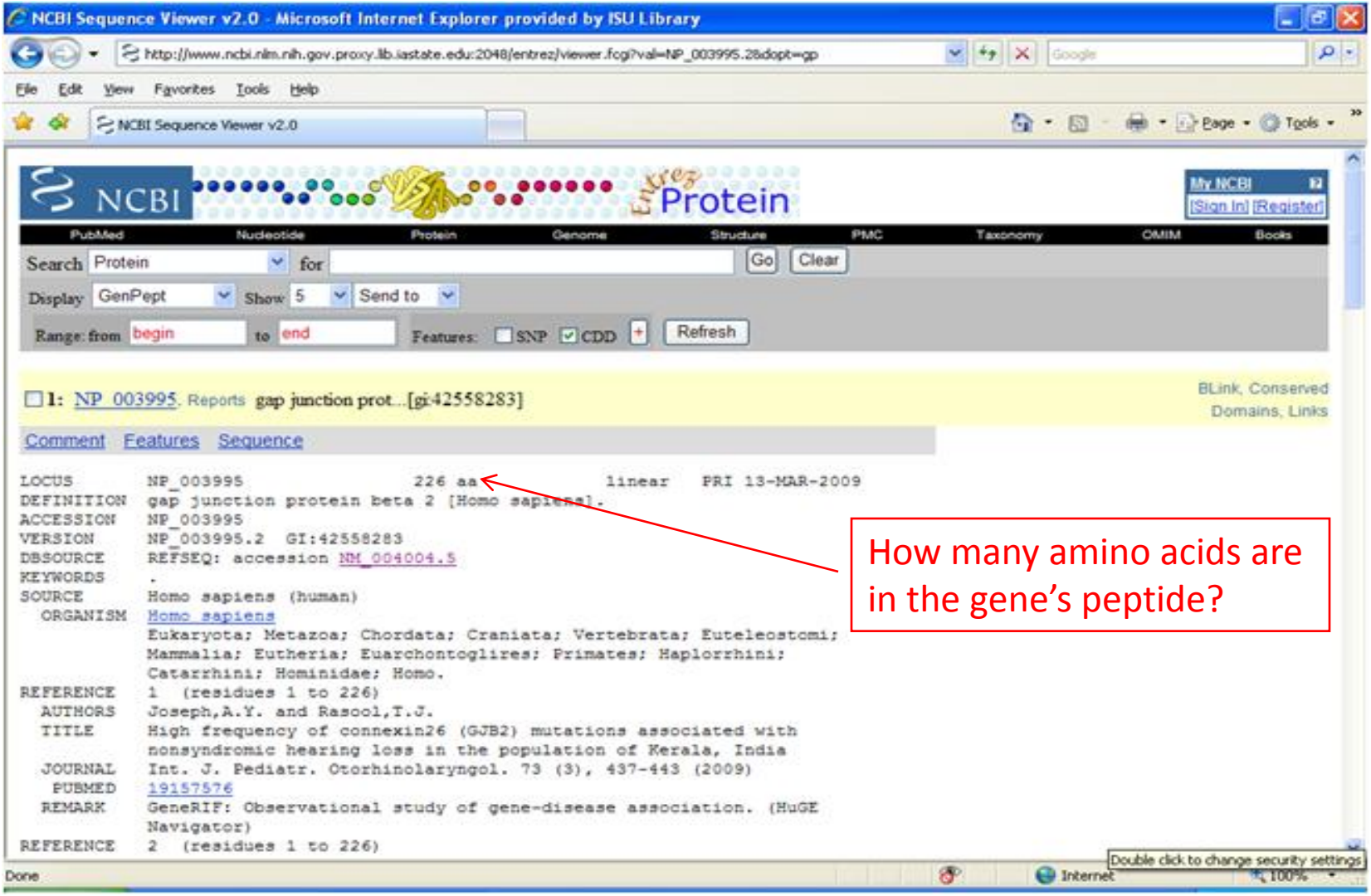


Genomic context

chromosome: 13; **Location:** 13q11-q12

See GJB2 in MapViewer

This will take you the record for the protein in the Entrez Protein database.



- What is the function of this gene's peptide?

There are several places in the Entrez GENE record which provide information about the peptide's function.

Summary

Official Symbol	GJB2 provided by HGNC
Official Full Name	gap junction protein, beta 2, 26kDa provided by HGNC
Primary source	HGNC:4284
See related	Ensembl:ENSG00000165474; HPRD:00413; MIM:121011
Gene type	protein coding
RefSeq status	REVIEWED
Organism	*Homo sapiens*
Lineage	*Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo*
Also known as	HID; KID; PPK; CX26; DFNA3; DFNB1; NSRD1; DFNA3A; DFNB1A; GJB2
Summary	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]

The summary may contain information.

KEGG pathway: Cell Communication
01430
Reactome Event:Gap junction trafficking and regulation
157858

Homology

Mouse, Rat
Map Viewer

More information about function in the GeneOntology section

GeneOntology

Provided by GOA

Function	Evidence
gap junction channel activity	IEA
protein binding	IEA

Refers to molecular function of the gene or gene product.

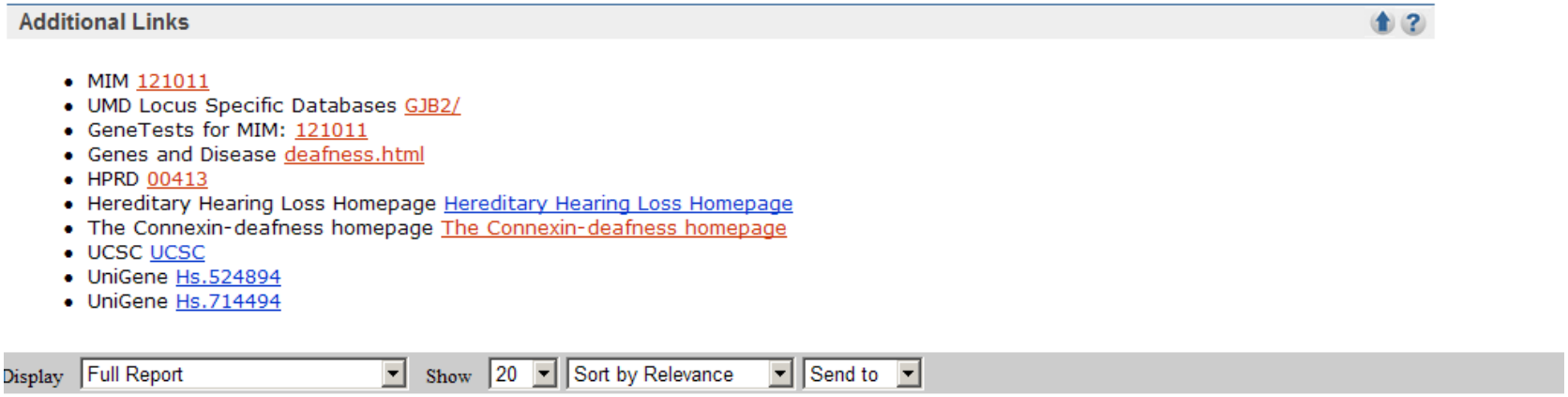
Process	Evidence	
cell-cell signaling	TAS	PubMed
male genitalia development	IEA	
sensory perception of sound	TAS	PubMed
transport	TAS	PubMed

Refers to biological processes that have been linked to the gene or gene product.

Component	Evidence	
Golgi membrane	EXP	PubMed
cell junction	IEA	
connexon complex	TAS	PubMed
integral to membrane	IEA	
lateral plasma membrane	IEA	
plasma membrane	EXP	PubMed
plasma membrane	TAS	PubMed

Refers to the location in the cell in which the gene product acts.

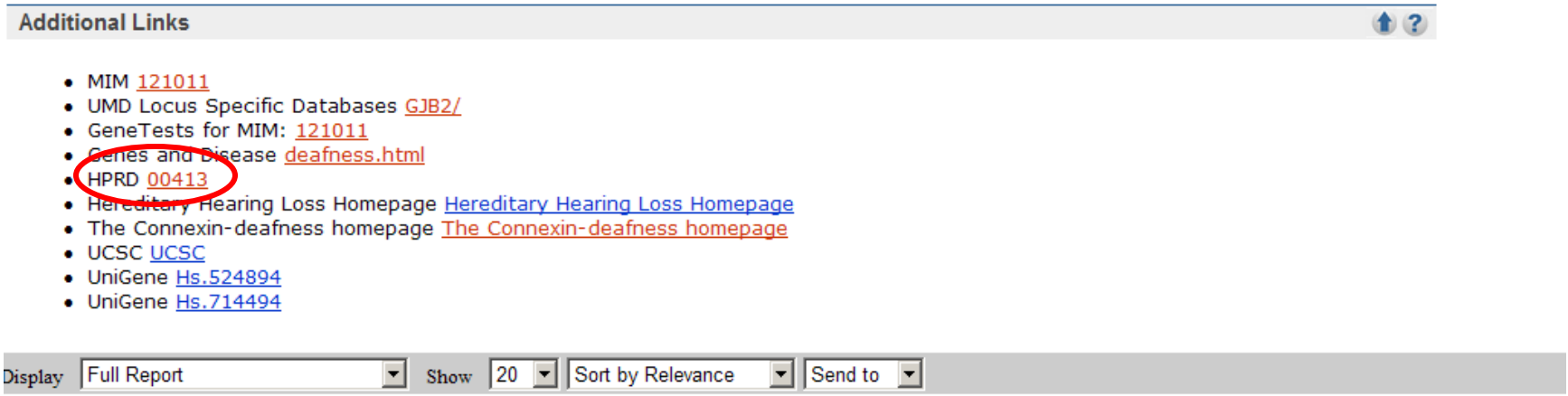
Scroll to the very bottom of the page to the “Additional Links” section. There are links to resources that were discussed earlier: the MIM link goes to OMIM, Genes & Disease book, as well as other resources. Both of these resources may give information about the peptide’s function.



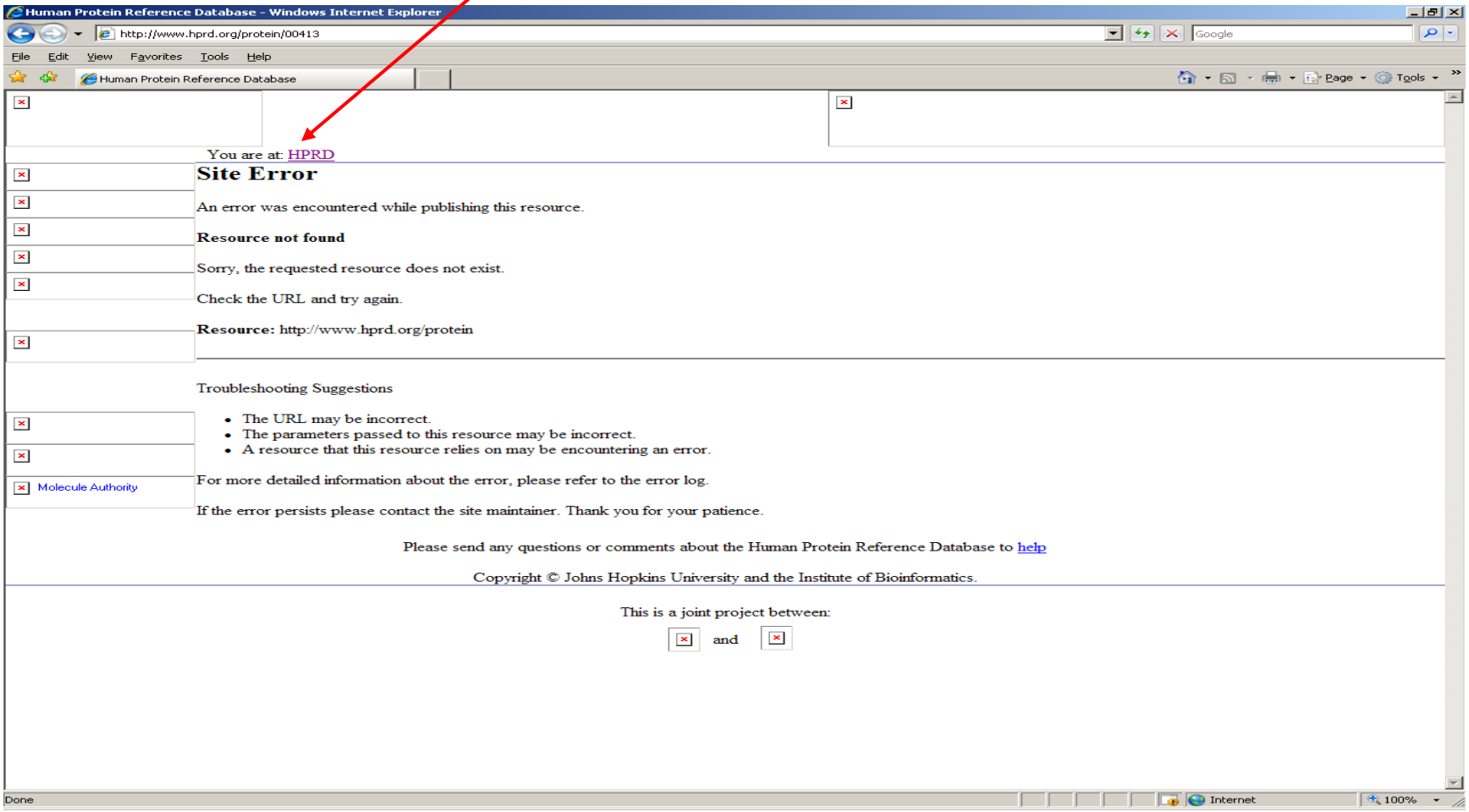
Where in the body is this peptide typically found?

HPRD stands for Human Protein Reference Database. It includes information about where the peptide is expressed in the body.

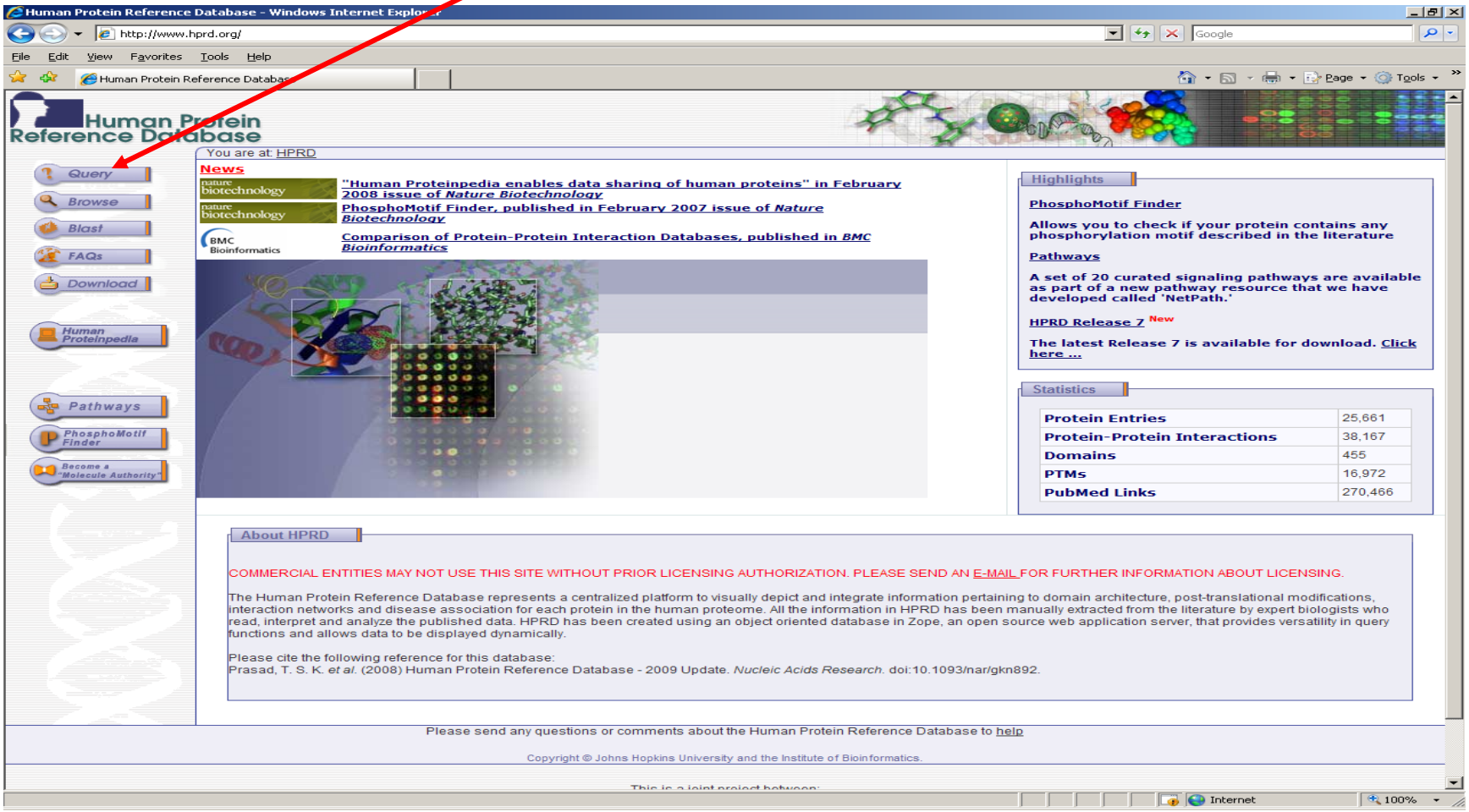
Unfortunately, the links to HPRD are not working quite right. So, click on the HPRD link below. Also, note the record number 00413.



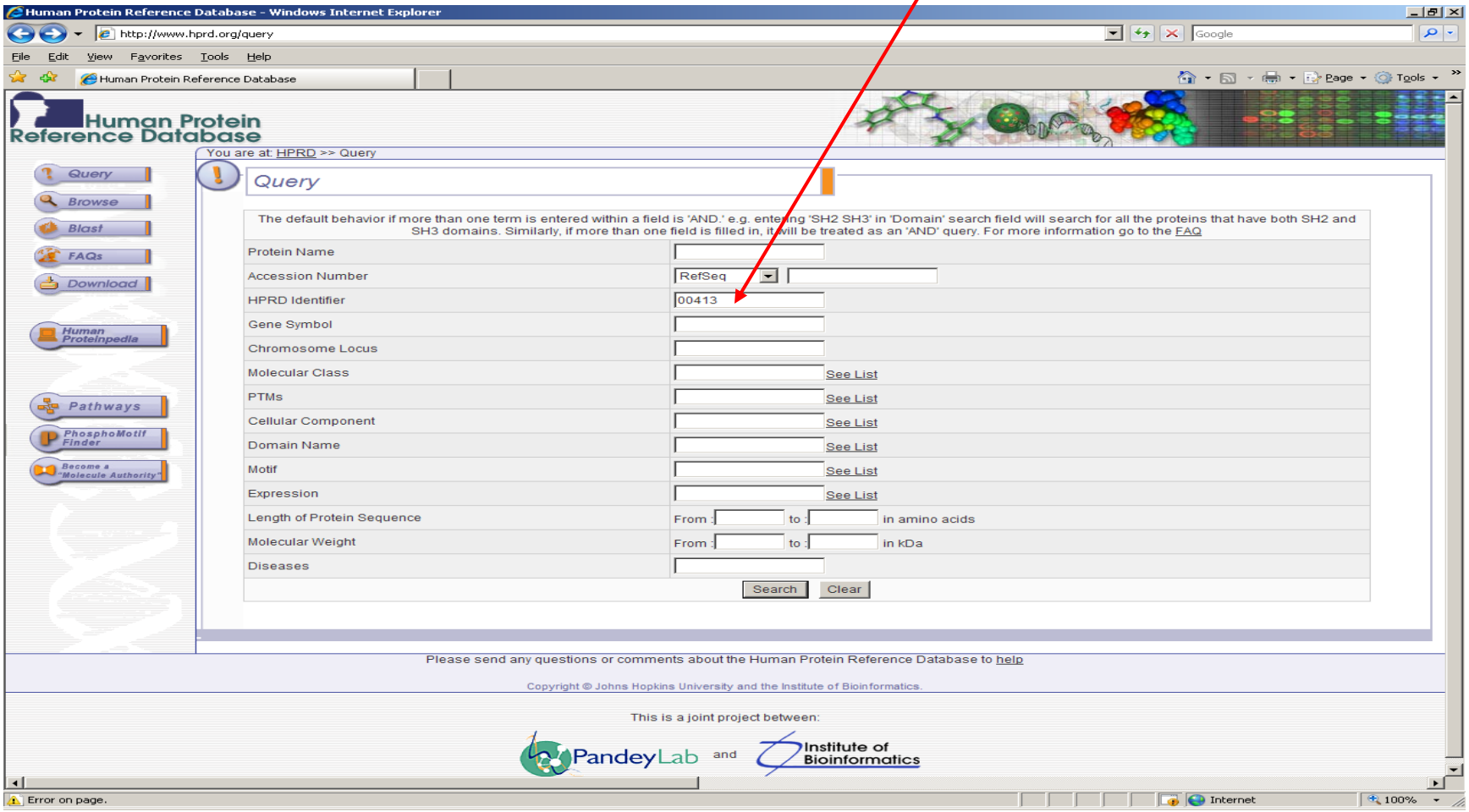
Then, click on HPRD. This will take you to the homepage of the database.



Click on Query.



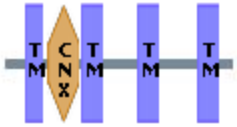
Enter the HPRD Identifier number, 00413, and click the Search button.



Connexin 26

Molecular Class	Membrane transport protein
Molecular Function	Auxiliary transport protein activity
Biological Process	Transport

This protein is expressed in the cochlea, liver, and skin.



ALTERNATE NAMES | DISEASES | PTMs & SUBSTRATES | SUMMARY | SEQUENCE | INTERACTIONS | EXTERNAL LINKS

General

Gene Symbol:	GJB2	Molecular Weight (Da) :	26215	Gene Map Locus:	13q11-q12

Localization

Primary	Plasma membrane GO	Alternate	

Domains and Motifs

Domains		Motifs
TM	132 - 154	
TM	193 - 215	
TM	21 - 40	
CNX	42 - 75	

Expression

Site of Expression
Cochlea
Liver
Skin