Disorder: Hearing loss affects about 1 in 500 newborns and a genetic etiology is suspected in two thirds of these patients. Hearing loss can be caused by mutations in many different genes which can be inherited in an autosomal dominant, autosomal recessive, X-linked or mitochondrial (maternal inheritance) manner.

Mutations in the GJB2 gene, which encodes for the connexin 26 protein, are the most frequent cause of autosomal recessive nonsyndromic hereditary hearing loss, known as DFNB1. Mutations in the GJB2 gene are found in various populations, with carrier rates of approximately 1-in-30 in the United States Caucasian population and 1-in-20 in the Ashkenazi Jewish population.

Indications:
• Sensorineural hearing loss of unknown etiology
• Carrier testing in a relative of a patient with a proven GJB2 mutation

Specimen: At least 2 mLs whole blood in a lavender top (EDTA) tube. Alternately, two cytobrushes (cheek swabs) may be collected. (Please call 1-513-636-4474 for a free cytobrush collection kit). Label each tube or brush with patient’s name, birth date, and date of collection.

Testing Methodology: PCR-based sequencing of the exon/intron boundaries and the entire coding sequence which is contained within exon 2 of the GJB2 gene.

Analysis of GJB2 is also offered as part of our Hearing Loss Panel Tier 1 and OtoSeq® Hearing Loss Panel which includes analyses for additional genes which cause hereditary hearing loss. Please refer to our web site for additional information.

Test Sensitivity: PCR-based sequencing detects 99% of the reported mutations in GJB2. Mutations in GJB2 account for 18% of congenital sensorineural hearing loss in the U.S. population.

The sensitivity of DNA sequencing is over 99% for the detection of nucleotide base changes, small deletions and insertions in the regions analyzed. Mutations in regulatory regions or other untranslated regions are not detected by this test. Large deletions involving entire single exons or multiple exons, large insertions and other complex genetic events are not identified using this test methodology. Rare primer site variants may lead to erroneous results.
Turn-Around Time: 28 days

**Cost:** Please call 1-866-450-4198 for current pricing, insurance precertification, or with any billing questions.

**CPT Codes:**
- **GJB2 full gene sequence analysis**
  - 81252
- **Family specific mutation analysis**
  - 81403

**Results:** Each test report includes a detailed interpretation of the genetic findings, the clinical significance of the result, and specific recommendations for clinical management and additional testing, if warranted. Results will be reported to the referring physician or health care provider as specified on the test requisition form.

**References:**


Additional information and test requisitions are available at: [www.cchmc.org/hearing-loss](http://www.cchmc.org/hearing-loss)

**Shipping Instructions**

Please enclose a completed test requisition, audiogram and MRI/CT report, if available with the sample. All information must be completed before the sample can be processed. Place samples in Styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Friday.

**Ship to:**

Cytogenetics and Molecular Genetics Laboratories
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229
513-636-4474