**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.

Approximately 7-10% of North American patients with a single GJB2 mutation also have large deletion involving GJB6 (digenic inheritance).

**Indications:**
- Sensorineural hearing loss of unknown etiology
- Sensorineural hearing loss and a single mutation in GJB2
- Carrier testing in relative of patient with a proven GJB6 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:** The 342kb deletion in the DFNB1 locus on chromosome 13q12 that affects connexin 30 (GJB6-D13S1830) is analyzed by PCR and gel electrophoresis. Analysis of this large deletion at GJB6 is also offered as part of our Hearing Loss Panel Tier I. Please refer to our web site for additional information.

**Test Sensitivity:** Heterozygous mutations in GJB6, in conjunction with a heterozygous mutation in GJB2 account for 1% of congenital sensorineural hearing loss in the U.S. population.

This test methodology detects the most common deletion involving GJB6. Other deletions and point mutations in GJB6, which are rare, are not detected with this test methodology.
Turn-Around Time: 28 days

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

CPT Codes: 81254

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

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