**Connexin 26 (Non-Syndromic Hereditary Hearing Loss/GJB2/DFNB1)**

**Test Code:** 1150  
**Department:** Molecular Genetics

**Test Synonyms:**  
Connexin  
Non-Syndromic Hereditary Hearing Loss  
Connexin/GJB2/DFNB1

**CPT Code(s):**  
83891  
83898  
83904 x 2

**Background:**  
DFNB1 is an autosomal recessive disorder characterized by congenital non-progressive hearing loss that is moderate to profound. No other systemic findings are associated with DFNB1, thus it is designated a non-syndromic hereditary hearing loss. Approximately 98% of cases of DFNB1 have been shown to be caused by mutations in the *GJB2* gene (connexin 26). *GJB2* encodes the gap junction protein, connexin 26, located on chromosome 13q12.1  
Incidence of DFNB1 is estimated to be 1-2 in 10,000 live births. Mutations in connexin 26 have also been shown to cause the autosomal dominant nonsyndromic hearing loss DFNA3, as well as keratitis-ichthyosis-deafness syndrome, palmoplantar keratoderma with deafness, and hystrix-like ichthyosis-deafness syndrome (Richard, 2003).

**Reason for Referral:**  
Non-syndromic congenital non-progressive hearing loss that is moderate to profound

**Methodology:**  
**Sequencing:** Testing for connexin 26 mutations in DFNB1 is done by sequencing of the entire coding region of the gene (entirely contained in exon 2). The coding region is amplified with a set of primers flanking exon 2 at the 5’ end and approximately 50 bases following the stop codon at the 3’ end (Wu et al., 2003). The PCR product is sequenced with four primers – the PCR primers plus two additional internal sequencing primers.  

Test reporting follows the American College of Medical Genetics (ACMG) guidelines.

**Specimen Requirements:**  
- Blood: EDTA (purple-top tube) or ACD (yellow-top tube)  
  - Adult: 5.0 mL  
  - Child: 5.0 mL  
  - Infant: 2.0-3.0 mL

**A REQUISITION FORM MUST ACCOMPANY ALL SAMPLES.** Please include detailed clinical information, including ethnicity, clinical history, and family history.

**Test Performed (Days):**  
Weekly

**Turn Around Time:**  
7 – 14 Days
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Shipment Sensitivity Requirements:
Package and ship specimen to remain cold, but not frozen. Ship via overnight express, using the FedEx priority overnight label provided. Contact Client Services for shipping kits and instructions at (855) 535-1522.

Additional Information:
Prior to any genetic testing we recommend genetic counseling. To receive forms and information about prenatal diagnostic testing, please contact Client Services at (855) 535-1522.

References: