Nonsyndromic Hearing Loss Secondary to *GJB6* (connexin 30) deletion

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.

Cincinnati Children's

Human Genetics

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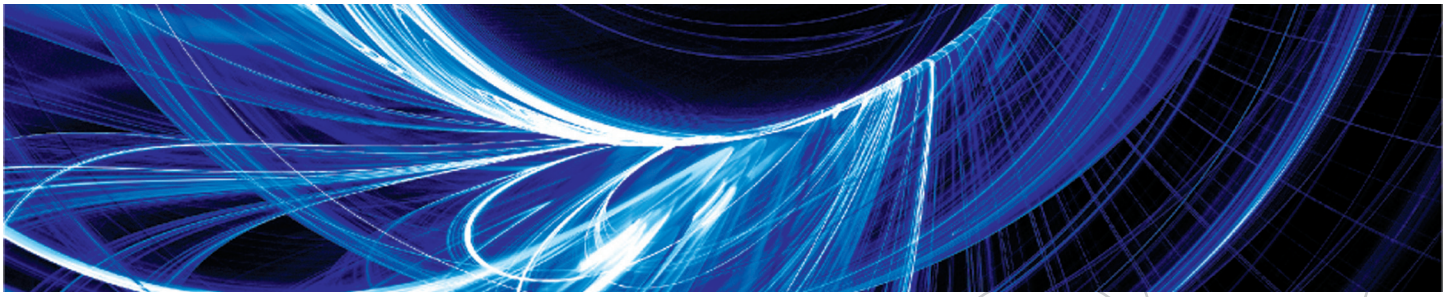

Turn-Around Time: 28 days

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:

del Castillo, F. J., M. Rodriguez-Ballesteros, et al. (2005). "A novel deletion involving the connexin-30 gene, del(GJB6-d13s1854), found in trans with mutations in the GJB2 gene (connexin-26) in subjects with DFNB1 non-syndromic hearing impairment." J Med Genet 42(7): 588-594.

Lim, L. H., J. K. Bradshaw, et al. (2003). "Genotypic and phenotypic correlations of DFNB1-related hearing impairment in the Midwestern United States." Arch Otolaryngol Head Neck Surg 129(8): 836-840.

Lin, J. W., N. Chowdhury, et al. (2011). "Comprehensive diagnostic battery for evaluating sensorineural hearing loss in children." Otol Neurotol 32(2): 259-264.

Marlin, S., D. Feldmann, et al. (2005). "GJB2 and GJB6 mutations: genotypic and phenotypic correlations in a large cohort of hearing-impaired patients." Arch Otolaryngol Head Neck Surg 131(6): 481-487.

Preciado, D. A., L. H. Lim, et al. (2004). "A diagnostic paradigm for childhood idiopathic sensorineural hearing loss." Otolaryngol Head Neck Surg 131(6): 804-809.

Scott, D. A., M. L. Kraft, et al. (1998). "Identification of mutations in the connexin 26 gene that cause autosomal recessive nonsyndromic hearing loss." Hum Mutat 11(5): 387-394.

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:

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