HOTLINE: Effective May 16, 2022

2001956 Hearing Loss, Nonsyndromic, Connexin 30 (*GJB6*) 2 Deletions GJB6 DEL

Specimen Required: Collect: Lavender (EDTA), pink (K_2EDTA), or yellow (ACD Solution A or B).
Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: Refrigerated. Also acceptable: Room temperature
Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: Unacceptable

Interpretive Data:

Background: Hearing Loss, Nonsyndromic, Connexin 30 (*GJB6*) 2 Deletions:
Characteristics: Moderate to profound nonsyndromic hearing loss (NSHL). Large *GJB6* gene deletions involving cis-regulatory elements for GJB2 (connexin 26) result in the loss of expression of GJB2. Thus, compound heterozygosity for a pathogenic *GJB2* variant and *GJB6* large deletion results in NSHL.
Incidence: Approximately 1 in 30 individuals with NSHL has a *GJB6* deletion; 1 in 100,000 in the general population. Twenty percent of *GJB2* heterozygotes with nonsyndromic hearing loss have a *GJB6* deletion; homozygosity for *GJB6* deletions is rare.
Inheritance: Autosomal recessive.
Cause: Pathogenic germline variants in *GJB6*.
Variants Tested: 309kb del(GJB6-D13S1830, also known as 342kb) and 232kb del(GJB6-D13S1854).
Clinical Sensitivity: Dependent on ethnicity.
Methodology: Multiplex PCR using deletion-specific primers, followed by capillary gel electrophoresis.
Analytical Sensitivity and Specificity: Greater than 99 percent.
Limitations: *GJB6* variants other than the two targeted deletions will not be identified. The etiology of hearing loss due to other genetic or environmental causes will not be determined. Diagnostic errors can occur due to rare sequence variations. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.