**ARUP LABORATORIES | aruplab.com**
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Tracy I. George, MD, Chief Medical Officer

Patient Report

Patient Age/Sex: Male

**Specimen Collected: 08-Mar-22 15:53**

**Connexin 30 (GJB6) 2 Deletions** | **Received: 08-Mar-22 15:53** **Report/Verified: 09-Mar-22 09:15**

| Procedure | Result | Units | Reference Interval |
|---|---|---|---|
| GJB6 DEL Specimen Type | Whole Blood | | |
| Connexin 30 GJB6 Deletion 309 | Hetero | | |
| Connexin 30 GJB6 Deletion 232 | Negative | | |
| Connexin 30 Interpretation | See Note [f1] [i1] | | |

## Result Footnote

f1: Connexin 30 Interpretation

Indication for testing: Determine etiology of nonsyndromic hearing loss (NSHL) or assess carrier status for the targeted GJB6 deletions.

Result: One copy of the pathogenic GJB6 309Kb deletion was detected.

Interpretation: One copy of the GJB6 (connexin 30) 309kb deletion (GJB6-D13S1830, also known as 342kb) was detected. This individual is at least a carrier for nonsyndromic hearing loss. If a pathogenic variant is also present in the GJB2 (connexin 26) gene, the combination may result in NSHL.

Recommendations: Medical management should rely on clinical findings and family history. This result should be combined with GJB2 sequencing and deletion/duplication results (ARUP test code 3004720) for optimal interpretation. At-risk family members should be offered testing for the identified deletion (ARUP test code 2001956). This individual's reproductive partner should be offered screening for pathogenic GJB2 and GJB6 variants. Genetic consultation is recommended.

This result has been reviewed and approved by [redacted]

## Test Information

i1: Connexin 30 Interpretation

BACKGROUND INFORMATION: 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.

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

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## Test Information

i1: Connexin 30 Interpretation

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 U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.