ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	11/24/2008
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Hearing Loss, Nonsyndromic, Connexin 30 (GJB6) 2 Deletions

ARUP test code 2001956

GJB6 DEL Specimen Type	Whole Blood
Connexin 30 GJB6 Deletion 309	Negative
Connexin 30 GJB6 Deletion 232	Negative
Connexin 30 Interpretation	See Note

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

Result: The GJB6 309kb and 232kb deletions were not detected.

Interpretation: Neither of the targeted GJB6 (connexin 30) deletions, 309kb (GJB6-D13S1830, also known as 342kb) and 232kb (GJB6-D13S1854), were identified. This result reduces, but does not exclude, a diagnosis of or carrier status for GJB6-related hearing loss. This individual could still be a carrier of or affected with hearing loss due to other genetic or environmental causes. Please refer to the background information included in this report for limitations of this test.

Recommendations: Medical screening and management should rely on clinical findings and family history. If suspicion for an inherited form of hearing loss remains, consider analysis of other associated genes (Expanded Hearing Loss Panel, Sequencing and Deletion/Duplication, ARUP test code 2008803). Genetic consultation is recommended.

This result has been reviewed and approved by [redacted]

H=High, L=Low, *=Abnormal, C=Critical

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.
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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
GJB6 DEL Specimen Type	23-076-401474	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Connexin 30 GJB6 Deletion 309	23-076-401474	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Connexin 30 GJB6 Deletion 232	23-076-401474	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Connexin 30 Interpretation	23-076-401474	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical