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	Unknown
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Usher Syndrome, Types 1F and 3 (PCDH15 and CLRN1), 2 Variants

ARUP test code 2013750

Usher Syndrome Types 1F and 3, Specimen	Whole Blood
Usher Syndrome Types 1F and 3, Allele 1	c.733C>T *
Usher Syndrome Types 1F and 3, Allele 2	Negative
Usher Syndrome Types 1F and 3, Interp	See Note

Indication for testing: Carrier screening or diagnostic testing for Usher syndrome types 1F and 3.

Positive: One pathogenic variant, p.R245X (c.733C>T), was detected in the PCDH15 gene; therefore, this individual is at least a carrier of Usher syndrome type 1F. Genetic counseling is recommended. This individual's reproductive partner should be offered screening for the disorder. At-risk family members should be offered testing to determine carrier status for the identified variant.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION; Usher Syndrome, Types 1F and 3 (PCDH15 and CLRN1), 2 Variants
CHARACTERISTICS: Usher syndrome type 1F is characterized by congenital, bilateral, profound sensorineural hearing loss, adolescent-onset retinitis pigmentosa and loss of vestibular function. Usher syndrome type 3 is characterized by post-lingual, progressive hearing loss, late-onset progressive vision loss due to retinitis pigmentosa and variable loss of vestibular function.
INCIDENCE: In Ashkenazi Jewish individuals - 1 in 20,500 for Usher syndrome type 1F; 1 in 82,000 for Usher syndrome type 3.
INHERITANCE: Autosomal recessive.
CAUSE: PCDH15 and CLRN1 pathogenic variants.
VARIANTS TESTED: PCDH15 p.R245X (c.733C>T), CLRN1 p.N48K (c.144T>G).
CLINICAL SENSITIVITY: In Ashkenazi Jewish individuals - 62 percent for Usher syndrome, type 1F; 98 percent for Usher syndrome, type 3. Sensitivities unknown in other ethnicities.
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.
LIMITATIONS: Variants other than those tested will not be detected. Diagnostic errors can occur due to rare sequence variations.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Usher Syndrome Types 1F and 3, Specimen	24-033-113089	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Usher Syndrome Types 1F and 3, Allele 1	24-033-113089	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Usher Syndrome Types 1F and 3, Allele 2	24-033-113089	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Usher Syndrome Types 1F and 3, Interp	24-033-113089	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