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

Joubert Syndrome Type 2 (TMEM216), 1 Variant

ARUP test code 2013909

Joubert Syndrome Type 2, Specimen	Whole Blood
Joubert Syndrome Type 2, Allele 1	c.218G>T *
Joubert Syndrome Type 2, Allele 2	Negative
Joubert Syndrome Type 2, Interp	See Note

Indication for testing: Carrier screening or diagnostic testing for Joubert syndrome type 2.

Positive: One copy of the p.R73L (c.218G>T) pathogenic variant in the TMEM216 gene was detected; therefore, this individual is at least a carrier of Joubert syndrome type 2. 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: Joubert Syndrome Type 2 (TMEM216), 1 Variant
CHARACTERISTICS: Joubert syndrome, type 2 is characterized by a "molar tooth sign" cerebellar and brain stem malformation, hypotonia and developmental delay. Clinical manifestations and severity of the syndrome vary.
INCIDENCE: 1 in 34,000 in Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: TMEM216 pathogenic variants.
VARIANTS TESTED: p.R73L (c.218G>T).
CLINICAL SENSITIVITY: 99 percent in Ashkenazi Jewish individuals; unknown in other ethnicities.
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.
LIMITATIONS: Variants other than p.R73L (c.218G>T) 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
Joubert Syndrome Type 2, Specimen	23-062-104500	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Joubert Syndrome Type 2, Allele 1	23-062-104500	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Joubert Syndrome Type 2, Allele 2	23-062-104500	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Joubert Syndrome Type 2, Interp	23-062-104500	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