ARUP LABORATORIES
800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
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:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Maple Syrup Urine Disease, Type 1B (BCKDHB), 3 Variants

ARUP test code 2013730

Maple Syrup Urine Disease, Specimen	Whole Blood
Maple Syrup Urine Disease, Allele 1	c.548G>C *
Maple Syrup Urine Disease, Allele 2	Negative
Maple Syrup Urine Disease, Interp	See Note

Indication for testing: Carrier screening or diagnostic testing for maple syrup urine disease type 1B.

Positive: One pathogenic variant, p.R183P (c.548G>C), was detected in the BCKDHB gene; therefore this individual is at least a carrier of maple syrup urine disease type 1B. At-risk family members should be offered testing to determine carrier status for the identified variant. This individual's reproductive partner should be offered screening for the disorder. Genetic counseling is recommended.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Maple Syrup Urine Disease, Type 1B (BCKDHB), 3 Variants
CHARACTERISTICS: Maple syrup urine disease (MSUD), type 1B most commonly presents in the first few days of life. Symptoms include irritability, poor feeding, lethargy, intermittent apnea and typically progresses to coma and death within 7 to 10 days if untreated.
INCIDENCE: 1 in 50,000 in Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: BCKDHB pathogenic variants.
VARIANTS TESTED: p.R183P (c.548G>C), p.G278S (c.832G>A), p.E372X (c.1114G>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 those tested will not be detected. Diagnostic errors can occur due to rare sequence variations.

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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Maple Syrup Urine Disease, Specimen	23-062-104237	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maple Syrup Urine Disease, Allele 1	23-062-104237	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maple Syrup Urine Disease, Allele 2	23-062-104237	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maple Syrup Urine Disease, Interp	23-062-104237	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