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

Lipoamide Dehydrogenase Deficiency (DLD), 2 Variants

ARUP test code 2013735

Lipoamide Dehydrogenase Def, Specimen	Whole Blood
Lipoamide Dehydrogenase Def, Allele 1	c.104dupA *
Lipoamide Dehydrogenase Def, Allele 2	Negative
Lipoamide Dehydrogenase Def, Interp	See Note

Indication for testing: Carrier screening or diagnostic testing for lipoamide dehydrogenase deficiency.

Positive: One pathogenic variant, p.Y35X (c.104dupA), was detected in the DLD gene; therefore, this individual is at least a carrier of lipoamide dehydrogenase deficiency. 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: Lipoamide Dehydrogenase Deficiency (DLD) 2 Variants
CHARACTERISTICS: Lipoamide dehydrogenase deficiency has a variable presentation that ranges from early-onset neurologic disease to adult-onset disease which is primarily hepatic. Early-onset neurologic disease presents in infancy with hypotonia, lethargy, vomiting and progressive encephalopathy resulting in death within the first or second year of life. Adult-onset primarily hepatic disease has a variable onset from infancy to the fourth decade and presents with liver injury or failure that is usually preceded by nausea and vomiting.
INCIDENCE: 1 in 35,000 in Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: DLD pathogenic variants.
VARIANTS TESTED: p.Y35X (c.104dupA), p.G229C (c.685G>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.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Lipoamide Dehydrogenase Def, Specimen	23-062-104284	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Lipoamide Dehydrogenase Def, Allele 1	23-062-104284	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Lipoamide Dehydrogenase Def, Allele 2	23-062-104284	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Lipoamide Dehydrogenase Def, Interp	23-062-104284	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