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

Canavan Disease (ASPA), 4 Variants

ARUP test code 0051453

Canavan Disease (ASPA), Specimen	Whole Blood
Canavan Disease (ASPA), Allele 1	c.693C>A *
Canavan Disease (ASPA), Allele 2	Negative
Canavan Disease (ASPA), Interpretation	See Note

Indication for testing: Carrier screening or diagnostic testing for Canavan disease.

Positive: One pathogenic variant, p.Y231X (c.693C>A), was detected in the ASPA gene; therefore, this individual is at least a carrier of Canavan disease. 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 this disorder. Genetic counseling is recommended.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Canavan Disease (ASPA), 4 Variants

CHARACTERISTICS: Canavan Disease is a neurodegenerative brain disorder that results in macrocephaly and lack of head control by 3 to 5 months of age. This progresses to a failure to achieve sitting, ambulation, or speech, and eventually leads to death typically in early childhood to teenage years.
INCIDENCE: 1 in 10,000 Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: ASPA pathogenic variants.
VARIANTS TESTED: c.433-2A>G, p.Y231X (c.693C>A), p.E285A (c.854A>C), and p.A305E (c.914C>A).
CLINICAL SENSITIVITY: 99 percent in Ashkenazi Jewish individuals; 55 percent 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
Canavan Disease (ASPA), Specimen	23-062-106318	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Canavan Disease (ASPA), Allele 1	23-062-106318	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Canavan Disease (ASPA), Allele 2	23-062-106318	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Canavan Disease (ASPA), Interpretation	23-062-106318	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