

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

ABCC8-Related Hyperinsulinism, 3 Variants

ARUP test code 2013725

ABCC-8 Related Hyperinsulinism, Specimen whole Blood

ABCC-8 Related Hyperinsulinism, Allele 1 Negative

ABCC-8 Related Hyperinsulinism, Allele 2 Negative

ABCC-8 Related Hyperinsulinism, Interp See Note

Indication for testing: Carrier screening or diagnostic testing for ABCC8-related hyperinsulinism.

Negative: This sample is negative for the three variants tested in the ABCC8 gene. If this is an asymptomatic individual of Ashkenazi Jewish descent, his/her risk of being a carrier of ABCC8-related hyperinsulinism has been reduced from 1 in 52 to approximately 1 in 1,700.

This result has been reviewed and approved by [REDACTED]

BACKGROUND INFORMATION: ABCC8-Related Hyperinsulinism, 3 Variants

CHARACTERISTICS: ABCC8-related hyperinsulinism is characterized by hypoglycemia varying in severity from mild symptoms to severe neonatal-onset. Infants with the severe neonatal-onset present with hypoglycemia within the first few days of life, which progresses causing seizures, brain damage and death if untreated.
INCIDENCE: 1 in 10,800 in Ashkenazi Jewish individuals.

INHERITANCE: Autosomal recessive.

CAUSE: ABCC8 pathogenic variants.

VARIANTS TESTED: p.F1388del (c.4163_4165del), p.V187D (c.560T>A), c.3992-9G>A.

CLINICAL SENSITIVITY: 97 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

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
ABCC-8 Related Hyperinsulinism, Specimen	23-304-101736	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
ABCC-8 Related Hyperinsulinism, Allele 1	23-304-101736	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
ABCC-8 Related Hyperinsulinism, Allele 2	23-304-101736	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
ABCC-8 Related Hyperinsulinism, Interp	23-304-101736	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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 23-304-101736
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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