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:	Female
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	c.4163_4165del *
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.

Positive: One pathogenic variant, p.F1388del (c.4163_4165del), was detected in the ABCC8 gene; therefore this individual is at least a carrier of ABCC8-related hyperinsulinism. 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: 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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
ABCC-8 Related Hyperinsulinism, Specimen	24-033-113088	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
ABCC-8 Related Hyperinsulinism, Allele 1	24-033-113088	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
ABCC-8 Related Hyperinsulinism, Allele 2	24-033-113088	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
ABCC-8 Related Hyperinsulinism, Interp	24-033-113088	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