ARUP LABORATORIES
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500 Chipeta Way, Salt Lake City, UT 84108-122
Tracy I. George, MD, Chief Medical Officer

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	[redacted]
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Fanconi Anemia, Group C (FANCC), 2 Variants

ARUP test code 0051468

Fanconi Anemia C (FANCC), Specimen	Whole Blood
Fanconi Anemia C (FANCC), Allele 1	c.456+4A>T *
Fanconi Anemia C (FANCC), Allele 2	Negative
Fanconi Anemia C (FANCC), Interp	See Note

Indication for testing: Carrier screening or diagnostic testing for Fanconi anemia, group C.

Positive: One pathogenic variant, c.456+4A>T, was detected in the FANCC gene; therefore, this individual is at least a carrier of Fanconi anemia group C. Genetic counseling is recommended. This individuals 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], Ph.D.

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

BACKGROUND INFORMATION: Fanconi Anemia, Group C (FANCC),
2 Variants:
CHARACTERISTICS: Fanconi anemia, group C is characterized by the following symptoms: short stature, abnormal skin pigmentation, and multiple malformations that may affect eyes, ears, heart, oral cavity, thumbs, forearms, kidneys, or urinary tract. Other symptoms may include hearing loss, hypogonadism, and developmental delay. Progressive bone marrow failure occurs during the first decade of life. Hematologic malignancies occur in approximately 20 percent of affected individuals. Nonhematologic malignancies occur in approximately 30 percent of affected individuals.
INCIDENCE: 1 in 32,000 Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: FANCC pathogenic variants.
VARIANTS TESTED: p.D23Ifs (c.67delG) and c.456+4A>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 p.D23Ifs (c.67delG) and c.456+4A>T 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
Fanconi Anemia C (FANCC), Specimen	21-114-400620	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fanconi Anemia C (FANCC), Allele 1	21-114-400620	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fanconi Anemia C (FANCC), Allele 2	21-114-400620	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fanconi Anemia C (FANCC), Interp	21-114-400620	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