ARUP LABORATORIES
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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	7/15/2000
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	Negative
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. Negative: This sample is negative for the two variants tested in the FANCC gene. If this is an asymptomatic individual of Ashkenazi Jewish descent, his/her risk of being a carrier of Fanconi anemia group C disease is reduced from 1 in 89 to approximately 1 in 8,800. This result has been reviewed and approved by [redacted]

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	23-332-400170	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fanconi Anemia C (FANCC), Allele 1	23-332-400170	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fanconi Anemia C (FANCC), Allele 2	23-332-400170	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Fanconi Anemia C (FANCC), Interp	23-332-400170	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