ARUP LABORATORIES
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Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108
UNITED STATES

Physician: arup, arup

Patient: Test, BLM

DOB
Sex: Female
Patient Identifiers: 46931
Visit Number (FIN): 47269
Collection Date: 3/3/2023 11:31

Bloom Syndrome (BLM), 1 Variant

ARUP test code 0051433

Bloom Syndrome (BLM), Specimen	Whole Blood
Bloom Syndrome (BLM), Allele 1	2207_2212delin *
Bloom Syndrome (BLM), Allele 2	Negative
Bloom Syndrome (BLM), Interpretation	See Note

Indication for testing: Carrier screening or diagnostic testing for Bloom syndrome.

Positive: One copy of the p.Y736Lfs (c.2207_2212delinsTAGATTC) pathogenic variant in the BLM gene was detected; therefore, this individual is at least a carrier of Bloom syndrome. Genetic counseling is recommended. This individual's 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]

BACKGROUND INFORMATION: Bloom Syndrome (BLM), 1 Variant

CHARACTERISTICS: Bloom syndrome is characterized by pre- and postnatal growth deficiency, sparse subcutaneous tissue, sun-sensitive telangiectatic hypo- and hyperpigmented skin lesions, chromosome instability causing benign and malignant tumors early in life, and male sterility.
INCIDENCE: 1 in 40,000 in Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: BLM pathogenic variants.
VARIANT TESTED: p.Y736Lfs (c.2207_2212delinsTAGATTC).
CLINICAL SENSITIVITY: 97 percent in Ashkenazi Jewish individuals, approximately 3 percent in other ethnicities.
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.
LIMITATIONS: Variants other than c.2207_2212delinsTAGATTC 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.

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

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Bloom Syndrome (BLM), Specimen	23-062-106139	3/3/2023 11:31:00 AM	3/3/2023 11:31:57 AM	4/13/2023 1:16:00 PM
Bloom Syndrome (BLM), Allele 1	23-062-106139	3/3/2023 11:31:00 AM	3/3/2023 11:31:57 AM	4/13/2023 1:16:00 PM
Bloom Syndrome (BLM), Allele 2	23-062-106139	3/3/2023 11:31:00 AM	3/3/2023 11:31:57 AM	4/13/2023 1:16:00 PM
Bloom Syndrome (BLM), Interpretation	23-062-106139	3/3/2023 11:31:00 AM	3/3/2023 11:31:57 AM	4/13/2023 1:16:00 PM

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