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	[redacted]
Sex:	[redacted]
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Bloom Syndrome (BLM), 1 Variant

ARUP test code 0051433

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

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

Negative: This sample is negative for the p.Y736Lfs (c.2207_2212delinsTAGATTC) pathogenic variant in the BLM gene. If this is an asymptomatic individual of Ashkenazi Jewish descent, his/her carrier risk is reduced from 1 in 100 to approximately 1 in 3,300.
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.

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

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Bloom Syndrome (BLM), Specimen	21-294-117443	10/21/2021 11:52 00 AM	10/22/2021 4:14:25 PM	10/28/2021 11:09:00 AM
Bloom Syndrome (BLM), Allele 1	21-294-117443	10/21/2021 11:52 00 AM	10/22/2021 4:14:25 PM	10/28/2021 11:09:00 AM
Bloom Syndrome (BLM), Allele 2	21-294-117443	10/21/2021 11:52 00 AM	10/22/2021 4:14:25 PM	10/28/2021 11:09:00 AM
Bloom Syndrome (BLM), Interpretation	21-294-117443	10/21/2021 11:52 00 AM	10/22/2021 4:14:25 PM	10/28/2021 11:09:00 AM

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

END OF CHART

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