

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

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 4/22/1999  
**Gender:** Unknown  
**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 **See Note** \*

Bloom Syndrome (BLM), Allele 2 **See Note** \*

Bloom Syndrome (BLM), Interpretation See Note

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.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

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

Unless otherwise indicated, testing performed at:

**ARUP LABORATORIES | 800-522-2787 | aruplab.com**  
500 Chipeta Way, Salt Lake City, UT 84108-1221  
Tracy I. George, MD, Laboratory Director

Patient: Patient, Example  
ARUP Accession: 19-350-118602  
Patient Identifiers: 01234567890ABCD, 012345  
Visit Number (FIN): 01234567890ABCD  
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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Bloom Syndrome (BLM), Specimen	19-350-118602	12/16/2019 12:05:00 PM	12/16/2019 3 27:38 PM	12/16/2019 3:50:00 PM
Bloom Syndrome (BLM), Allele 1	19-350-118602	12/16/2019 12:05:00 PM	12/16/2019 3 27:38 PM	12/16/2019 3:50:00 PM
Bloom Syndrome (BLM), Allele 2	19-350-118602	12/16/2019 12:05:00 PM	12/16/2019 3 27:38 PM	12/16/2019 3:50:00 PM
Bloom Syndrome (BLM), Interpretation	19-350-118602	12/16/2019 12:05:00 PM	12/16/2019 3 27:38 PM	12/16/2019 3:50:00 PM

END OF CHART

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

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