

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 12/31/1752
Gender: Male
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

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:

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Bloom Syndrome (BLM), Specimen	20-042-101019	2/11/2020 6:06 00 AM	2/11/2020 6:06:40 AM	2/11/2020 10 00:00 AM
Bloom Syndrome (BLM), Allele 1	20-042-101019	2/11/2020 6:06 00 AM	2/11/2020 6:06:40 AM	2/11/2020 10 00:00 AM
Bloom Syndrome (BLM), Allele 2	20-042-101019	2/11/2020 6:06 00 AM	2/11/2020 6:06:40 AM	2/11/2020 10 00:00 AM
Bloom Syndrome (BLM), Interpretation	20-042-101019	2/11/2020 6:06 00 AM	2/11/2020 6:06:40 AM	2/11/2020 10 00:00 AM

END OF CHART

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: 20-042-101019
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Page 2 of 2 | Printed: 1/29/2021 6:26:48 AM
4848