

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 7/29/2017
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 01/01/2017 12:34

Chromosome Analysis - Breakage, Fanconi Anemia, Whole Blood

ARUP test code 0097688

Chromosome Analysis, Breakage, Fanconi See Note

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

Unless otherwise indicated, testing performed at:

Test(s) Ordered: Fanconi Breakage

Specimen Submitted: PERIPHERAL BLOOD

Submitted ICD9 Code: none provided

Clinical History: none provided

Breakage analysis*:

Cells analyzed per clastogen:	50
DEB [0.1 mcg/mL] cells with radials:	21
DEB [0.1 mcg/mL] breaks/cell w/o radials:	1.13
MMC [40 ng/mL] cells with radials:	30
MMC [40 ng/mL] breaks/cell w/o radials:	0.5

*DEB control values have been previously established in this laboratory. Absent radial figures and <0.5 breaks/cell is considered negative; few or no radials and 0.5-1.0 breaks/cell is considered negative but may indicate need for retesting; multiple radials and >1.0 breaks/cell is considered positive. MMC breakage values are variable and interpreted in the context of concurrent normal control cultures.

Chromosome Analysis:

Peripheral blood was cultured with the clastogenic agents diepoxybutane (DEB) and mitomycin C (MMC) to induce Fanconi Anemia (FA)-related chromosome breakage. Breakage analysis was performed on unbanded chromosome preparations. Fifty cells each from the two culture systems were screened for chromosome breakage with the above results. When compared to negative control values, this analysis is considered positive for FA-related chromosome breakage.

Comments:

Both DEB and MMC clastogenic stress culture systems are positive for FA-related breakage. In particular, MMC culture demonstrated radial figures, the hallmark of FA-related chromosome breakage repair deficit, in a majority of the 50 examined cells. Genetic counseling is recommended.

Interpretation:

Positive for Fanconi-related chromosome breakage

I have reviewed the specimen and agree with the interpretation above. MELANIE MANNING, M.D.
Electronically signed
Stanford University MC Cytogenetics Laboratory, 3375 Hillview Ave., Palo Alto, CA 94304

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: 18-115-112955
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Chromosome Analysis, Breakage, Fanconi	18-115-112955	4/25/2018 12:52:00 PM	4/26/2018 6:07:50 AM	5/14/2018 6:00:00 PM

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
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Patient: Patient, Example
ARUP Accession: 18-115-112955
Patient Identifiers: 01234567890ABCD, 012345
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