LABORATORY REPORT

Redcliffe labs

Patient Name	:	Bill Date	:
DOB/Age/Gender	:	Sample Collected	:
Patient ID / UHID	:	Sample Received	:
Referred By	:	Report Date	:
Sample Type	:	Barcode No	:
Client	:	Report Status	:

CYTOGENETICS REPORT

Fanconi Anemia (FA) Stress Cytogenetics

Mitomycin-C induced Chromosome Breakage Study For Fanconi Anaemia (FA).

CLINICAL HISTORY To rule out any chromosomal breakages

SUMMARY OF RESULTS **NOT SENSTIVE TO MITOMYCIN C**

MITOMYCIN TREATED CULTURE FOR SOLID GEIMSA STAIN **Page 2**

No significant numbers of (a) Chromatid gap, (b) Chromatid break (c) Triradials (d) Quadriradial (e) Chromatid interchange observed in both patient and control sample.

CLINICAL INTERPRETATION **Page 3**

No significant number of chromatid gap, chromatid break, triradials, quadriradial and chromatid interchange observed in both patient and control sample. These results should be interpreted in correlation of other clinical features/symptoms. Fanconi's anaemia is one of the chromosomal instability syndromes characterized by chromosomal breakages due to a deficient DNA repair system. When FA is suspected, a chromosome stress test with MMC should is performed in order to look for increased chromosome breakage, particularly radial formations.The breakages appear in the form of chromatid gaps and reunion give rise to chromosomal radial formation. The test assay is based on the observation of increased breakages and radial formation in patients as opposed to sex and age matched control because the patients blood cells are hypersensitive to Mitomycin C.

TEST DESCRIPTION

72 hours of PHA stimulated unsynchronized peripheral blood culture was initated in PB-MAX (Thermofischer) Karyotyping medium along with the two concentrations (50 ng/mL & 100 ng/mL) of Mitomycin C. Age and sex matched normal control peripheral blood was also set as above using Mitomycin C.

RECOMMENDATION Genetic Counselling for the family is recommended.

Dr. Ashish Fauzdar
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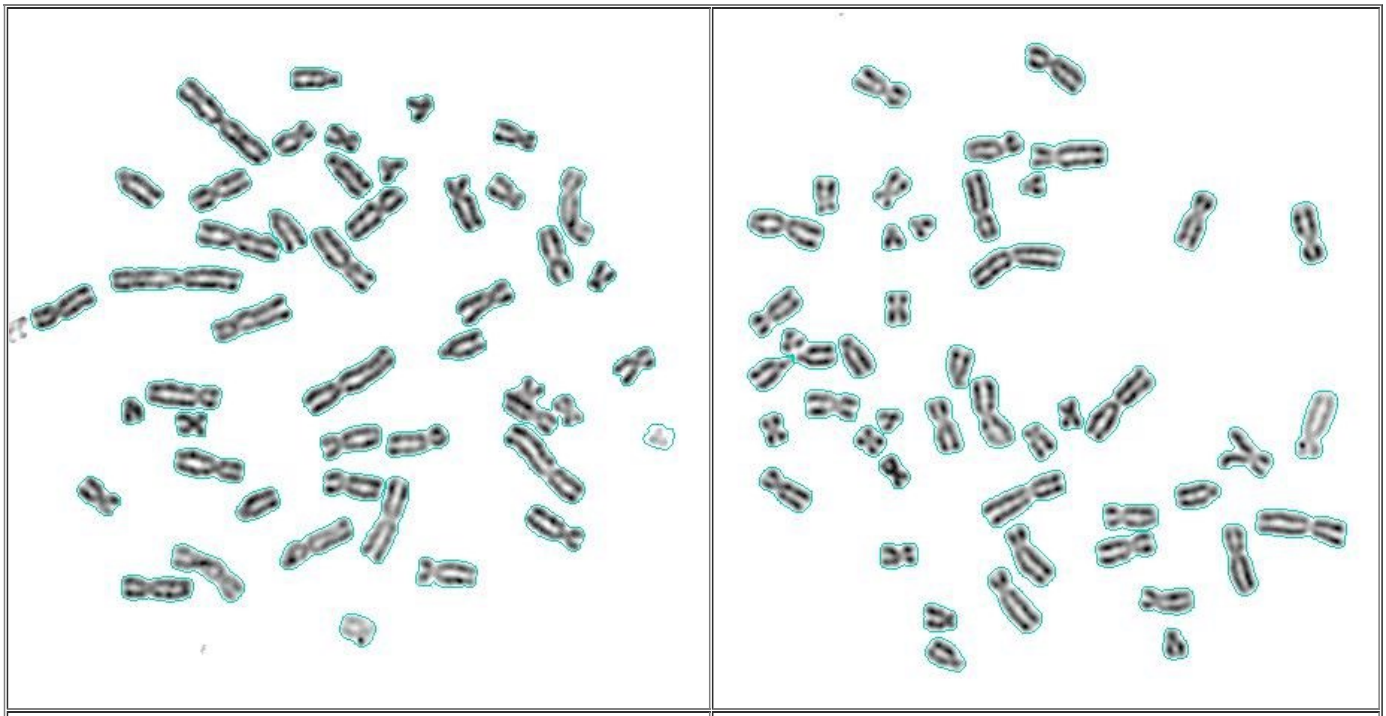
MITOMYCIN-C TREATED METAPHASES WITH SOLID GEIMA STAIN



Patient's Sample	Control Sample

ANALYSIS	Patient	Control
Mitomycin C added / Culture	50ng/mL and 100ng/mL	50ng/mL and 100ng/mL
Number of metaphases analyzed	50	50
Number of cells with chromatid gap, chromatid break, triradials, quadriradial and chromatid interchange	2	1
Number of chromatid gap, chromatid break, triradials, quadriradial and chromatid interchangeformation	1	1
Percentage of aberrant cells (%)	4%	2%

RESULTS:

The patient is not sensitive to Mitomycin-C without any significant numbers of chromatid gap, chromatid break, triradials, quadriradial and chromatid interchange observed in both patient and control sample. Kindly correlate clinically.

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INTERPRETATION OF CHROMOSOME BREAKAGE TEST RESULTS:

Formula to calculate sensitivity to Mitomycin C (% of aberrant cells):

Percentage of cells with triradicals + 1.6 times the total number of triradials

>40	Hypersensitive to MMC
<10-40	Equivocal to MMC
0-10	Not sensitive to MMC

=1+(1.6x4)

=7.4

Sensitivity of Mitomycin C: Identify the disease-causing genetic mutation using the molecular methods.

Non-Sensitivity of Mitomycin C: If clinical evidence of Fanconi Anemia is weak, no further studies are needed, if there is strong clinical suspicion of FA

1. Skin fibroblast testing should be performed to rule out the possibility of mosaicism.

2. Mutation analysis for other disorders that have some clinical features in common with FA and are associated with some form of chromosome instability.

Equivocal:
1. Skin fibroblast testing should be performed to rule the mosaicism.
2. Mutation analysis for condition other than FA that manifests with increased chromosomal breakage such as Nijmegen breakage syndrome, ataxia telangiectasia like disorder, DNA ligase 4 syndrome, Seckel syndrome1, Bloom syndrome, dyskeratosis congenita, Roberts syndrome, Warsaw breakage syndrome, Cornelia de Lange syndrome, or FAN1 deficiency.

Limitations:
1. The MMC stress test focuses on chromosome breakage and does not identify constitutional chromosome abnormalities. If a constitutional chromosome abnormality is suspected, a separate conventional cytogenetic study is necessary, if clinically required.
2. Since underlying genes of the chromosomal breakage syndrome have been identified and molecular methods are also available and may be offered to the patient if clinically indicated.

References:
1. Daniel g. Kuffel *et al*., Mitomycin c Chromosome Stress Test. *Mayo Clin proc* 1997; 72:579-580.
2. Fanconi Anaemia: Guidelines for Diagnostic and Managements, 4th Edition, 2014.
3. Anneke B. Oostra etal.,Diagnosis of Fanconi Anemia: Chromosomal Breakage Analysis, *Anemia, vol 2012, Article ID 238731.*

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