CYTOGENETICS

#Stress Cytogenetics

KARYOTYPE : 46,XX

STRUCTURAL ABERRATIONS : Nil

NUMERICAL ABERRATIONS : Nil

OBSERVATIONS : The percentage of breaks and radials are not increased in the patient at highest dose of MMC.

Normal culture and 50 ng/ml MMC induced culture did not show any significant breakages as compared to control individual.

0.27 breaks/cell were observed in cultures at a final concentration of 100 ng/ml compared to 0.04 breaks/cell in control.

The percentage of breaks is given in table 1, 2 & 3.

The karyotype of the patient is normal.

COMMENTS : The report is enclosed herewith and is normal for chromosomal breakage syndrome. There are no significant structural aberrations found in metaphases studied as compared to control individual.

PROTOCOL ADAPTED FOR CHROMOSOME BREAKAGE STUDY FOR FANCONI'S ANEMIA

The peripheral blood was collected in sodium heparin vacutainer in sterile conditions. A sex and age matched healthy individual with no history of alcohol, smoke and drugs was taken as control. The peripheral blood culture were set-up in Karyomax, (Gibco, USA) by following 3 different methods:

1. A set of two blood cultures of patient and healthy individual, single for each, were set in Karyomax, (Gibco, USA) for 72 hours with no addition of any breakage inducing agent.

2. An another set of two blood cultures of patient and healthy individual, single for each, were set in Karyomax, (Gibco, USA) for 72 hours. A breakage inducing agent Mitomycin-C at a concentration of 50 ng/ml was added at 48 hours of initiation of culture.

3. A third set of two blood cultures of patient and healthy individual, single for each, were set in Karyomax, (Gibco, USA) for 72 hours. A breakage inducing agent Mitomycin-C at a concentration of 100 ng/ml was added at 48 hours of initiation of culture.

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TABLE 1, 2 AND 3 LISTING THE RESULTS OF ABERRATION ANALYSIS OF METAPHASES.

Table 1. Aberration Analysis of Non-Induced Culture

	Total Cells	Aberrant Cells	Percent Aberration	Total cht gap	Total cht break	Tri radial	Quadri radial	Other	Total breaks	Breaks/cell
Patient	50	0	0	0	0	0	0	0	0	0.00
Healthy Individual	50	0	0	0	0	0	0	0	0	0.00

Table 2. Aberration Analysis of Mitomycin-C Induced Culture- (50 ng/ml concentration)

	Total Cells	Aberrant Cells	Percent Aberration	Total cht gap	Total cht break	Tri radial	Quadri radial	Other	Total breaks	Breaks/cell
Patient	50	4	8	4	3	0	0	0	7	0.13
Healthy Individual	50	1	2	1	0	0	0	0	1	0.02

Table 3. Aberration Analysis of Mitomycin-C Induced Culture- (100 ng/ml concentration)

	Total Cells	Aberrant Cells	Percent Aberration	Total cht gap	Total cht break	Tri radial	Quadri radial	Other	Total breaks	Breaks/cell
Patient	50	7	14	8	6	0	0	0	14	0.27
Healthy Individual	50	2	4	2	0	0	0	0	2	0.04

Calculation of Aberration Rate:

1. Chromatid gaps and chromatid breaks are counted as one breakage event.
1. Tri radials and quadri radials are counted as 2 break events each.
1. Other chromatid interchange figures are counted as 8 break events.

*cht g: chromatid gap, cht b: chromatid break, tri: tri radial, quad: quadri radial

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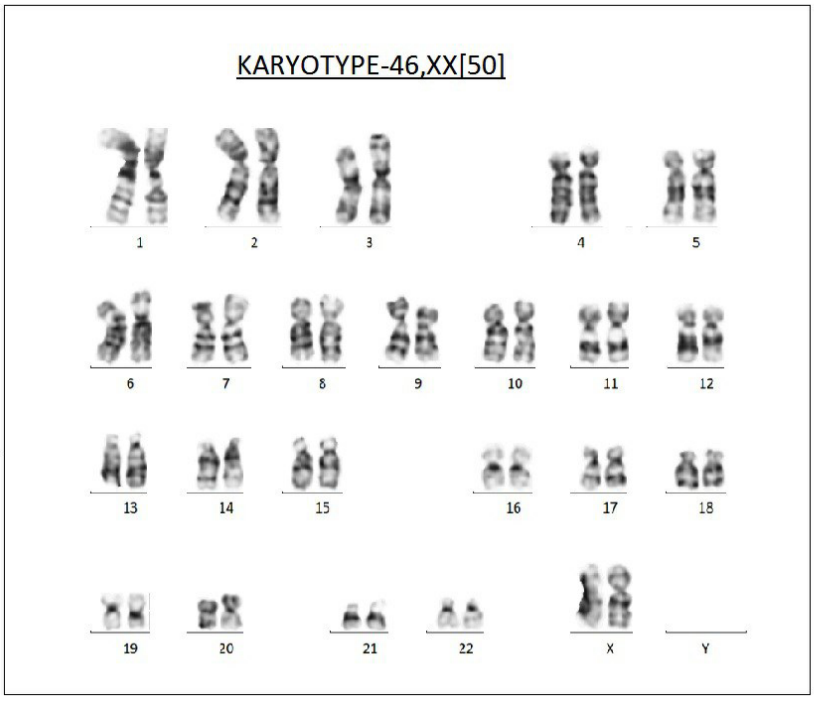

Correlate Clinically.

*** End Of Report ***