

Stress Cytogenetics Test for Fanconi's Anaemia

SPECIMEN: Peripheral blood

CLINICAL HISTORY:

PREPARATION: 72 hours PHA stimulated unsynchronized culture using two concentrations (50 ng/mL and 100 ng/mL) of Mitomycin C. Age and sex matched normal control peripheral blood was also set as above using Mitomycin C.

STAINING: Giemsa

	Patient	Control
Mitomycin C added / culture	50 ng/mL & 100 ng/mL	50 ng/mL & 100 ng/mL
Number of metaphases analyzed	50	50
Number of cells with triradial / quadriradial formation		
Number of triradials formed		
Percentage of aberrant cells		

Formula to calculate sensitivity to Mitomycin C*

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

Cut off value for sensitivity to Mitomycin C > 40

Free text 5 lines

RESULT: SENSITIVE / NOT SENSITIVE TO MITOMYCIN C.

Note: The patient is not sensitive for Mitomycin C; therefore, these results must be interpreted in the light of the clinical features.

*Ref: Mayo Clin Proc 1997; 72: 579-580.

PHOTO

Fanconi's anaemia is one of the Chromosomal Instability Syndromes characterized by chromosomal breakages due to a deficient DNA repair system. The breakages appear in the form of chromatid gaps and reunion give rise to chromosomal radial formations. The test assay is based on the observation of increased breakages and radial formations in patients as opposed to sex and age matched controls because the patient's blood cells are hypersensitive to Mitomycin C.

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