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Photo Imaging

Images in Clinical Medicine

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A 15-year-old girl presented with progressive fatigue for 6 months, multiple purpuric spots on both lower limbs, and bleeding gums for the last couple of days, for which she was admitted to Dhaka Medical College Hospital. She acknowledged having repeated upper respiratory tract infections, low-grade fever, and becoming progressively pale in recent months. During her previous admission, she was diagnosed as a case of pancytopenia of unknown aetiology with a normal bone marrow cellularity. She got two units of whole blood transfusion.

Physical examination revealed a severely anaemic girl of short stature (4'6", <5th percentile), frontal bossing, polydactyly (six digits of the left hand), Café au lait spots, and a hypopigmented macule on the back, non-palpable purpuric spots and ecchymoses on limbs, gum bleeding, and ecchymoses on the tongue. There was no organomegaly or lymphadenopathy. We had a provisional diagnosis of **Aplastic anemia due to Fanconi's anemia.**

Investigation revealed: hemoglobin- 4.8 gm/dl, total leukocyte count- 2.07 /mm3, platelet 20.000/mm3, ESR-91, Peripheral blood film: anisocytosis, anisochromic RBC with few macrocytes and a few rouleaux formation; comment: pancytopenia. LDH, ALT, S. bilirubin, and S. vitamin B12- all were within normal limits. HBsAg and Anti-HCV were negative, USG of the whole abdomen was normal. X-ray hands: polydactyly and clinodactyly(left), Bone marrow: Hypocellular marrow due to aplastic anaemia.

Chromosome breakage test: Sensitive to mitomycin C, a Significant number of chromosomal breaks, chromatid gaps, and tri/quadri radials were observed in the supplied sample, as compared to controls.

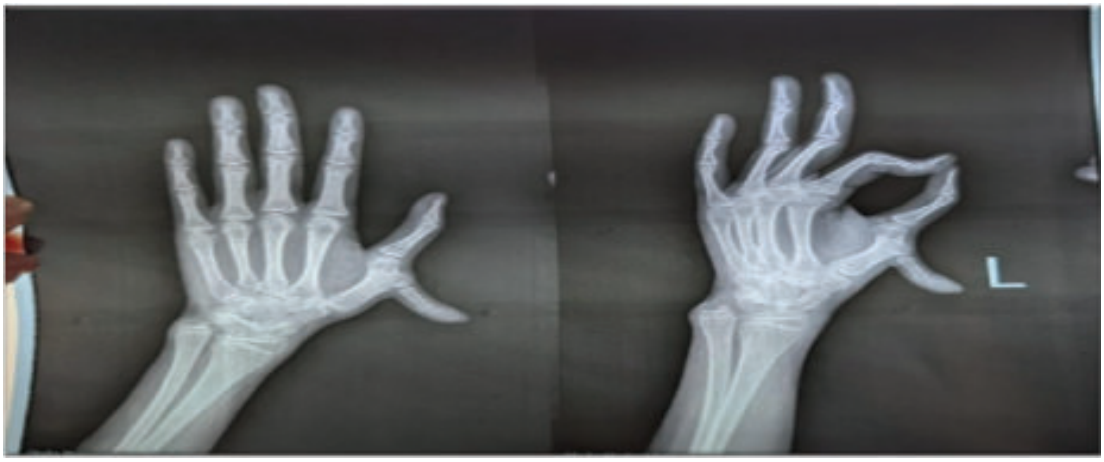

Photo 1: X-ray, clinodactyly and polydactyly

Inference: Positive for chromosomal breakage (suggestive of Fanconi's anemia)

- **Fanconi's anemia (FA):** Is a rare inherited bone marrow failure syndrome (IBMFS) characterized by pancytopenia, predisposition to malignancy, and characteristic physical abnormalities and congenital malformations. FA is caused by pathogenic variants (i.e., mutations) in one of numerous genes involved with deoxyribonucleic acid (DNA) repair. DNA damage that occurs during foetal development leads to congenital anomalies, childhood & adulthood bone marrow failure, organ damage, cancer predisposition.

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STRESS CYTOGENETICS TEST FOR FANCONI ANAEMIA @

Result Summary
SENSITIVE TO MITOMYCIN C

Interpretation
Significant number of chromatid breaks, chromatid gaps and tri/quadri radials were observed in patient sample as compared to control.

Positive for chromosomal breakages.

Advised: Kindly correlate with clinical findings including CBP parameters and treatment history.

Specimen
Peripheral Blood (in Heparin Vacutainer)

Reason for Referral
Anaemia/Cytopenia under evaluation

Method
PHA-stimulated (72 hours) cultures using two concentrati (50 ng/ml and 100 ng/ml) of Mitomycin C. The metaphas were stained using Giemsa stain.

Cut off values

Range	Category
0-10	Not Sensitive to MMC
<10-40	Equivocal to MMC
>40	Sensitive to MMC

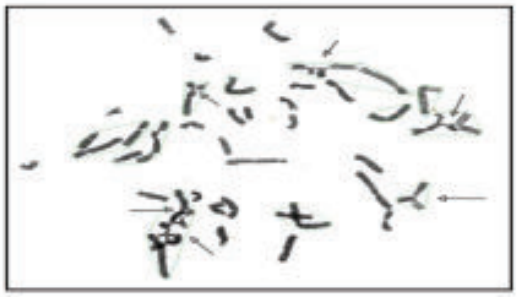
Formula to calculate sensitivity to Mitomycin C

Percentage of cells with Tri radials/Quadri radials + 1.6 times the total number of Tri radials/Quadri radials

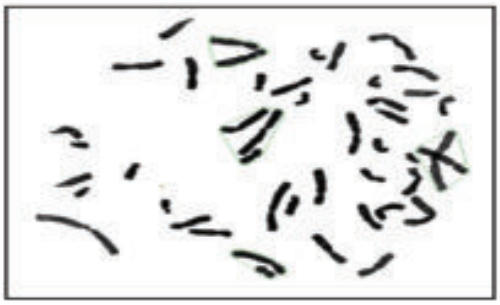
58.5 + [1.6×162] = 58.5 + 259.2 =	**317.7**

Photo 2: Stress Cytogenetics Test for Falconi Anaemia

Representative Metaphases



PATIENT SAMPLE



CONTROL SAMPLE

Chromosomal Aberrations Analysis

Sample Details	Chromosomal aberrations	Non – induced culture (No Mitomycin C added)	Mitomycin C induced culture (Conc.: 50 ng/ml)	Mitomycin C induced culture (Conc.:100 ng/ml)
Patient sample	Total cells analysed	25	25	25
	Total aberrant cells	0	23	22
	Percentage Aberration	0	29.9	28.6
	Total number of Tri radials	0	46	58
	Total number of Quadri - radials	0	28	30
	Total breaks & chromatid gaps	0	42	49
	Breaks/aberrant cell	0	20	19

Photo 3: Microscopic Chromosomal Aberrations Analysis