A RARE CASE OF PANCYTOPENIA

Dr Manaswini Edara

DEPT OF GENERAL MEDICINE

A 20-year-old female, student, presented with c/o

- shortness of breath
- easy fatiguability
- generalised weakness.

} since 3 months

- No history of chest pain, cough, cold, orthopnea or PND.

PAST HISTORY:

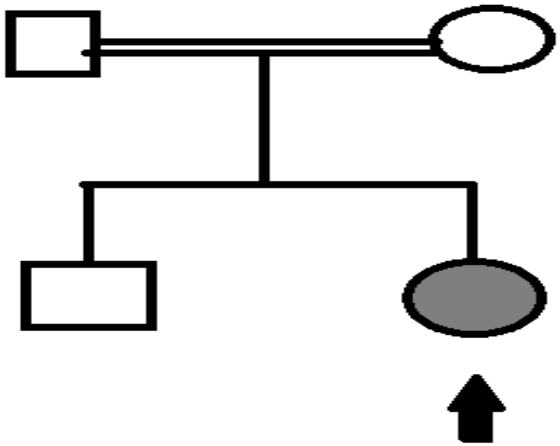
No h/o Diabetes mellitus, Hypertension, Tuberculosis, bronchial asthma.

No previous h/o blood transfusions, jaundice in the past.

No h/o drug intake

FAMILY HISTORY:

Born in a second degree consanguinous marriage.



PERSONAL HISTORY:

- Mixed diet
- Apetite -Normal
- Sleep- normal
- Bowel and bladder habits unaltered
- No addictions

MENSTRUAL HISTORY:

Menarche at 12 years of age.

Regular cycles.

-

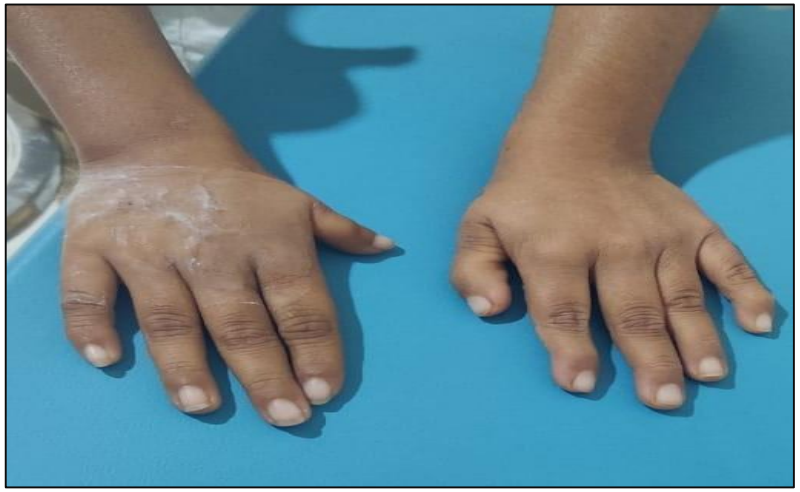
ON EXAMINATION :

Patient was conscious, oriented to time, place and person.

- Afebrile
- Pulse rate-98 bpm
- BP-90/60 mm of Hg
- SpO2 -98% on Room Air
- RR- 18/min

- Severe pallor present.
- Mild pitting pedal edema present.
- No cyanosis, clubbing, lymphadenopathy, koilonychia.
- No knuckle hyperpigmentation.

- Patient had short stature and right hand hypoplastic thumb.
- Height-143cm
- Upper segment-80 cm
- Lower segment-63cm
- US:LS=1.2
- Mother’s height-154 cm
- Father’s height-164 cm
- Brother’s height-168 cm



- CVS- S1S2 present
- RS-AEBE
- P/A- Soft, No tenderness

  No Organomegaly
- CNS- Conscious

  No motor or sensory neurological deficit

Hb	3.4 gm/dl (14 - 17.5)
TLC	3800 cells/cumm (4000-10,000)
PLATELET	48000cells/cumm (150000-450000)
HCT	9.6% (41-51.4)
MCV	111 fL (80-96)
RDW	15.6% (12.2-16.1)
RETIC COUNT	2.7% (0.6%)

HIV	Negative
HBsAg	Negative
Anti -HCV	Negative

Total Bilirubin	0.6
Direct bilirubin	0.19
Indirect bilirun	0.42
SGOT	20
SGPT	19
ALP	81

Total protein	6.5
Albumin	4.4
Globulin	2.1

Urea	18
Creatinine	0.61

Na	138
K+	3.6

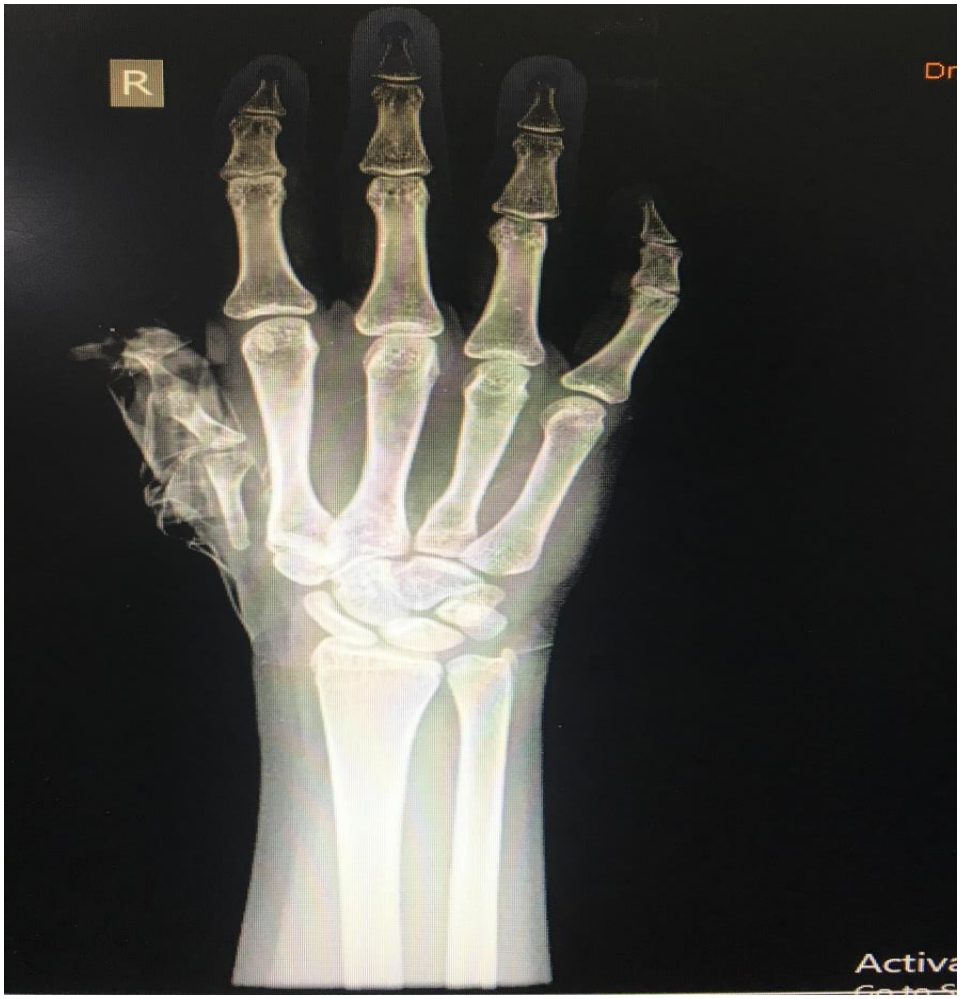
VIT B12	238 pg/mL
LDH	198 U/L
ESR	34 mm/hr
CRP	6.2 mg/L

- **BONE MARROW ASPIRATION** : Dry tap

- **BONE MARROW BIOPSY**-Bone marrow shows marrow spaces mostly occupied by mature adipocytes and markedly reduced hematopoietic cells s/o aplastic anemia.

- **Provisional diagnosis: Aplastic anemia(? Fanconi anemia)**

➢**X-Ray right hand:** Right 1st finger appears shortened with short and thin metacarpal,proximal and distal phalanx. Significant gap is noted at the 1st carpo-metacarpal joint.

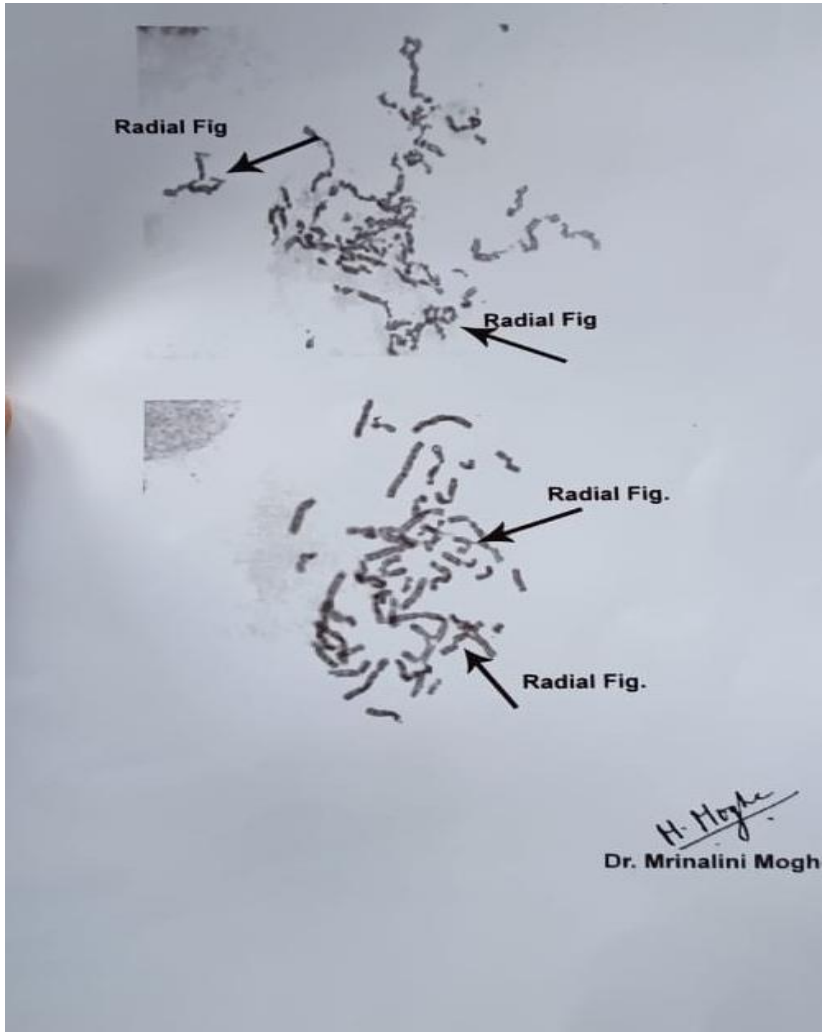


- **2D ECHO**- Ejection fraction-60%
  Normal chambers, normal valves, No RWMA.

- **USG (ABD/PELVIS) :** No organomegaly. No other abnormality detected**.**

- **Chromosomal breakage test:**

Significant increase in chromosomal breaks and chromatid exchange radial formations caused by mitomycin C s/o Fanconi anemia.



- **Final Diagnosis : FANCONI ANEMIA**

- Patient was started on Tablet DANAZOL 200mg BD

- Patient and relatives were counselled regarding bone marrow transplantation.

- Patient is in regular follow-up in Medicine OPD.

FANCONI ANEMIA

- Fanconi anemia- an autosomal recessive disorder.
- It manifests as congenital developmental anomalies, progressive pancytopenia, and an increased risk of malignancy.
- Patients of Fanconi anemia typically have short stature, café au lait spots, and anomalies involving thumb, radius, and genitourinary tract.
- Atleast 17 different genetic defects have been defined. The most common, type A Fanconi anemia, is due to mutation of FANCA.
- Most of the Fanconi anemia gene produts form a protein complex that activates FANCD2 by monoubiquitination to play a role in the cellular response to DNA damage and especially interstrand cross-linking.
- Chromosomes in Fanconi anemia are susceptible to DNA cross-linking agents, which is the basis for the diagnostic assay.

TAKE HOME MESSAGE

- Young patients with pancytopenia have to be carefully assessed for any other congenital abnormalities to rule out any inherited causes of aplastic anemia which are treatable.

- Fanconi anemia is a rare cause of aplastic anemia which may progress to bone marrow failure or AML or Myelodysplastic syndrome.

- Early detection and treatment with bone marrow transplantation may prevent the complications.