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# Congestive Cardiac Failure in a Case of Fanconi Anaemia: Usual Presentation in a Rare Disease

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***Authors' contributions***

*This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.*

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***Case Study***

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## ABSTRACT

The present study highlights an usual case of congestive cardiac failure. Cardiovascular, gastrointestinal or renal anomalies can also occur. FA "facies" microcephaly, small eyes, epicanthal folds, abnormal ears. A 11 year old male presented in emergency with complain of generalized weakness and loss of apetite from 10 days, fever and cough from 5 days, vomiting(3-4 episodes/day) from 3 days. He had a history of similar episode and blood transfusion 2 months back. In our case patient presented in shock with CHF. He had severe anemia with hypoplastic thumb with short stature underdeveloped penis and abnormal pigmentation.

*Keywords: Pancytopenia; hypoplastic thumb; depigmentation; short stature; congestive cardiac failure.*

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## 1. INTRODUCTION

Fanconi anemia is the most common type of inherited pancytopenia .It is an autosomal recessive disease characterized by spontaneous chromosomal fragility,which is increased after exposure of peripheral blood lymphocytes to DNA crosslinking agents such as mitomycin C,melphalan. Often appear as thrombocytopenia-granulocytopenia –micro-cytopenia.

Generally presented with hyperpigmentation (café-au-lait spots and vitiligo), short stature, hypothyroidism,absence of radii,anomalies of thumb,feet,hip dislocation. Males can have underdeveloped penis and females can have grnital anomalies. Cardiovascular, gastro-intestinal or renal anomalies can also occur. Fanconi anemia facies, microcephaly, small eyes, epicanthal folds, abnormal ears.

## 2. CASE PRESENTRATION

**HOPI** :A 11 year old male presented in emergency with complain of generalized weakness and loss of apetite from 10 days,fever and cough from 5 days, vomiting(3-4 episodes/day) from 3 days.He had a history of similar episode and blood transfusion 2 months back.

**O/E:** Physical examination revealed severe pallor, hypoplastic thumb, dystrophic nails, short staure and abnormal pigmentation in hand and foot underdeveloped penis.

PP was feeble. 4 limb bp RA -92/60(<50%) LA-90/56(50%)LL-88/44(<10%)RL-86/40(<10%)

CVS examination- S2 is loud,systolic murmur of grade 3 was present in left lower sternal border radiating to carotid and aortic area.

Bilateral basal crepts were present. On P/A examination tense hypocondrium with no organomegaly was present.

**Course during hospital stay:** i/v/o severe pallor and hb 1gm/dl 1 unit of prbc was transfused with mid BT lasix and inotropes (dopamine and dobutamine) were given for 36 hrs then slowly tappered and stopped.

On day 3,1 prbc was transfused again@ 15 ml/kg with mid BT lasix and 1 platlet @ 1o ml/kg. prophylacyic antibiotics(piptaz and linezolid) were given for 7 days.

**Investigations:** Hb was 1gm/dl Hct -4.2% corrected retic count -0.08%on admission with pancytopenia.

LDH -463 U/L(H) FERRITIN-1566ng/ml(H) vit B12 -1058(H) TRANSFERRIN -96.4%(H) serum iron -295ug/dl TIBC-306ugm/dl UIBC-11ugm/dl.

Bone Marrow shows reduced erythropoiesis, myelopoiesis and thrombopoiesis with occasional megakaryocytes M:E -2:1 suggestive of Hypoplastic Bone Marrow with no signs of neoplastic pathology or hemoparasites.

I/v/o hypocellular bonemarrow cytogenetics studies were done to confirm the diagnosis which shows Chromosome Breakage sensitivity to mitomycin c.

On discharge hb was 12.1g/dl hct 26.4% platlet 2.24L/ul.

## 3. DISCUSSION

Fanconi anaemia is a rare genetic condition with annual incidence of 1 in 129000 live births with male to femaleratio1:1and median age of onset is 7 yrs and average life expectancy 25yrs.Mostly patients present with moderate anemia and bleeding tendencies (9,10). TGF-β1 is an important T helper 3 (TH3) immune suppressor cytokine that promotes B cell and T helper cell interaction [1]. TGF-b1 has been shown to selectively inhibit the growth and differentiation of early HSCs, contributing to marrow failure in Fanconi anemia [2]. IL-10 is an immune-regulatory cytokine having anti-tumor effect [3]. Fanconi anemia patients presenting severe cytopenia had elevated IL-10 levels [4]. Toll like receptors play a considerable role in the host defense against microorganism [5]. TLR-dependent overproduction of TNFα is an important element in the pathogenesis of BM failure of Fanconi anemia [6]. The higher expression of PD-L1 was associated with poor response to induction therapy in AML patients [7]. There is a high mutational load in Fanconi anemia - acute myeloid leukemia (FA-AML) cell lines, which also express PD-L1 [8].

In our case patient presented in shock with CHF. He had severe anemia with hypoplastic thumb with short stature underdeveloped penis and abnormal pigmentation. Bone marrow biopsy was markedly hypocellular and chromosome breakage studies were positive after exposure to mitomycin C [9-11].

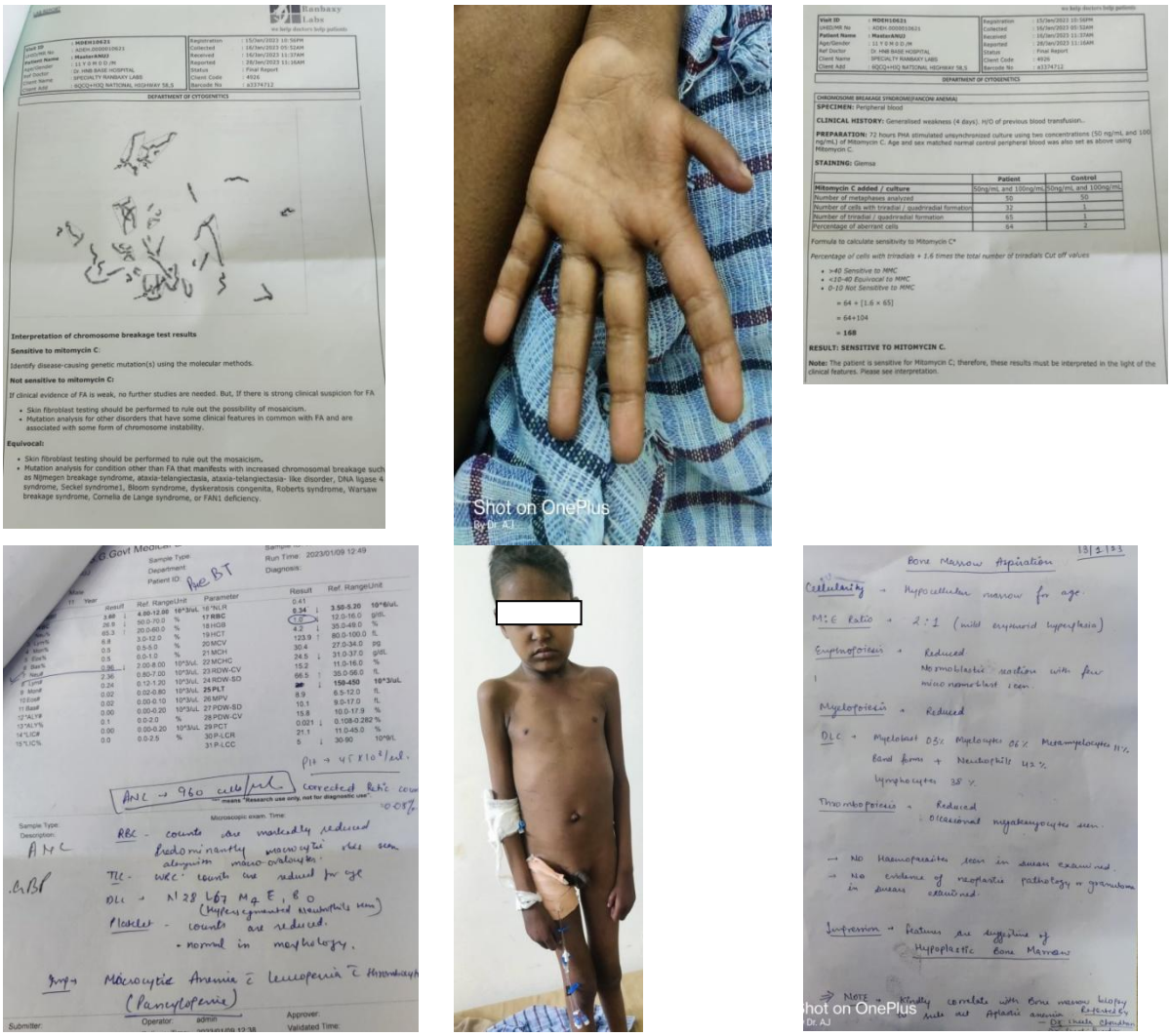


**Fig. 1. Disorder in patients and clinical reports**

## 4. CONCLUSION

It is concluded that although it was rare presentation, the possibility of such association should be kept in mind while dealing the patient. Echocardiography and CVS examination should be done to rule out cardiac involvement. It will help us to plan appropriate treatment and follow up.

## CONSENT

As per international standard or university standard, parental(s) written consent has been collected and preserved by the author(s).

## ETHICAL APPROVAL

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

## COMPETING INTERESTS

Authors have declared that no competing interests exist.

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