Case study

An usual case of congestive cardiac failure

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,shortstature,congestive cardiac failure

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 –microcytopenia.

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,gastrointestinal or renal anomalies can also occur.FA “facies” microcephaly,small eyes,epicanthal folds,abnormal ears.

CASE HISTORY

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.

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.

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.

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.

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. 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 mitomycinC.

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 followup.

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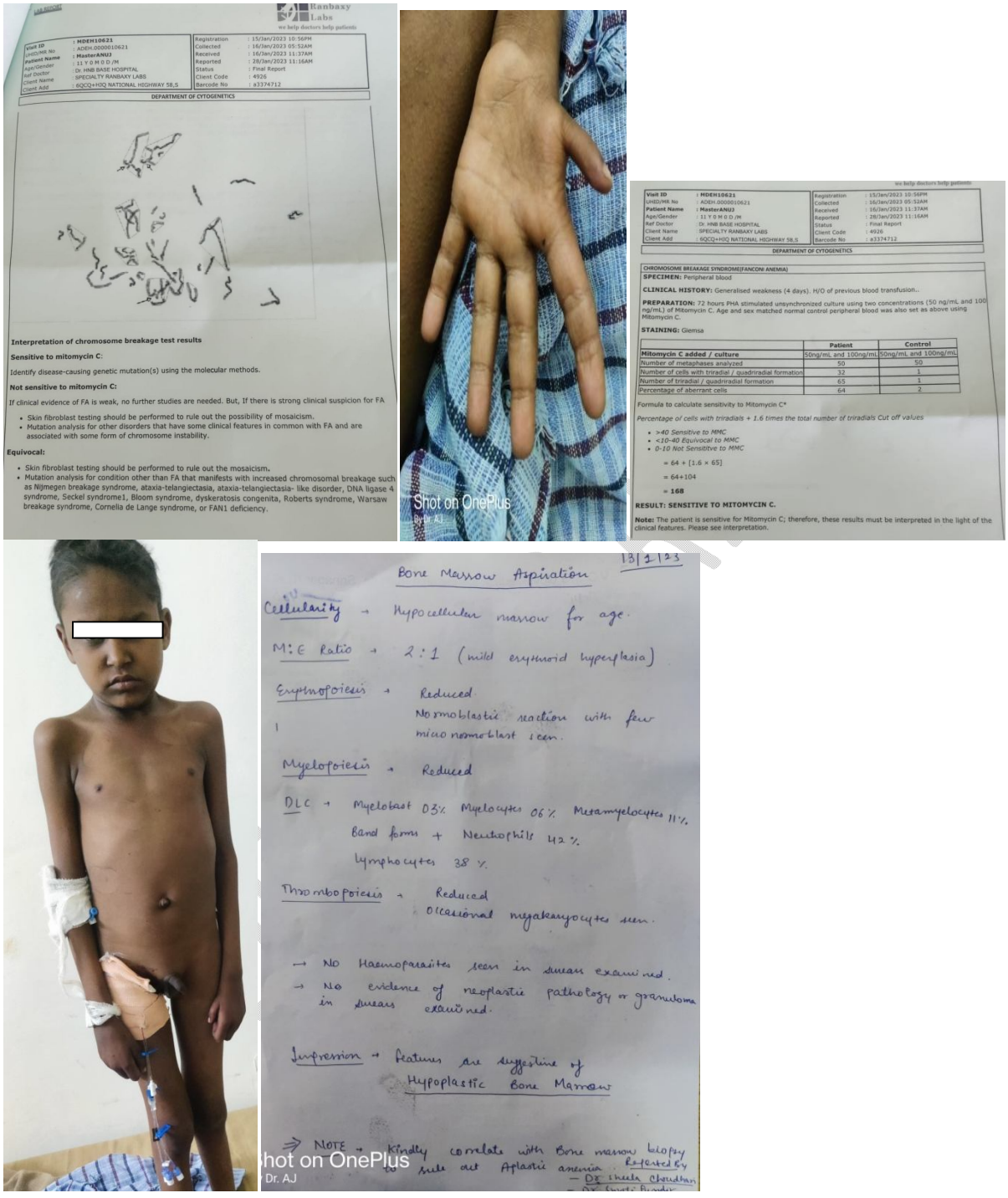

Fig. 1. Disorder in patients and clinical reports