

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 appetite 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 appetite 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 tapered 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.

REFERENCE

Indian general of paediatrics,Ital J Pediatr.2015,41:38.

Nelson Textbook of pediatrics .chap 1605.e1.priciples and clinical indication of hematopoietic stem cell transplantation..

Deaconu A,voda D,Bulucea D.Fanconi Anemia-case report of rare aplastic anemia at child.Acta Medica Marisiensis,2014 Jun 1,60(3):125-8.

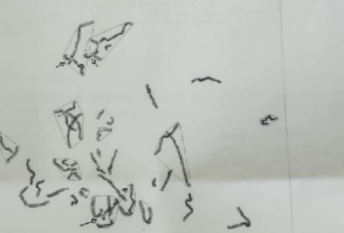
Goswami M, Bhushan U.Dental prespective of rare disease of Fanconi anemia case report with review.Clinical Medicine Insight Casereport.2016 Sep9:25-30

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Client ID: MDEH1021
 UNDO# No: ADEH-000010621
 Patient Name: MASTHANUJ
 Age/Gender: 11 Y 9 M 0 D JM
 Ref Doctor: DR. HNB BASE HOSPITAL
 Client Name: SPECIALTY RANBAXY LABS
 Client Add: SCCC+HIQ NATIONAL HIGHWAY 58.5

Registration: 15/06/2023 10:56PM
 Collected: 16/06/2023 09:52AM
 Received: 16/06/2023 11:37AM
 Reported: 16/06/2023 11:16AM
 Status: Final Report
 Client Code: 4926
 Barcode No: 83374712

DEPARTMENT OF CYTOGENETICS



Interpretation of chromosome breakage test results
Sensitive to mitomycin C:
 Identify disease-causing genetic mutation(s) using the molecular methods.

Not sensitive to mitomycin C:
 If clinical evidence of FA is weak, no further studies are needed. But, if there is strong clinical suspicion for FA

- Skin fibroblast testing should be performed to rule out the possibility of mosaicism.
- Mutation analysis for other disorders that have some clinical features in common with FA and are associated with some form of chromosome instability.

Equivocal:

- Skin fibroblast testing should be performed to rule out the mosaicism.
- Mutation analysis for condition other than FA that manifests with increased chromosomal breakage such as Nijmegen breakage syndrome, ataxia-telangiectasia-like disorder, DNA ligase 4 syndrome, Seckel syndrome, Bloom syndrome, dyskeratosis congenita, Roberts syndrome, Warsaw breakage syndrome, Cornelia de Lange syndrome, or FANL1 deficiency.



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CHROMOSOME BREAKAGE SYNDROME (FANCONI ANEMIA)
SPECIMEN: Peripheral blood

CLINICAL HISTORY: Generalised weakness (4 days), H/O of previous blood transfusion...

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

Mitomycin C added / culture	Patient	Control
50ng/mL and 100ng/mL	50	50
Number of metaphases analyzed	50	50
Number of cells with triradial / quadriradial formation	32	1
Number of triradial / quadriradial formation	65	1
Percentage of aberrant cells	64	2

Formula to calculate sensitivity to Mitomycin C*
 Percentage of cells with triradials + 1.6 times the total number of triradials Cut off values

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

= 64 + [1.6 x 65]
 = 64 + 104
 = 168

RESULT: SENSITIVE TO MITOMYCIN C.

Note: The patient is sensitive to Mitomycin C; therefore, these results must be interpreted in the light of the clinical features. Please see interpretation.



Bone Marrow Aspiration 18/1/23

Cellularity → Hypocellular marrow for age.

M:E Ratio → 2:1 (mild erythroid hyperplasia)

Erythropoiesis → Reduced
 Normoblastic reaction with few micro-normoblast seen.

Myelopoiesis → Reduced

DLC → Myeloblast 03%, Myelocytes 06%, Metamyelocytes 11%,
 Band forms + Neutrophils 42%,
 Lymphocytes 38%.

Thrombopoiesis → Reduced
 Occasional megakaryocytes seen.

→ NO Haemoparasites seen in smears examined.
 → No evidence of neoplastic pathology or granuloma in smears examined.

Impression → Features are suggestive of Hypoplastic Bone Marrow

NOTE → Kindly correlate with Bone marrow biopsy
 rule out Aplastic anemia. Referated by
 Dr. Sneha Choudhary
 Dr. Anurag Prasad

hot on OnePlus
Dr. AJ

Fig. 1. Disorder in patients and clinical reports