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# Fanconi's Anemia - Case Report

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**SUMMARY**

*Fanconi's Anemia is an inherited condition characterized by pancytopenia combined with certain congenital physical anomalies. One of our cases, an 8-year old girl, presented with pallor. Investigations showed pancytopenia and hypocellular bone marrow. She was treated with blood transfusions, steroids and androgens.*

## INTRODUCTION

Fanconi's Anemia, a constitutional pancytopenia, is inherited as an autosomal recessive manner with a heterozygote frequency of about 1 in 2001. As described first by Fanconi, patients have pancytopenia combined with physical anomalies like skin pigmentary changes, short statue, upper limb abnormalities, males often have gonadal and genitalia abnormalities including underdeveloped or micropenis; undescended, atrophic or absent testes, hypospadias, phimosis or an abnormal urethra, renal malformations and eye, ear and gastrointestinal/cardiopulmonary malformations[2]. Differential diagnosis is acquired aplastic anemia, Schwachman-Diamond syndrome, dyskeratosis congenita, amegakaryocytic thrombocytopenia and TAR (Thrombocytopenia with absent radii syndrome.

## CASE PRESENTATION

An 8 years old girl presented with progressive pallor for 6 years. There was also history of bruises over body and fever off and on. She was born of consanguinous marriage and had normal birth and developmental history.

On examination she was short statured with a height of 112 cm (<5th centile), thin built (weight=18 kg; <5th centile). She had marked pallor and was also microcephalic. She had circumoral hyperpigmentation and dysmorphic thumbs of both hands. Her vital signs were normal and systemic examination was unremarkable.

Initial investigations showed Haemoglobin 5.3 g/dl, TLC $1.6x10^9/l$ polymorphs 40% and lymphocytes 60%, platelets $56x10^9/l$. Peripheral smear showed a mixed population of cells. BUN, serum creatinine, serum bilirubin, ALT, AST and alkaline phosphate were normal. Bone marrow trephine biopsy was done and it showed hypocellular marrow.

Urine complete examination was normal. Urine culture and blood culture showed no growth. During stay in the hospital she was given whole blood transfusion, broad-spectrum antibiotics (Inj. Cefotaxime 100mg/kg/day I/V), oral prednisolone 5mg on alternate days. Androgens in the form of injection Nandrolone (Decadurabolin) 1mg/kg/week I/M were also started. Her symptoms and peripheral blood picture improved with this treatment and she was discharged on steroids and androgens. On follow up visits she had symptomatic improvement. Her liver function tests and blood counts were checked repeatedly and were found to be normal.

Another case is of a 12-year old girl who presented with pallor for the last 7 years. She also had bifid thumbs. Investigations showed pancytopenia and hypocellular marrow. She was given steroids and androgens. On follow-up, her clinical condition improved but she required multiple transfusions, her liver functions were deranged and was found to be anti-HCV positive.

She was followed up on regular basis for anemia, petechiac, bruises and side effects of drugs. She was transfused accordingly and her symptoms improved.

## DISCUSSION

About 75% of patients of Fanconi's Anemia are between 3 and 14 years of age at the time of diagnosis[1]. The presence of one or more characteristic congenital physical anomalies combined with pancytopenia favour the diagnosis of Fanconi's Anemia. Thrombocytopenia usually develops initially with subsequent onset of granulocytopenia and then anaemia. The red blood cells are macrocytic and there is increased fetal hemoglobin.

As the disease progresses, the marrow becomes hypocellular and fatty. A major finding is abnormal chromosomal fragility and the karyotype shows spontaneously occurring chromatid breaks, rearrangements, gaps, end reduplications and chromatid exchanges in cells[1]. A prenatal diagnosis can be made by abnormal chromosomal picture.Diagnostic testing can be performed on fetal amniotic fluid cells at 16 wks of gestation or on chorionic villus biopsy specimen at 9-12 wks.

Overall incidence of malignant transformation is 20%.The defect in DNA repair and the cellular damage that occur in these patients predispose them to malignancy like acute myeloid leukemia and liver tumours[2,3].

Bone marrow transplantation is the only curative therapy. Androgens and steroids are also used. The response rate with andogren therapy is 50%[2]. Other recent advances in the management are cord blood cells transplantation, use of hematopoietic growth factors and gene therapy.

## REFERENCES

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