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Fanconi Anemia: In Newborn-unusual presentation

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Abstract

Fanconi anemia (FA) is a rare autosomal recessive disorder associated with pancytopenia, congenital anomalies and chromosomal instability. Few cases have been identified in the first month of life, and few have described the management of FA in resource-limited settings. In this case report, we describe a newborn from Ethiopia with cardiac anomalies, left-sided skeletal abnormalities, absent left kidney, and thrombocytopenia strongly believed to have FA.

Keywords

fanconi anemia; genetics; newborn; cardiac anomalies

Introduction

Fanconi anemia (FA) is a rare, genetically heterogeneous autosomal recessive disorder characterized by congenital malformations, hematological problems and predisposition to malignancies [1]. Patients may be severely affected with multiple congenital malformations or may have a mild phenotype with no malformations. Affected individuals may exhibit varying degrees of low birth weight, growth retardation, skin pigmentation, gastrointestinal malformations, hypoplasia or aplasia of the radius and thumbs, skeletal, renal, cardiac and other anomalies. Hematologic abnormality at birth is very rare [2]. To the authors' knowledge, no case has been reported from Ethiopia. Although chromosomal breakage test for the diagnosis of FA is recommended, we were unable to perform this test due to limitations in our hospital's laboratory.

Case Presentation

A 14 days old female child from northern Gondar Amhara regional state, born to a 27-year-old Para2, abortion zero, mother at term. The pregnancy was uneventful, mother received only iron sulfate during ANC follow up but ultra sound was not done. There is no history of consanguinity or physical malformation in other family members. The mother has no history of radiation exposure, use of traditional medicine or chemotherapy during pregnancy.

Physical examination: her vital signs were normal (PR=142, RR=42, T=36.5^{0}C). Weight-3000gm length-48cm and Head circumference-34cm, all anthropometric measurements are within the standard range, she has oral thrush and a grade III systolic murmur best heard at the left lower sternal border. No dysmorphisms or abnormal skin pigmentation were appreciated. The left hand was hypoplastic with absent left thumb and index finger and deformity of the wrist joint. The left lower extremity had preaxial

polydactyly with 7 toes and severe deformity at the ankle joint (Figure 1). Skeletal radiographs revealed mesomelic deformity of the left upper extremity with absent radius, thumb and index finger (Figure 2). A mesomelic deformity of the left lower extremity with absent tibia was also observed. Echocardiography revealed a 5mm perimembraneous ventricular septal defect, a 2mm secundum-type atrial septal defect, and mild pulmonary stenosis with left to right blood flow. Abdominal ultrasound revealed absent left kidney. The liver, spleen, and right kidney were normal. At presentation, her complete blood count was normal except for platelet count of 93,000 cells/ml. The family declined a bone marrow aspirate. Our facility does not have the capacity to perform genetic study to diagnosis FA.

Discussion

This report describes a neonate with skeletal malformations, renal anomalies, cardiac defects, and hematologic derangements consistent with FA. Although chromosomal breakage test is recommended for the diagnosis of FA, we were unable to perform this test due to limitations in our hospital's laboratory.

Relatively little is known about FA in neonates. A report from India described a term, growth-restricted neonate with right radial ray defect, absent left kidney, mild thrombocytopenia and herniated sac of the left lumbar region [3]. The finding in our case is similar to that of Indian case except for cardiac anomaly.

Radial ray defects also occur in Holt-Oram syndrome, thrombocytopenia-absent radius syndrome (TAR), and vertebral, anorectal, cardiac, tracheal, esophageal, renal, and radial limb sequence (VACTERL). TAR presents at birth with severe thrombocytopenia and radial ray defects, but the thumb is typically present, although some kidney malformations were described; absent kidney was not identified in some literatures. Heart anomaly like VSD, ASD were also reported [4]. Holt-Oram syndrome always presents with radial ray and cardiac defects but is not associated with renal and lower extremity abnormalities [5]. Even though in Noonan syndrome; congenital heart disease, characteristic facial dysmorphism, hydronephrosis, bleeding tendency with thrombocytopenia, joint deformity and skin pigmentation are well documented in different literatures, very few of these characters are described in our case [6].

Other than the congenital anomalies seen in FA, it is also associated with a gradual bone marrow failure and propensity for malignancy. The resources available for the management of FA in our setup are limited, the surgical option for this patient was communicated to orthopedic surgeon in Addis Ababa, who recommended waiting till the age of one year for possible correction of extremities, and expertise on hematology, cardiac disease and endocrinologist suggested strict follow up of the child with a serial hematologic workup; (including blood counts to be done every 1-3 months to look for thrombocytopenia and anemia, bone marrow examination to be done annually for leukemia and myelodysplastic syndrome surveillance, and patients to be assessed annually for solid tumors), echocardiogram, lipid profile, thyroid function test and diabetic screening. In addition to these, parents were counseled about the course of the disease and locally available treatment options. Genetic counseling was not possible in our setup.

Conclusion

Diagnosis of FA in a resource constrains setting requires high index of suspicion, early diagnosis of

the cases will help health professionals to carryout appropriate steps of management and follow up. As the disease involves multiple organ and affects body physiology a multidisciplinary management approach is required to improve the quality of life. We recommend further research to determine the optimal management including the option of bone marrow transplantation in resource limited settings.

Figures

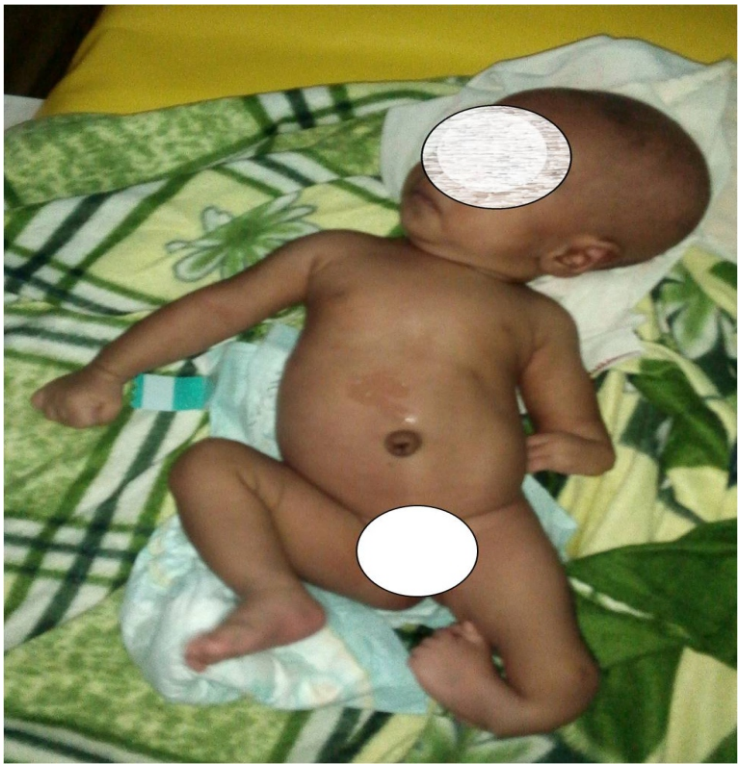

Figure 1: Skeletal malformation on the left side

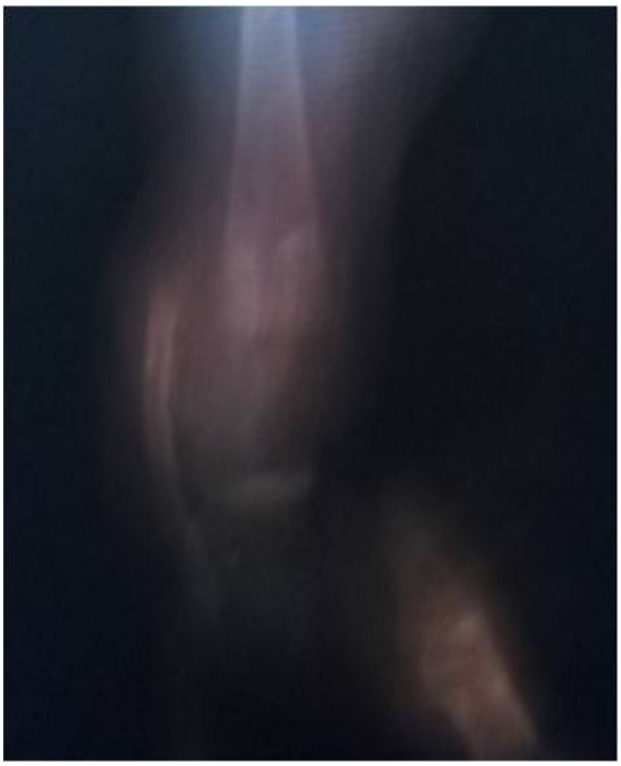

Figure 2: absent radial bone of left upper extremity

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