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Research Paper

Fanconi's Anaemia in Newborn – Rare Presentation

Medical Science

KEYWORDS : Fanconi's anaemia, thrombocytopenia, physical malformations

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ABSTRACT

Fanconi's anaemia is a rare autosomal recessive disorder associated with pancytopenia, congenital anomalies and chromosomal instability. Identification of the disease at birth is based on charecteristics physical malformations, as hematological menifestations at birth is very rare. Only very few cases were identified in the first month of life and reported in the world literature. We report a case of fanconi's anaemia in a newborn. Who presented with right radial ray defect, absent left kidney, left lumber region herniated sac and mild thrombocytopenia.

Introduction:

First description of Fanconi's anemia dates back to 1927.[1] Since then more than 1000 cases of Fanconi's anemia have been reported in the world literature.[2] However of these only very few cases were identified in the first month of life.Fanconi anemia is one of the well known chromosome instability syndromes. 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, gastrointestinal malformations, hypoplasia or aplasia of the radius and thumbs, skeletal, renal and other anomalies.Hematologic abnormality at birth is very rare.[4] We report a case of a newborn with varying degree of physical malformations.

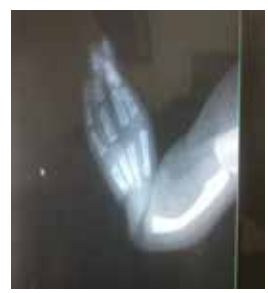
Case report:

A 37 week, 1900 grams, growth restricted male neonate, born to a first gravida mother, with uneventful antenatal and intrapartum period, was admitted to our NICU on day one of life. There was no history of consanguinity or physical malformation in other family members.

Examination of the newborn revealed microcephaly, high arched palate, depressed nasal bridge, low set ears, webbed neck, micrognathia, widely spaced nipples, radially curved right forearm and absent right thumb, left ape thumb, Left lumber region herniated sac, hypoplasia of thenar muscle, No café au lait spots were noted.

Skeletal roentogenic survey revealed right absent radius, curved ulna and abdominal & KUB sonography revealed herniated bowel loops in left lumber region and absent left kidney. Hematologic profile revealed significant thrombocytopenia - Platelet count 92,000/cmm on day two of life. Hemoglobin, red blood cell morphology and white blood cell count were within normal limits. Mother's platelet count was normal.

In view of poor long-term prognosis, parents refused any further treatment and baby was discharged against medical advice on 6th day of life.



Discussion:

Diagnosis of Fanconi's anemia (FA) is Microcephaly, short webbed neck and skeletal anomalies, mild thrombocytopenia noted in the present case, though rare, have been reported in patients with TAR syndrome.[2,3] In FA, if the radii are affected, the thumbs are always abnormal (absent / hypoplastic); in TAR, in which radii are absent, the thumbs are always present.[2] The present case had an absent right thumb. TAR syndrome presents at birth with severe thrombocytopenia with bleeding manifestations and radial ray defects.[5] Holt-Oram syndrome has radial ray defects with cardiac defects (100% cases)In the present case there were no cardiac defects.[5]

Trisomy 18 can rarely have radialray defects and eye anomalies. However trisomy 18 has host of different physical abnormalities and a typical facies.[2] FA has considerable overlap in the physical abnormalities with VATER / VACTERAL syndromes. In a large series of FA patients, 10% patients had three principal clinical features and additional 20% patients had two major defects found in VATER syndrome.[6] But in VACTERAL association renal and radial ray defect present on same side while in FA radial ray defect and renal malformations are opposite side.

Conclusion:

Early diagnosis also offers options of planning next pregnancy; as the umbilical cord blood can be used for stem cell transplantation. Bone marrow or umbilical cord blood transplantation from an HLA identical sibling is now considered the treatment of choice for FA.[3,7] Diagnosis of FA requires high index of suspicion as it presents with physical abnormalities involving multiple systems and hematologic abnormalities at birth are extremely rare. Early diagnosis in FA is very important as long term survival depends on the age of onset of hematologic abnormalities or malignancies.[8]

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