FANCONI FAMILIAL HYPOPLASTIC ANAEMIA

Pages with reference to book, From 88 To 89

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Introduction

Fanconi Hypoplastic Anaemia affects all three elements of the bone marrow. The disease is inherited on an autosomal recessive basis and a familial incidence is usual (Fanconi, 1967). The most common abnormalities associated with the disease are short stature, microcephaly, microphthalmos and hypoplastic or accessory thumb.
This report describes a female patient with the same syndrome having some interesting skeletal anomalies.

Case Report

S.K. a 4 years old female child, was born after an uneventful full term pregnancy. There was no family history of consanguinity. She did not have any problem till the end of 2nd year. During the next two years she got repeated attacks of epistaxis and respiratory infections. The child had not taken any antibiotic or other drugs which could cause bone marrow depression. The family history is negative for skeletal or definite hematologic diseases. She had two normal sisters and one brother, all older to her. Examination during the initial visit to Khyber Hospital Peshawar revealed a well developed female child as regards height and weight. She had an accessory rudimenbary thumb on the right side. The neck was webbed and there was sprengel deformity of the scapula on the right side. No abnormal skin pigmentation was found. There was a Grade II apical systolic murmur. The hematological studies showed a Hemoglobin at 7.0 gm/dl, hematocrit 20%, WBC 2500/cmm, and a relative lymphocytosis. Platelet count was 80,000/cmm and no reticulocytes were seen. The red blood cells were normocytic and normochromic. Bone marrow was hypocellular and all elements were equally depressed. In addition reticulum cells and plasma cells were also found.
Roentgenogram of the skull, cervical spine and chest were normal except for synostosis between C3 and C4 vertebrae. Electrocardiogram was normal and coupled with X-ray and on auscultatory findings it was concluded that she had a functional hemic murmur.
Chromosome studies could not be done because of lack of facilities.
A diagnosis of Fanconis pancytopenia, based on skeletal anomalies and hematological findings was thus made and the child put on prednisolone and oxymethalone. A remission was obtained for a period of three months. Subsequently the child died of intracranial haemorrhage and associated bronchopneumonia.

Discussion

Syndromes which include hypoplasia of the bone marrow and skeletal anomalies include Fanconis pancytopenia and Thrombocytopenia with radial aplasia. In 1969 Aaes and Smith (1969) reported male siblings with congenital hypoplastic anaemia and triphalangeal thumb and distinguished their disorder as a separate entity from the other two, thus adding a new member to the list of these syndromes. Skeletal anomalies observed in Fanconis pancytopenia include hypoplasia or aplasia of thumb, radius and carpal bones, or rarely accessory thumb. Our patient had an accessory thumb and also webbed neck and sprengel deformity of scapula. The last two have not been described previously in literature. The hematologic manifestations of Fanconis pancytopenia may have its onset in infancy but it generally

occurs later, and so was in our case as she got symptoms at the age of two years. Thrombocytopenia, leukopenia and anaemia are all apparent in fanconis pancytopenia. Our patient had pancytopenia. She had to receive four transfusions over a period of three months. She had frequent infections and episodes of epistaxis.

Microcephaly is a finding' in 40% cases of Fanconis pancytopenia and distinguishes it from the other two syndromes in which it is not a feature. Our patient had microcephaly, however she was of normal intelligence.

Chromosomal studies have proved to be of some help. In some cases high number of chromatid breaks, endoreduplications and other minor abnormalities have been observed (Bloom et al., 1966).

The overall prognosis is gloomy. Remission can be obtained with prednisolone and oxymethalone but it is usually not a prolonged one. Repeated blood transfusions can prolong life but death is common in early childhood.

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References

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