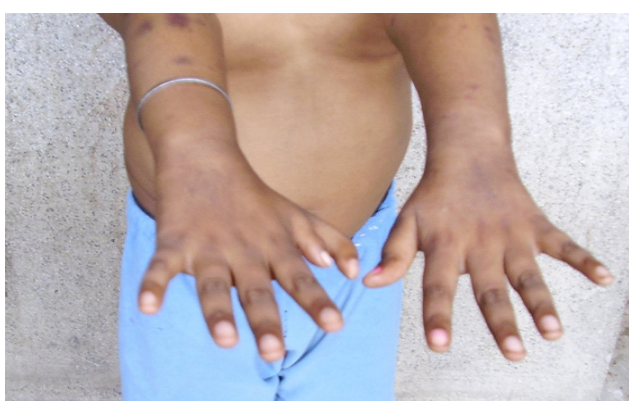
SPOT DIAGNOSIS (IMAGE GALLERY)



APLASTIC ANEMIA

Rakesh Kumar, Puja Bhagat
Department of Pediatrics, Katihar Medical College, Katihar, Bihar, India.

Address for Correspondence: Dr. Rakesh Kumar, Assistant Professor, Department of Pediatrics, Katihar Medical College, Katihar, Bihar, India. E-mail: drjaiswalrakesh@yahoo.co.in

A 6-years old male child, born of non-consanguineous parents was admitted with complains of paleness of the body and bleeding from gums since one year. He had been treated with multiple blood transfusions. Bone marrow examination done outside showed hypocellular marrow with depression of all the three cell lines. The diagnosis of referring physician was aplastic anemia. General examination of the body revealed marked pallor, multiple ecchymotic patches and polydactyly of the right hand. There was a supernumerary triphalangeal thumb on right hand. (Fig.1) He also had short stature, facial hyperpigmentation, microcephaly and undescended testes of the right side. The child had mild mental retardation. History of delayed achievement of milestones was also present.

What is the diagnosis?

Fanconi Anemia (FA) is a rare autosomal recessive disorder. FA was first described by Professor Fanconi in Switzerland in the year 1927. (1) In this disorder, at least seven genes are implicated encoding proteins which form complex possibly involved in sensing DNA damage and facilitating repair. (2) Usual age of detection is about 7 years but it may be apparent at birth or the disease may not be diagnosed until third or fourth decade of life. (3) Affected male to female ratio is 1.3:1. Every cell and every organ of the FA patient may be missing the contribution of an essential gene function. Consequently physical anomalies may involve almost all organ systems. The various manifestations of FA include pancytopenia, single lineage defects (particularly thrombocytopenia especially in neonates) (4) or malignancy (like leukemia, myelodysplastic syndromes or solid tumors especially in those over the age of 20). (2,5) Only half of the patients have classic phenotype that includes short stature, microcephaly, radial ray defects (hypoplasticperbsent thumb and radii, supernumerary, bifid or triphalangeal thumb), café au lait and hypopigmented spots, and a characteristic facial appearance (a broadbase nose, epicanthal folds and micrognathia). (1) Approximately one-third of the patients have

genitourinary abnormalities. Mental retardation especially learning disabilities may be present in some cases. (1) Diagnosis is based on Mitomycin C or Diepoxybutane induced chromosomal breaks and rearrangements in cultured lymphocytes. (3,5) The only curative therapy to date has been bone marrow transplantation. High dose androgen therapy alone or in combination with steroids produces haematologic improvement in more than two third of the patients. Once an index case is diagnosed, genetic counseling of the parents is important. The prenatal diagnosis of the disease could be done using the same chromosomal breakage study. The prognosis of FA is still poor.

References:

1. Glanz A, Fraser FC. Spectrum of anomalies in Fanconi's Anemia. J Med Genet 1982; 19: 412-416.
2. Rosenberg PS, Greene MH, Alter BP. Cancer incidence in persons with Fanconi anemia. Blood. 2003; 101: 822-826.
3. Zatterale A, Calzone R, Renda S, Catalano L, Selleri C, Notaro R, Rotoli B. Identification and treatment of late onset Fanconi's anemia. Hematologica. 1995; 80: 535-538.
4. Parikh TB, Udani RH, Nanavati RN, Rao B. Fanconi's Anemia in newborn. Indian Pediatr. 2005; 42: 285-287.
5. Tischkowitz MD, Chisholm J, Gaze M, Michalski A, Rosser EM. Medulloblastoma as a first presentation of Fanconi's anemia. J Pediatr Hematol Oncol 2004; 26: 52-55.

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