CASE REPORTS

FANCONI'S ANEMIA*

Report of 3 Cases

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Fanconi in 1927 for the first time described a “familial infantile pernicious like anemia” in three sibs, who presented with pancytopenia, bone-marrow hypoplasia, multiple congenital anomalies, hyperpigmentation of skin, hypogonadism, strabismus and growth retardation. Since then the entity has been recognised as Fanconi’s anemia or Fanconi’s syndrome and about 165 cases have been recorded in the world literature (Reinhold et al. 1952, Fanconi 1964 and 1965, Bryan and Nixon 1965, London et al. 1965 and Wintrobe, 1967). Rare occurrence of the entity and scanty reports from India (Joshi et al. 1963, Paul et al. 1966 and Athavale, 1968) prompted us to record our observations on three children with Fanconi’s anemia.

Material

Case 1. A.K., a 7½-year-old boy was admitted to the pediatric service of the All India Institute of Medical Sciences hospital in March, 1965 with the presenting symptoms of low grade continuous pyrexia ranging from 99°-100°F, progressive pallor and pigmentation of the skin of six months’ duration. The past, personal and family survey were non-contributory. There was no evidence of consanguinity among his parents.

He was underbuilt and undernourished (weight 15 Kg., height 104.5 cm.). He looked pale and had patches of pigmentation on the hard palate, buccal mucosa, and tongue along with circumoral pigmentation. He had slanting eyes, strabismus and microphthalmia. The head circumference was 46.5 cm. He had an accessory finger on the radial side of the right thumb. The liver was palpable 1 cm. below the costal margin in the right hypocondrium in the mid-clavicular line. It was soft, nontender, had a smooth surface and round margin. The spleen and lymph nodes were not palpable.

Systemic examination revealed cardiomegaly and a hemic ejection systolic murmur grade II/VI, and hypogonadism. A clinical diagnosis of Fanconi’s anemia was made.

Investigations revealed a hemoglobin concentration of 2.2 G%, packed cell volume of 9%, total red cell
count of 1.1 million/cu.mm., and a reticulocyte count of 0.5%. The total leucocyte count was 2750 cells/cu.mm., with 5% polymorphs, 90% lymphocytes, 4% myelocytes and 1% metamyelocytes. Platelet count was 30,000/cu.mm. The erythrocytes showed aniso-poikilocytosis and moderate hypochromia, a few micro- and macrocytes were also present. A large number of broken forms, burr cells, and other atypical forms were seen. No immature cells of the erythrocyte series were seen. Granulocytes were markedly reduced and lymphocytes were the predominating cells. Occasional immature cells of the leucocyte series were seen. Platelets were markedly reduced in number. The bone marrow was extremely hypocellular. Among the cells present the erythrocyte precursors were predominant with a shift to the right. The cells were mostly intermediate and late normoblasts. The hemoglobinisation was adequate. Only mild myelopoiesis was seen. Myelocytes and metamyelocytes were predominant. No megakaryocytes were seen. Bone marrow ironstaining was 4+. His serum iron was 272 ug/l with an unsaturated iron binding capacity of 316 ug/l and 46% saturation of transferrin with iron. His serum vitamin B₁₂ was 160 ug/ml. Serum bilirubin was 1.1 mg%. Foetal hemo
globin content was 2.5%.

A skiagram of the chest showed cardiomegaly and bony changes seen in chronic anemia. A skiagram of the spines revealed spina bifida occulta of the first sacral vertebra. On intravenous pyelography the left kidney and ureter appeared normal in shape, size and position. The right kidney was small and displaced medially. The urine was normal. His intelligence quotient was 74-76.

He was given a packed cell transfusion and put on prednisolone 15 mg per day and testosterone 50 mg subcutaneously every two weeks. His general condition improved. There was no improvement in his hematological status except for the rise in hemoglobin. He was discharged from the hospital in May, 1965 on prednisolone and testosterone, but he did not return for follow up.

Case 2. A.S., a 14-year-old boy was admitted in September, 1967, with the presenting symptoms of progressive pallor, exertional dyspnoea, palpitation and bleeding from gums, nose and gastrointestinal tract off and on for five years. In February, 1967, he had a bout of hematemesis and was given eleven blood transfusions in Ludhiana. Following this episode he remained all right for about three weeks when again he became pale and received 2 more blood transfusions. Investigations done at that time in the Dayanand Medical College, Ludhiana, revealed pancytopenia and hypocellular bone marrow. He was put on prednisolone and testosterone as he was considered to be a case of aplastic anemia. He developed megaloblastic bone marrow reaction while on prednisolone and testosterone and was given both folic acid and vitamin B₁₂ injections. He had smallpox preceeding the present illness. His younger brother had died at the age of 14 years due to a similar illness and a younger sister had a similar illness (vide infra). The past and personal histories were non-contributory. There was no evidence of consanguinity.

He was underbuilt and undernourished. His measurements were: weight 25 Kg., height 143 cm., upper segment