Cutaneous Manifestations of Fanconi’s Anemia in Two Siblings

Sir,
Fanconi’s anaemia is a rare, autosomal recessive disorder characterized by diverse congenital malformations, developmental defects, progressive pancytopenia, predisposition to both hematologic malignancies and solid tumors, and widespread mottled skin pigmentation.

Two brothers (aged 7 and 13 years) born of nonconsanguinous marriage and studying in second and sixth standard presently, presented to our pediatrics department 2 years back with the complaints of fever, not gaining weight and height, paleness of body, as well as an additional complaint of incomplete voiding of urine in the younger brother. On anthropometric examination, the weight of both the brothers was less than the 3rd centile. The height of the elder brother was at the 3rd centile and younger brother was less than the 3rd centile. The head, chest, and mid arm circumference was 48 cm, 49 cm, and 14.5 cm, respectively, for the elder brother and 47.5 cm, 46 cm, and 12 cm, respectively, for the younger brother. Microcephaly and micrognathia was present in the younger brother. On hematological evaluations, there was progressive pancytopenia. Peripheral blood picture showed moderate anisopoikilocytois with microcytic hypochromic anemia in both the brothers. Bone marrow aspirate examination revealed hypoplastic anemia. Liver function, renal function tests, and X-rays of the hands were normal. On ultrasonography of the abdomen, there was hepatomegaly, and cystitis with a large echogenic calculus of size 1.2 cm in the dependent part of urinary bladder in the elder and younger brother, respectively. Karyotyping done from a cytogenetics lab, Department of Zoology was normal. The patients were referred to the dermatology outpatient department for the cutaneous manifestations. On general examination, there was pallor, generalized olive brown pigmentation of the whole body, with discrete, asymptomatic, hypopigmented macules distributed over the trunk [Figures 1 and 2], neck, bilateral upper limbs, lower limbs, face, palms [Figure 3], and soles in both the brothers. No cafe-au-lait macules, palmoplantar keratoderma, and mucosal pigmentation was present. Fingers, thumb and nails, and testes were normal. No hearing loss, structural abnormalities of external ears, and squint were found. Fundoscopy, superficial, and deep reflexes were normal. Histopathological examination of the skin revealed sparse superficial perivascular lymphocytic infiltrate with numerous
Fanconi anemia is a genetically and phenotypically heterogeneous recessive disorder. There is an intrafamilial phenotypic variation in the specific types of congenital malformations among affected siblings. Other findings include skin pigmentation abnormalities, hypoplastic thenar eminence, microcephaly, and/or microphthalmia.

Renal aplasia, horseshoe kidney, and syndactyly have also been reported. However, in our case, only cystitis and large echogenic calculus of size 1.2 cm was present in the dependent part of the urinary bladder, in the younger brother.

In our patients, mottled pigmentation all over the body, short stature with height and weight below the 3rd centile, microcephaly in the younger brother, progressive pancytopenia with hypoplastic anaemia on bone marrow aspiration, led to the diagnosis of Fanconi’s anemia.

The patients were given blood transfusions and anabolic steroids (stanozolol 2 mg/kg/day initially which is still continuing). They are on continuous follow up. There is weight gain, increase in height, and improvement in blood counts.
**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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