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Congenital Erythropoietic Porphyria

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Photosensitivity reactions associated with blister formation are rare in Indian children. We report one of the causes of this condition, namely congenital erythropoietic porphyria.

Case Report

A 7-year-old boy was seen with the history of recurrent episodes of blisters over exposed parts of the body since the age of three and a half years. The blisters healed within two to three days with scarring and disfigurement. He had two to three episodes a month. There were no other systemic symptoms. He was the second child born to

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a second degree consanguineous marriage. There was no history of a similar illness in the family.

On examination the patient was alert and intelligent. He had mild pallor and weighed 16.3 kg. The exposed parts of the body were hyperpigmented with areas of hypopigmentation and extensive scarring. Hypertrichosis was also prominent over the exposed areas. There was mutilation and contractures of the fingers and toes (*Fig. 1*). The teeth were normal. Spleen was palpable 1 cm below the costal margin. The rest of the systemic examination was within normal limits.

The level of hemoglobin was 9.4 g/dl, reticulocyte count 2.5%, total leucocyte count 7,600/cu mm, with 49% neutrophils, 45% lymphocytes and 6% eosinophils. The platelet count was 269,000/cu mm. The red blood cell fluorescence was positive. Urine on standing became pink and was positive for porphyrin. There was no abnormal aminoaciduria. Stool porphyrin was positive. The child was diagnosed as congenital erythropoietic porphyria.

Discussion

Schultz is credited with the first clinical description of congenital erythropoietic porphyria(1). Taneja and Sheth published the first report from India in two brothers in 1956(2). Subsequently, a number of reports have appeared(3,4). Congenital erythro-



Fig. 1. Photosensitivity causing mutilating scars of face and fingers.

poietic porphyria is a rare inborn error of haem-biosynthesis in which there is severe mutilating photosensitivity. Hemolytic anemia with splenomegaly and a decreased life expectancy. Consanguinity is frequent and inheritance is autosomal recessive. Most cases present in childhood. The primary defect is a deficiency of uroporphyrinogen III cosynthase. The main site of enzyme defect is the bone marrow due to which uroporphyrin I which is photosensitive is overproduced, diffuses into the circulation and is deposited in various tissues including skin, teeth and bone, and is excreted in the urine and feces. The photosensitivity leads to bullous dermatosis which heals

slowly with mutilating scar formation of face, ears, nose and fingers. Hypertrichosis and irregular hyperpigmentation occur in areas of scarring. Photophobia, keratoconjunctivitis and loss of vision due to corneal scarring can also occur. The teeth are usually brown and fluoresce a reddish pink under Wood's light.

The management of these patients is symptomatic. Protection from sunlight is essential. Splenectomy for intractable hemolytic anemia may be required and has occasionally resulted in marked improvement both in anemia and in cutaneous photosensitivity(1). Hypertransfusion with packed cells are helpful as they suppress erythropoiesis and depress the production of porphyrins. Bone marrow transplantation may eventually be a possibility(1).

This case illustrates the fact that congenital erythropoietic porphyria should be considered in the differential diagnosis when a child presents with blistering skin lesions over exposed parts of the body.

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