PORPHYRIA ERYTHROPOIETICA
(A case report)

K. C. VERMA, KRISHNABIR SINGH

Porphyria erythropoietica is one of the very rare human diseases. The first authentic case was reported by Baumstrak in 1874 under the term pemphigus leprosum. Schmid et al. (1954) accepted only 34 cases as the authentic cases of congenital porphyria. Waldanstrom (1957) reported 7 cases of congenital porphyria in his series of 250 cases of porphyria. Stich (1959) reported two cases in brothers Goldberg and Rimington (1962) reviewed a total of 52 cases. These were followed by reports of many workers and a total of about 100 cases have so far been published. In India cases have been reported by Taneja and Seth (1956), Chaudhry et al. (1958), Ram (1962), Chatterjee and Chatterjea (1962), Mandal (1961) Chatterjee (1964), Handa (1965) and Sadana et al (1969).

Case Report:—

Patient, Ajit Singh, 10 years, male child was admitted in the Skin & V. D. ward of Medical College Hospital, Rohtak on 2nd September, 1970, with the complaints of passing red colored urine and eruption of bullae on the exposed parts of the body since the age of 3 months. On interrogation, the parents informed that urine of the child used to stain the clothes red ever since that age. The patient had repeatedly been having bullous lesions on the extremities and face. Bullous fluid often turned red and on healing of these bullae thin scars were formed. The trouble, though more marked in summer, persisted throughout the year. Excessive growth of the hair on face was noted since the age of three years. There was history that slight trauma or friction caused abrasion of the skin resulting in bleeding. There was no complaint of any discoloration of teeth so far. There was no history of acute pain in abdomen or any paralysis.

Examination :—

On examination the patient was moderately built and moderately nourished. He had paralytic squint in his left eye. B.P. was 110/80 m.m. Hg. Liver and spleen were not palpable. Examination of chest and nervous system revealed nothing abnormal.

Examination of skin showed multiple thin scars and pigmented macules on face, back of trunk and extremities. There were few crusted lesions on the fingers. Face had typical withered and "Monkey like" appearance (Fig. 1). On the legs there were some crusted lesions. Mucous membranes, nails and teeth were normal. Hypertrichosis was notable on face and limbs.

Investigations :—

Blood : Hb. 10.5g., T.L.C. 9500/c.m.m., P 53, L 35, M 3, E 9; E.S.R. 36 mm. Blood was examined by Rimington and Cripps (1965) method for prophyrins. Red flourescence was seen under wood's lamp.

Department of Skin & V. D.,
Medical College, Rohtak
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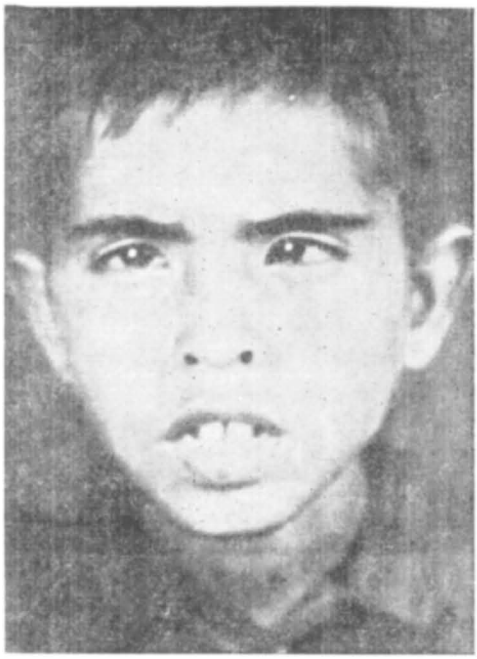

Fig. I

Urine : Urine was positive for porphyrins when seen directly under Wood's lamp. Urine was treated with glacial acetic acid and amyl alcohol. Red flourescence was noted in the upper alcohol layer.

Stool : Stool was examined for porphyrins. It was treated with glacial acetic acid, ether and hydrochloric acid. Red flourescence was seen under Wood's lamp.

Blood urea : 25 mgs.

S.S.T. : Negative.

L.F.T. : N.A.D.

Blood sugar : 80 mgs%

E.C.G. : Normal.

Skin biopsy did not reveal anything abnormal.

Family history :

The child had three sisters and two brothers. He was fourth in order of birth. No other member of the family had such trouble. There was no history of consanguinity. All his brothers and sisters were brought for examination. On provocative test with grisovin only one (younger brother) of them showed red urine. No other abnormality was detected in any of them.

Treatment :—

Patient was put on non-specific treatment. He did not show any response to riboflavin, pot-p-aminobenzoic acid or chloroquin.

Discussion :—

Congenital porphyria is a very rare disorder. There is some metabolic disorder in the synthesis of haem which results in accumulation of porphyrins in red cells and other tissues giving rise to a characteristic clinical picture. Porphyria congenita is transmitted as a recessive gene and its occurrence among several siblings is rare. There are only few reports where familial incidence has been noted in cases of congenital porphyria. These are by Harnando (1938), Mey et al (1948), Taneja (1956), Anderson (1898), Handa (1965) and Sadana et al (1968).

Clinically the condition presents with any combination of the following features :

1. Excretion of red colored urine.
2. Bullous eruptions on the exposed parts of the body due to photosensitivity caused by porphyrins.
3. Hypertrichosis.
4. Erythrodontia.
5. Hepatosplenomegaly.
6. Haemolytic anaemia.

All these features may or may not be present in one patient or at one time. In our patient history of bullous eruptions, passing of red colored urine and hypertrichosis were present.

Treatment of congenital porphyria is unsatisfactory. Stich (1950) claimed that riboflavin caused sudden and complete disappearance of porphyrins from the urine, but Neuberger's findings were contrary to that. Taneja (1956) suggested improvement of the condition after splenectomy. Protection from sunlight and trauma is generally required in these patients. Role of chloroquin therapy has been stressed by Sadana et al. (1969), Zarafonetis (1953) found good results with potassium-p-amino-benzoic acid given in the doses of 1 g. t.d.s. for four days.

Summary :

A case of congenital porphyria with brief review of literature is presented.

Acknowledgment :

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False

Studies on blood flow with tissue clearance technique using iodoantipyrine I 131 (I.A.P.) shows that there is no statistical difference between increased blood flow of the normal and atopic individual. Vessels are actually dilated in areas of blanching but the visible evidence of hyperemia is obscured by the local accumulation of edema fluid.

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