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# Congenital Erythropoetic Porphyria

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Congenital erythropoetic porphyria (CEP) or Gunther's disease is a rare autosomal recessive disorder that causes chronic photosensitivity with severe mutilating lesions(1). Of the various types of porphyria, CEP is the least common and about 70 cases have been reported from different parts of the world(2). From India, Bhutani *et al.* previously reported a case of photodermatosis due to erythropoetic protoporphyria(3). We describe a case of CEP which was diagnosed on the basis of clinical, histological and bio-chemical features at our hospital.

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*Received for publication: May 13, 1992; Accepted: April 29, 1993*

## Case Report

An 18-month-old Muslim boy born of consanguineous marriage was admitted in the Pediatric ward of North Bengal Medical Hospital with complaints of diffuse skin lesions since birth. His parents also noticed that he passed normal urine which became reddish (burgundy red) on standing. History revealed that the skin rash appeared first over the face and gradually spread to the upper part of the trunk, mainly the back.

The boy weighed 6 kg, length was 77.5 cm and had a head circumference 41 cm (all below one fifth percentile for age). Examination of the skin revealed diffuse areas of hyperpigmented macules with few areas of hypopigmented patches interpersed within them. The skin lesions were seen mostly over the photosensitive areas, being prominent on the face, scalp, neck, shoulder, extensor, extensor surface of arms and back (*Fig. 1*). The involvement of the skin was progressive with successive bouts of bullous formation followed by healing and scarring. On exposure to sunlight even for a few minutes the child felt extremely uneasy and irritated possibly due to intense itching. The sclap showed cicatrised alopecia and there was no evidence of hypertrichosis in any part of the body. There was abnormal yellowish mottling of the teeth with hypertrophic gums. The liver was palpable 2 cm and spleen 1 cm below the costal margin. The nervous system examination was normal.

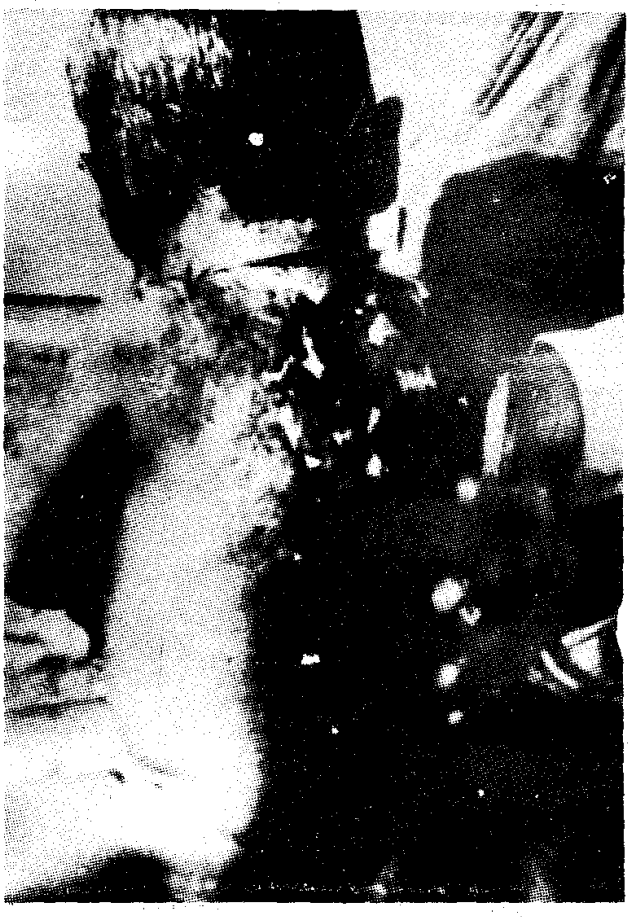

*Fig. 1. Photograph shows the extensive skin lesion, especially at the back.*

The hemogolobin level was 7.8 g/dl. Peripheral blood smear showed normal erythrocyte morphology with mild polychromasia. The total and differential leucocyte count were normal and the corrected reticulocyte count 3.1%.

As the clinical history and findings were suggestive of CEP, simple screening tests for porphyrin in blood, urine and stool were undertaken by the method of Haining *et al.*(4). The porphyrins were significantly raised in blood, urine and stool specimens.

Tests for uroporphyrin I (URO I), coproporphyrin I (COPRO I) and protoporphyrin in the blood was done by the method of Schwartz *et al.*(5). Tests for URO I and COPRO I in 24 hours specimen of urine was done by the method of Scott *et al.*(6). Tests for fecal COPRO I, URO I and protoporphyrin was done by the method of Schwartz *et al.*(7).

URO I was markedly increased in blood and urine and slightly increased in fecal specimens. COPRO I was slightly increased in blood and urine but was markedly elevated in the fecal specimen. Protoporphyrin was slightly increased in feces and normal in blood.

Histological examination of skin obtained from the back showed marked acanthosis and hyperkeratosis of epidermis with evidences of subepidermal bullae formation which were consistent with the diagnosis of CEP.

**Discussion**

The term porphyria refers to a group of diseases characterized by the excessive production and excretion of porphyrins, porphyrin precursor or both(8). The diagnosis of different types of porphyrias require the analysis and differentiation of porphyrin precursors in blood, urine and stool(1,9). The pattern of the changes in porphyrin metabolism is of paramount importance in labelling differing types of porphyria(1,9). This, in the presence of a medical history permits an exact diagnosis in most cases except in those in which the metabolic defect is latent(2,9). Enzymatic assay only helps to substantiate the diagnosis(9).

CEP is one of the rare in born errors of metabolism where vastly increased amounts of URO I are found in the marrow normoblasts, circulating erythrocytes, plasma, urine and feces due to subnormal levels of the enzyme uroporphyrinogen III synthase(8). The accumulation of URO I in the tissues and the associated hemolytic anemia ac-

ount for the clinical manifestation of the lisease(1,9). The photodermatitis of this lisease is devastating and often causes permanent disfiguration as seen in our case. The excretion of burgendy red urine in CEP begins at birth or shortly thereafter and continues throughout life(1,9). Spelenectomy s often beneficial in CEP with definite decrease in photosensitivity and reduction in urinary porphyrin excretion(10). It was, however, not undertaken in our case.

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# Congenital Lobar Emphysema


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Congenital lobar emphysema (Synonym –Panlobular emphysema of infancy) is the postnatal over distention of one or more lobes of a histologically normal lung, usually presenting with respiratory distress in infancy(1,2). This condition is rare but should


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*Received for publication: December 1, 1993; Accepted: May 12, 1993*