Günther disease, also known as congenital erythropoietic porphyria or uroporphyrinogen III synthase (UROS) deficiency, is a rare autosomal recessively inherited erythropoietic porphyria caused by mutation in the uroporphyrinogen III cosynthase gene (locus 10q25.2-q26.3). In this disorder of heme biosynthesis, patients have elevated levels of uroporphyrin 1 and coproporphyrin 1 in the erythrocytes. These porphyrin precursors are deposited in many tissues, causing photosensitivity, blistering and scarring of exposed areas leading to mutilating deformity, hypertrichosis, red urine, erythrocytosis and retarded growth. Hemolytic anemia, splenomegaly and acroosteolysis may be present. Porphyrins are markedly increased in the bone marrow, red blood cells, plasma, urine and feces.

CASE

A 1-year old male infant, born of normal consanguineous parents after a full-term normal delivery, third in birth order, with one normal sibling and one having neonatal death, presented to us with bullae on the face, dorsa of the hands and feet, and on the scalp and ear helices from nine months of age. The bullae would rupture after 3-4 days followed by crusting and scarring. The patient also had a history of failure to thrive, weakness, persistent lethargy, and passage of red-colored urine since seven months of age. General physical examination showed a weight of 8 kgs and height of 66 cm with severe pallor, hypertrichosis of the eyelashes and brownish discoloured incisor teeth (Figure 1). Systemic examination was normal except for hepatosplenomegaly. Cutaneous examination revealed crusted ulcers, hypopigmented atrophic scars on scalp, forehead, dorsa of hands and feet (Figure 2), hyperpigmentation of the distal nails of both hands with onycholysis of right index finger (Figure 3). The ophthalmological examination was normal.

Laboratory investigations revealed severe anemia (hemoglobin, 6.4 g/dL) with anisocytosis, leucopenia, thrombocytopenia, prominent monocytosis with no blasts. Bone marrow aspiration showed normoblastic erythroid hyperplasia with dysplasia. Liver and kidney function tests were normal. Ultrasound of the abdomen confirmed hepatosplenomegaly. A Wood’s lamp examination of the urine, blood and faeces showed bright pink fluorescence. Biochemical tests were positive for urinary, fecal, erythrocyte (Remington method) and plasma porphyrins, suggestive of congenital
erythropoietic porphyria. Histopathology of the bul- 
lae showed subepidermal separation with minimal in-
flammation.

**DISCUSSION**

The porphyrins are diseases caused by defects in heme biosynthesis, resulting in the accumulation and in-
creased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic de-
pending on whether the enzyme deficiency occurs in 
red blood cells or in the liver. Congenital erythropoi-
etic porphyria is the most dramatic form of porphyria, 
recognised first by Gunther, in which the enzyme uro-
porphyrinogen III cosynthetase is deficient in the bone 
marrow. This leads to elevated levels of uroporphyrin 1 
and coproporphyrin 1 in erythrocytes and urine, and cop-
roporphyrin 1 in feces. The clinical features include ex-

treme photosensitivity leading to erythema, edema and 
blisters on photo-exposed parts. The lesions heal over 
time with scarring, milia, and hypo- or hyperpigmenta-
tion. Repeated episodes of blistering result in mutila-
cion of ears, nose and hands. As the porphyrins within the 
RBCs in the cutaneous microvasculature are exposed to 
light, haemolytic anemia can occur. Treatment modal-
ities include sun protection, betacarotene, intravenous 
haematin, oral activated charcoal, transfusion of eryth-

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