Images in Clinical Practice

Congenital Erythropoeitic Porphyria

A 4-year-old boy, presented with blistering and scarring of skin. He had vesicles and erosions with mutilations of the skin over exposed surfaces (*Fig.1*). There was hypertrichosis over face and extremities, teeth were stained red (*Fig. 2*) with moderate splenomegaly. The urine was red with increased levels of urinary and erythrocyte porphyrins. A diagnosis of Congenital Erythropoeitic Porphyria was made.

This autosomal recessive condition besides its typical skin, dental and urine findings can also have ocular and hematologic findings. Erythrodontia can also be seen in fluorosis, tetracycline therapy, food stains or dentinogenesis imperfecta. Porphyrial skin lesions must be differentiated from xeroderma pigmentosum, epidermolysis bullosa and pemphigoid. The best therapy is avoidance of sunlight, while oral beta-carotenes have been tried with limited benefit.

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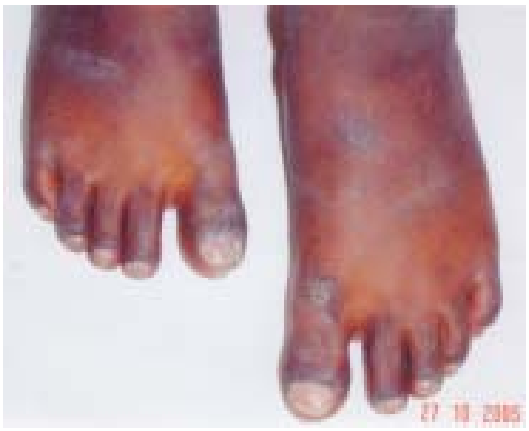

Fig. 1. Mutilated skin oner dorsum of first toe (left), and tip of first toe (right) in porphyria.

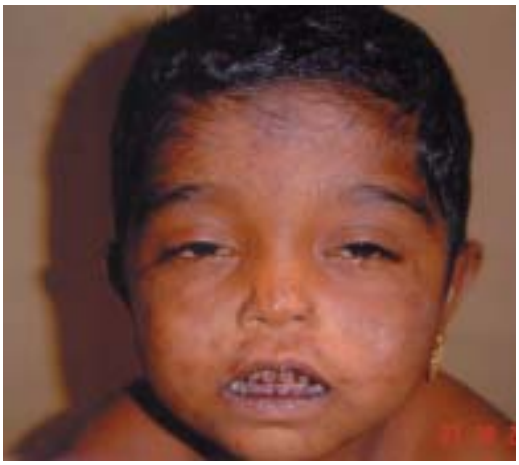

Fig. 2. Facial scarring, erosion, hyper pigmentation and erythrodontia in a patient with congenital erythropoeitic porphyria.