Dyskeratosis Congenita with Esophageal Stricture and Dermatological Manifestations

Dyskeratosis congenita, also termed Zinsser-Cole-Engman syndrome is a very rare condition characterized by pigmentation, dystrophia unguium, and leukoplakia oris (1,2).

An eight-year-old boy who complained of regurgitation of food since birth, intermittent vomiting, recurrent upper respiratory tract infection, and progressive dysphagia, was misdiagnosed as having achalasia, in a district hospital, and had also undergone a Heller's myotomy with no relief of symptoms. On examination, the child was short, had oxycephaly, periorbital pigmentation, and diffuse pigmentation on the wrists and ankles. There was leukoplakia of the tongue and pigmentation of the buccal mucosa. The blood picture and liver function tests were normal. The barium swallow showed a lower third esophageal narrowing. An upper gastrointestinal endoscopy showed a stricture with ulceration at 24 cm. The stricture was dilated, leading to complete relief of the dysphagia, and the child was able to eat solids for the first time. Over a six-month follow-up, the dysphagia has not recurred.

Dysphagia occurs in 50 % of the cases of dyskeratosis congenita, and two-thirds have congenital stenosis or diverticuli (4). The prog-

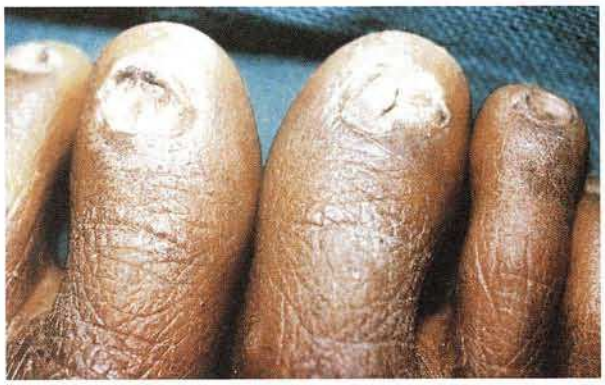

Figure 1: Dystrophy of all the nails.

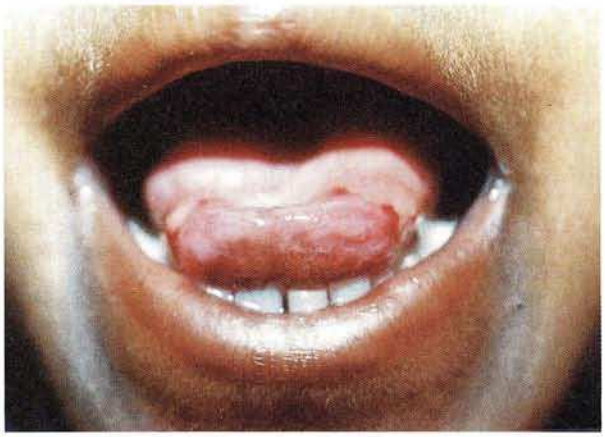

Figure 2: Leukoplakia of the tongue.

nosis is usually poor, and blood dyscrasia or carcinoma occurs in the third decade and can prove fatal (2,4,5).

P. Sawant, N. M. Chopda, D. C. Desai, U. R. Dave,
R. P. Satarkar, S. A. Nanivadekar
Department of Gastroenterology, Lokmanya Tilak Municipal Medical College and Lokmanya Tilak Muncipal General Hospital, Bombay, India

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Corresponding Author
P. Sawant, M.D., Associate Professor
Department of Gastroenterology, Lokmanya Tilak Municipal General Hospital, Sion, Bombay 400 022, India