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Residents' Corner

Rare cause of pancytopenia in a 5-year-old boy

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A 5-year-old male presented with a history of generalized weakness and progressive pallor requiring blood transfusion along with brittle nails changes in two fingers of hand since the age of 3 years. Over a span of 1 year, her mother gave a history of progressive nail changes along with whitish patches over the tongue. Since then, he has been regularly on red blood cell and platelet concentrate transfusions. Examination revealed severe pallor, oral leukoplakia, dystrophic nails, and hypopigmented spots in both the upper and lower limb [Figure 1a-c].

A complete hemogram showed pancytopenia (hemoglobin 5 g/dL, total leukocyte count 2100/cu mm, and platelet 10,000/cu mm). The corrected reticulocyte count was 0.15%. Biochemical workup revealed normal results of renal and liver function tests. Bone marrow examination revealed markedly hypocellular marrow (cellularity 10%). Secondary causes of hypoplastic marrow were ruled out (negative paroxysmal nocturnal hemoglobinuria (PNH) clone and stress cytogenetics). No abnormality was detected on ultrasonography of the abdomen. A diagnosis of dyskeratosis congenita (DKC) was made based on a clinical triad of oral leukoplakia, dystrophic nails, and skin pigmentation. He was started on anabolic steroids along with other supportive care including blood transfusions. Gradually, the patient's condition improved, and is now on regular follow-up. DKC is a rare disease; but in a young patient presenting with pancytopenia

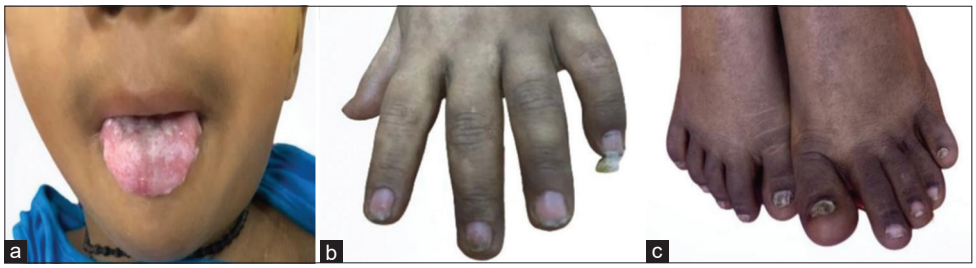

Figure 1: (a) Oral leukoplakia (b and c) Dystrophic nails of upper and lower limbs.

with a characteristic triad of skin, nail, and oral mucosa changes, this diagnosis should always be kept in mind.[1]

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

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