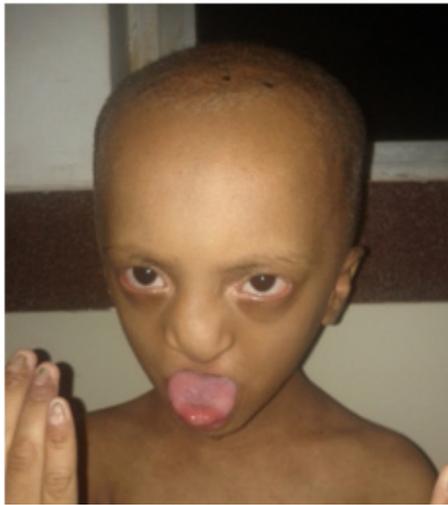


SPOT DIAGNOSIS (IMAGE GALLERY)



EPIPHORA, DYSTROPHIC NAILS AND TELANGIECTATIC ERYTHEMATOUS

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A 4years 6 months old male child born of non-consanguineous marriage presented with dysmorphic features excessive tearing from eyes. He had been operated for cleft palate. His development and birth history were normal. On examination, he had alopecia, epiphora, increased salivation, hypertelorism, low set ears, loss of eye lashes, bald tongue, oral leukoplakia, dystrophic nails, dental caries, loss of teeth, hypoplastic nipples, kyphosis and telangiectatic erythematousus.

What is the diagnosis?

Dyskeratosis congenita. It is an inherited multisystem disorder characterized by mucocutaneous abnormalities, bone marrow failure and predisposition to cancer and myelodysplastic syndrome. The diagnostic mucocutaneous (ectodermal) triad is reticulate skin pigmentation of upper body, mucosal leukoplakia and nail dystrophy. About 73 percent patients are males compatible with X-linked recessive inheritance. The remainder has either an autosomal dominant or autosomal recessive mode of inheritance. (1)The X-linked recessive form maps to Xq28, and many mutations have been identified in the DKC1 gene, which codes for the nuclear protein dyskerin. Because of impaired telomere maintenance in all 3 inherited forms, short telomeres are demonstrated in the peripheral blood cells of all patients and are a cardinal marker for marrow failure. Androgens combined with low-dose prednisone, can induce improvement of marrow function in approximately 50 percent of patients. Allogeneic hemopoetic stem cell transplantation corrects marrow failure, but with only a 50 percent survival rate. (1) .

References

1. Kliegman RM, Stanton BMD, St. Geme J, Schor N, Behrman RE. Nelson Textbook of Pediatrics, 19thed, Saunders. Philadelphia. 2011: 1687-88

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