Dyskeratosis congenita presenting with dysphagia

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ABSTRACT

Dyskeratosiscongenita (DKC) is a genetically heterogeneous disease of defective telomere maintenance that may demonstrate different patterns of inheritance. It is characterized by thetriad of dystrophy of the nails, leukokeratosis of the oral mucosa, and extensive net-like pigmentation of the skin. We report a case ofDKC who presented with a chief complaint of dysphagia.

Key words: Dyskeratosis congenita, dysphagia, leukoplakia, nail dystrophy, reticulate hyperpigmentation

INTRODUCTION

DyskeratosisCongenita (DKC) or the Zinsser–Cole–Engman syndrome is a multisystem genetic disease characterized by a triad of pigmentation and atrophy of skin, nail dystrophy, and leukoplakia. Multisystem involvement (dental, gastrointestinal, genitourinary, neurological, ophthalmic, pulmonary, and skeletal) isknown.[1] We report a case of recurrent dysphagia under care of gastroenterologist, which was referred to us for cutaneous, oral, and nail lesions. He was diagnosed to have DKC; his dysphagia was part of systemic involvement of DKC.

CASE REPORT

An 18-year-old boy, born of a nonconsanguineous marriage, presented to the hospital emergency with a three-day history of dysphagia.The patient gave a history of two similar episodes of dysphagia that were relievedon treatment. He was under the care of a gastroenterologist who performed an endoscopy and a barium swallow that revealed a very tight stricture at the upper end of esophagus [Figure 1]. It was diagnosed asacongenital web and dilated after passing a guide wire. He was referred to dermatology departmentfor pigmentation of skin and nail dystrophy. A detailed history revealed that the patient developed splitting and dystrophy of his fingernails and toenails at 5 years of age. This was followed by pigmentation of the skin within a year. Pigmentation first appeared on the neck and lower limbs, followed by the extensive involvement of the entire body with sparing of the face. Around this time he started developing oral ulcers that were recurrent despite treatment. He gave no history of eye complaints, loss of hair, dental caries, or difficulty in keeping up with studies. Except dysphagia, patient gave no history of systemic symptoms. The patient's maternal uncle had a history of similar type of skin pigmentation and nail dystrophy, and died of unknown causes at 55 years of age.

A thorough examination of the patient revealed generalized fine, lacy, reticulated, greyish-brown pattern of hyperpigmentation with areas of epidermal atrophy (poikiloderma), sparing the face [Figure 2a and b]. Examination of the genitals revealed no signs of testicular atrophy. Splitting of nails with pterygium formation and dystrophy was present, more over the fingers than the toes [Figure 2c]. Examination of the oral mucosa revealed the presence of a whitish, well-defined leukoplakic plaque on the dorsal

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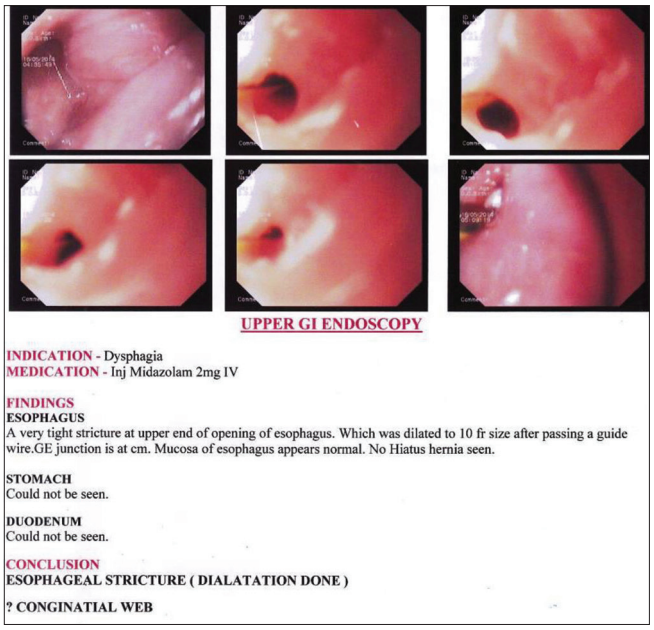

UPPER GI ENDOSCOPY

INDICATION - Dysphagia
MEDICATION - Inj Midazolam 2mg IV

FINDINGS
ESOPHAGUS
A very tight stricture at upper end of opening of esophagus. Which was dilated to 10 fr size after passing a guide wire.GE junction is at cm. Mucosa of esophagus appears normal. No Hiatus hernia seen.

STOMACH
Could not be seen.

DUODENUM
Could not be seen.

CONCLUSION
ESOPHAGEAL STRICTURE (DIALATATION DONE)

? CONGINATIAL WEB

Figure 1: Endoscopy report showing esophageal stricture

surface of the tongue, interrupted by lingual ulcers [Figure 2d]. Buccal and labial mucosa were spared. There were no significant systemic findings. Routine investigations along with skin biopsy were done. Histopathological examination of skin sample revealed melanophages in the upper dermis and vacuolization of basal cells. Routine investigations of the patient were normal. Bone marrow biopsy and lingual biopsy were refused by the patient. The classic clinical findings, family history and consistent histopathology enabled us to make a diagnosis of DKC.

DISCUSSION

DKC (Zinsser–Cole–Engman syndrome) is a rare disorder of telomere biology,[2] estimated to affect 1 in 1 million people and transmitted as an X-linked recessive (which is the most common), autosomal dominant or autosomal recessivetrait. It is characterized by the classic triad of dystrophic nails, lacy reticular pigmentation of the upper part of chest and/or neck, and oral leukoplakia. Nail changes are the first to appear and lead to splitting, dystrophy, and shedding of nails. Reticulate pigmentation and atrophy of epidermis (poikiloderma) also manifests simultaneously, or within 2–3 years. Mucous membrane lesions are common such as blisters and erosions of the lingual and buccal mucous membrane, which are succeeded by irregular patches of leukoplakia. Other mucous membranes such as conjuctival, anorectal, esophageal (or throughout the gastrointestinal mucosa) and urethral mucosa may get involved causing leukoplakia and/or stenosis. Other reported dermatological manifestations include palmoplantar thickening and hyperhidrosis, premature canities, and cicatricialalopecia.[3] The teeth tend to be defective and irregularly implanted, and periodontal disease

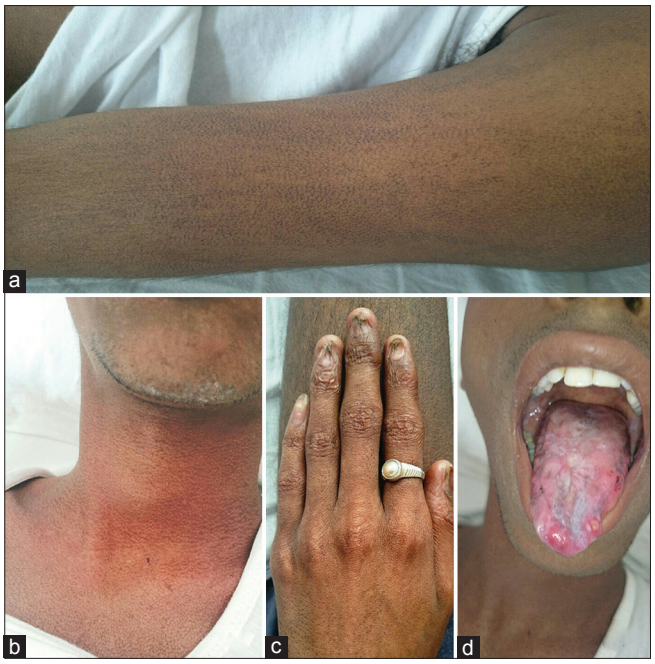

Figure 2: Patient having classical features of dyskeratosis congenita which include (a and b) reticulate pigmentation (c) nail dystrophy (d) oral leukoplakia with ulceration

and early caries are usual. General physical and mental development is sometimes delayed, and osteoporosis may occur. Immunodeficiency, intrauterine growth retardation, microcephaly, cerebellar hypoplasia, and intracranial calcification have also been reported. Prognosis is usually poor, as either blood dyscrasias (bone marrow failure, pancytopenia) or malignancies (carcinoma of mouth, Hodgkin's disease and pancreatic adenocarcinoma) may prove fatal.[4]

Our patient presented with chief complaints of recurrent dysphagia, which had been treated in the past with endoscopic dilatation. This was the patient's third episode. On examination, was found to have the classical triad of DKC which enabled us to make thediagnosis. Although dysphagia has been reported in DKC,[5-8] it is not a very common finding and it is unusual for patients to present with it as the chief complaint. Other positive findings included family history, hyperhidrosis of palms and soles, and premature greying of hair.

This case is being reported because it is a rare disease that presented to us in an uncommon manner. Dermatologists should be aware of the possibility of esophageal involvement, and history regarding dysphagia should be taken in patients of DKC. Also, the diagnosis of DKC could be made because the physician sought the help for interpreting the dermatologic findings. The authors want to stress the importance of multidisciplinary care in healthcare institutions. More often than not, it helps in establishment of correct diagnosis and better healthcare delivery through prognostication, genetic counseling, and appropriate management.

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Conflicts of interest

There are no conflicts of interest.

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