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Short communication

Squamous cell carcinoma of the tongue in a patient with dyskeratosis congenita: a rare entity

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Abstract

Dyskeratosis congenita is a rare genetic disorder that results from progressive failure of the bone marrow. It presents with a classic triad of reticular pigmentations of the skin, dystrophic nails, and oral leukoplakia; patients rarely develop cancers. We report a patient with dyskeratosis congenita who presented with squamous cell carcinoma of the oral tongue.

Keywords: Dyskeratosis congenita; Oral squamous cell carcinoma

Introduction

Dyskeratosis congenita is a rare disorder caused by failure of the bone marrow,[1] inherited as either an X-linked, recessive, autosomal dominant, or autosomal recessive, pattern.[2] It was first reported by Zinsser in 1910, followed by Engman in 1926, and Cole in 1930, and is also known as Zinsser-Engman-Cole syndrome. It classically presents as a triad of dystrophic nails, reticular pigmentation, and leukoplakia in the oral cavity.[3]

Its primary pathogenesis is defects in the maintenance of telomeres: the enzyme telomerase maintains the length of telomeres, which is the main mechanism for counteracting the process of continuous shortening.[2,4,5]

Case report

A 41-year-old man who neither smoked nor drank alcohol presented with a three-month history of an ulcer on the left lateral border of the tongue. He had reticular skin pigmentations, predominantly on his neck, trunk, and upper and lower limbs (Fig. 1), which he had first noticed at the age of 10 years. He also had dystrophic nails, which had developed first on his hands and subsequently on his feet (Fig. 2).

Intraoral examination showed a patch of erythroleukoplakia on the left lateral border of the oral tongue that extended to the dorsal surface (Fig. 3). No neck nodes were palpable. He had a low haemoglobin concentration (83 g/L) and a reduced erythrocyte count (2.24/L). The white blood cell and platelet counts were within normal limits. The triad of reticular pigmentations of skin, oral leukoplakia, and dystrophic nails, with reduced blood counts was highly suggestive of dyskeratosis congenita, so the opinion of a dermatologist was sought, who concurred with our findings.

Biopsy of the erythroleukoplakic lesion showed a moderately-differentiated squamous cell carcinoma (SCC). The patient was treated by wide local excision of the lesion on the tongue, together with selective neck dissection and reconstruction with a free radial forearm flap. He was then referred for genetic testing to the cancer genetic clinic, at which a P53 mutation analysis was positive. However, because of finan-

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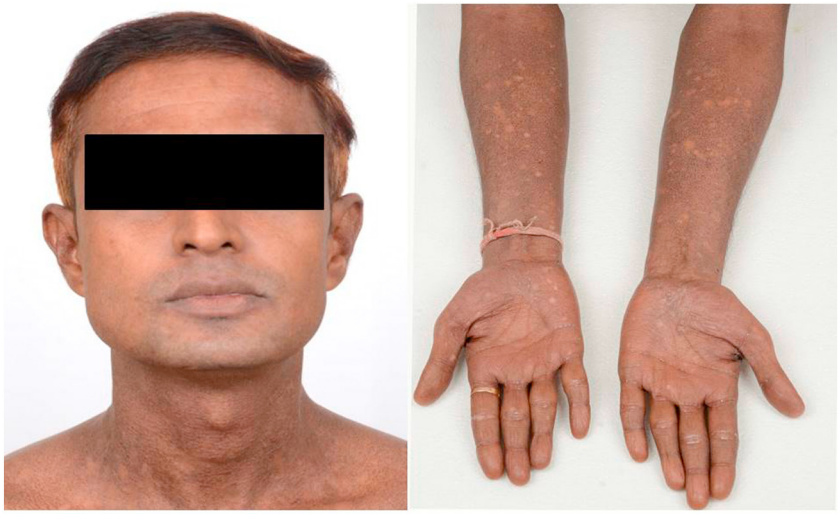

Fig. 1. Reticular skin pigmentation.

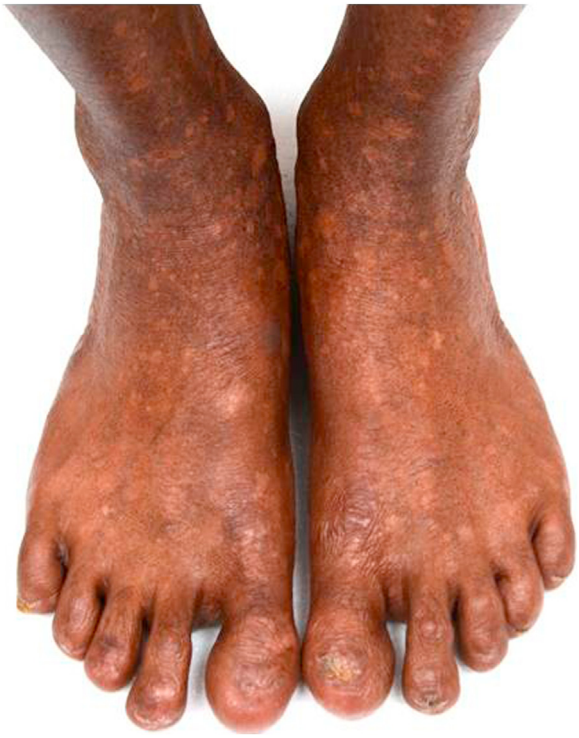

Fig. 2. Dystrophic nails.

cial constraints other genes (such as DKC1, TERC, TINF2, and TERT) could not be tested.

The histopathology report showed a lesion $5.2 \times 3.3 \times 0.3$ cm, which confirmed moderately-differentiated SCC (pT3N0). The patient was advised to have adjuvant radiotherapy.

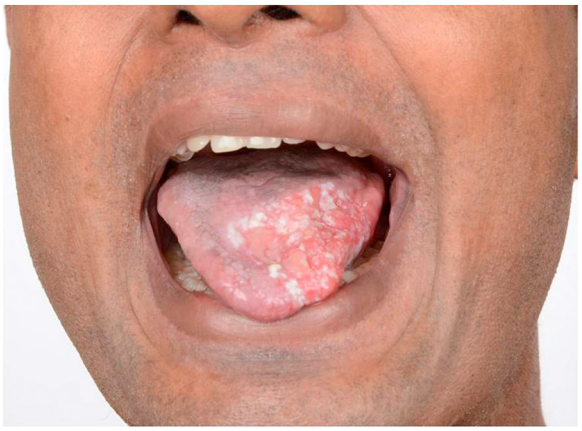

Fig. 3. Squamous cell carcinoma of the oral tongue.

Discussion

Dyskeratosis congenita can present in many ways. Apart from the classic triad, there are often other symptoms such as epiphora, blepharitis, periodontitis, taurodontism, congenital heart disease, and pulmonary fibrosis.[5]

There has been a lack of understanding of the pathogenesis for a long time, but in recent years abnormalities in the maintenance of telomeres has been thought to be the underlying cause. Among the various inheritance patterns X-linked recessive is most common, so men are more likely to be affected than women,[2] and the affected genes are DKC1, TERC, TERT, NOP10, NHP2, TINF2, TCAB1, C16orf57, CTC1, and RTEL1.[3]

The diagnosis of dyskeratosis congenita is generally made if the classic mucocutaneous triad (oral leukoplakia, dystrophic nails, and reticular pigmentation) is present, together with the features of failure of the bone marrow. At presentation this may not be overt but limited only to certain cell lines.[3] Many research workers have tried to make the diagno-

sis based on the shortening of telomeres in a sample of blood, but this has high sensitivity but low specificity.[3]

Most affected patients die of progressive failure of the bone marrow, pulmonary disease, infections, and (rarely) malignancy, with a median survival of 44 years.[4] The associated cancers are tumours of the blood such as acute myeloid leukaemia and solid tumours of the mucosa and skin.[3] A review by Alter et al showed that only 62 cases of cancer have been reported (blood or solid tumour) in patients with dyskeratosis congenita.[6] The lesions of oral leukoplakia are prone to develop oral SCC, and the most common site is the tongue, followed by the buccal mucosa. Only 24 cases of SCC in the head and neck region have been reported, with no mention of the site involved within the head and neck.[6]

Conflict of interest

We have no conflicts of interest.

Ethics statement/confirmation of patient's permission

We did not obtain ethics committee approval, but the patient gave his permission for the report to be published.

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