Letter to the Editor

A Vietnamese case of dyskeratosis congenita

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Sir,

Dyskeratosis congenita (DC) is an inherited bone marrow failure (BMF) syndrome characterized by abnormal skin pigmentation, nail dystrophy, oral leukoplakia, and cancer predisposition, with increased risk of squamous cell carcinoma and hematolymphoid neoplasms. This is a rare disease with an estimated annual incidence of < 1 in 1 million [1]. To the best of our knowledge, there is no report of DC in Vietnam in PubMed or Vietnamese Dermatology literature. Here, we report a case of DC in a Vietnamese man with the classic triad of signs.

A 22-year-old Vietnamese man presented to our hospital with hyperpigmentation and nail dystrophy. The patient had no history of alcoholism, smoking, chewing tobacco, or chronic drug use. He had white and thickened patches in the mouth since birth but had noticed asymptomatic skin pigmentation all over the body, predominantly on the chest, trunk, and arms since 10 years of age. In addition, he had noticed nail deformity when he was 15 years old. Although his sister had no abnormal skin pigmentation or nail dystrophy, his cousin (maternal side) had similar skin symptoms, including hyperpigmentation, nail dystrophy, and leukoplakia.

Clinical examination revealed fine, reticular, grey-brown pigmentation over the whole body, particularly on the neck, chest, arms, and thighs (Fig. 1a). All of the patients nails exhibited dystrophy (Fig. 1b). Leukoplakia was present on the tongue (Fig. 1c). He also had epiphora and hyperhidrosis. Blood pressure and body temperature were within normal ranges. Chest X-ray and abdominal ultrasound revealed no abnormality.

Laboratory examination found a normal white blood cell count (6,350/μL) with 69.9% neutrophils (4,400/μL). However, a low red blood cell count (3,120,000/μL), a low level of hemoglobin (116 g/L), and a low platelet count (38,000/μL) were detected. In addition, bone marrow histology further supported BMF. Histopathology of a skin biopsy obtained from the lesion with pigmentation revealed no abnormal findings, with an increasing amount of melanin and number of melanocytes within the epidermis. However, melanophages and perivascular lymphocytic infiltrates were detected within the dermis (Fig. 1d). We diagnosed DC and the patient has been advised to follow-up every 3 months with a dermatologist and hematologist.

DC is characterized by a triad of nail dystrophy, skin pigmentation, and leukoplakia. It can be associated with BMF, as presented in this case. The skin pigmentation and nail changes usually appear first, before the age of 10 years, and then BMF often develops before the age of 20 years, which was similar to our case [2]. Because a higher prevalence is found in men than in women, with a ratio of 13:1 reported for DC [3], X-linked recessive conditions are suggested to be related to the pathogenesis of this disease. Considering that the patient's cousin had a similar clinical appearance, we should still take the genetic background into account in our case.

Many studies have demonstrated that DC is principally a disease of defective telomere maintenance [1,4]. DC patients normally have very short telomeres, which could be related to the fact that some DC patients are reported to have mutations in different genes encoding components of the telomerase complex [2,4,5]. Previous

How to cite this article: Thi PH, Huu DL, Thai HV, Thanh TT, Dinh HP, Hau KT, Sugawara K, Tsuruta D. A Vietnamese case of dyskeratosis congenita. Our Dermatol Online. 2018;9(3):331-332.

Submission: 07.02.2018; **Acceptance:** 29.04.2018

DOI:10.7241/ourd.20183.27

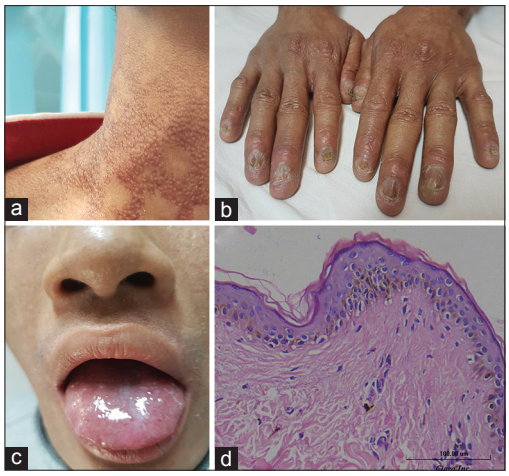

Figure 1: Clinical manifestations and histopathology of the patient. (a) Reticular, grey brown pigmentation on the neck and chest. (b) Nail dystrophy. (c) Oral leukoplakia. (d) Skin histopathology shows increased melanin and melanocytes in the epidermis and melanophages and perivascular lymphocytic infiltrate in the dermis.

electron microscopy studies revealed that cells isolated from DC patients have an embryonic immature nucleus, which could induce malignant transformation [6]. Cancer usually develops after the third decade; the most frequent solid malignancies are head and neck squamous cell carcinomas [7]. Although we have not detected any malignant changes, we need to follow-up with this first case of DC in Vietnam regularly.

CONSENT

The examination of the patient was conducted according to the Declaration of Helsinki principles.

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Source of Support: Nil, **Conflict of Interest:** None declared.