Dermatoscopic Findings of Dyskeratosis Congenita: Case Report

Dyskeratosis congenita (DC) is a rare inherited syndrome characterized by reticular pigmentation of the skin, nail dystrophy, mucosal leukoplakia, predisposition to cancer and bone marrow failure. Dermatoscopy is a non-invasive and valuable method of skin imaging in the diagnosis of pigmented skin lesions. Nevertheless, to our knowledge, the dermoscopic features of the pigmented lesions in DC have not been described previously.

CASE REPORT

A 30-year-old man presented to our clinic for evaluation of his mucosal nonhealing lesions. These lesions had grown over 5 years without a symptom. The medical history of the patient was well until the age of 8 when hy-
perpigmentation was noted. He continued to develop areas of hyperpigmentation over the trunk, face, neck, and extremities. Nail dystrophy was also started at 10 years of age. There was no evidence of mental or growth retardation. His father had also dystrophic nails but no other symptoms. Dermatologic examination of this patient revealed skin, nail, and mucosal lesions. He had subtle reticular hyperpigmentation of the skin of his face, neck, trunk, arms and legs. This finding was more evident around the flexor and sacral areas (Figure 1a). Three fingernails were missing, with the rest affected by splitting and longitudinal ridging with pterygium formation (Figure 1b). There were thick, white, rough, painless plaques measuring 4 x 4 cm on both buccal mucosa consistent with leukoplakia. Pulmonary function tests, haematologic studies and further investigations to exclude malignancy were normal. Biopsies of the skin and lateral tongue plaque were obtained. Histopathology of oral leukoplakia lesion demonstrated benign features with simple hyperkeratosis. The skin biopsy showed mildly atrophic skin with scattered dermal macrophages and foci of increased pigment within the basal layer consistent with skin involvement by DC. Dermatoscopy was performed with the use of a Mole max II™ videodermoscope (MoleMax-Derma Medical Systems, Vienna, Austria). One of the major finding was the generalized light to dark brown coloured pigmented lesions showing a delicate and regularly meshed pigment network corresponding to actinic lentigo or lentigo simplex. Additionally dark brown coloured dots and globules were observed throughout the body (Figure 2a). They were regular in size and shape, and are quite evenly distributed. Around the flexor regions, the reticular pigment network disappeared (Figure 2b). Instead we have detected the clusters of dots and globules, in these very dark brown coloured areas. The nail dermatoscopy revealed no melanocytic lesion. There were reddish, longitudinal bands on the affected nails (Figure 3).

**DISCUSSION**

DC presents with mucocutaneous and nail changes in the 1st or 2nd decade of life. A variety of other abnormalities in these patients have been documented, including dental, gastrointestinal, genitourinary, neurologic, ophthalmic, pulmonary, and skeletal involvement. Recently, there have been significant advances in DC and these had been facilitated by the DC Registry established at Hammersmith Hospital. Regular follow-up of the patient is essential owing to the possibility of malignant changes within oral, other mucosal and cutaneous sites.

DC should be differentiated from a great variety of cutaneous lesions that cause reticular hy-
perpigmentation. DC should especially be distinguished from Fanconi’s anemia, which is also associated with pigmented abnormalities, pancytopenia and an increased risk of neoplasia. The lacy reticulated pigmentation in DC favors the neck and upper chest, with onset during the first decade. However in Fancony anemia the pigmentation occurs more generalized and diffuse. Although clinical appearance have distinct features, in most cases, the clinical diagnosis should be confirmed by further expensive and complex diagnostic methods including flow fluorescence in situ hybridization and detection of the response of patients’ lymphocytes to mitomycin C. Other important disorders characterized by reticulated pigmentation are erythema ab igne, Dowling-Degos disease, confluent and reticulated papillomatosis of Gougerot and Carteaud and lichenoid dermatoses including lichen planus pigmentosus. Dermatoscopy is a noninvasive technique that has been used to make more accurate diagnoses of pigmented skin lesions. Over the past few years, it has been proven to be useful in a variety of cutaneous disorders, including ectoparasitic infestations, infections, hair and nail abnormalities and psoriasis. It represents an important and relatively simple diagnostic technique in daily practice.

Nowadays, this non-invasive technique has been started to use in pigmented skin disorders in addition to the differential diagnosis of melanocytic lesions. Gil et al demonstrated the dermoscopic features of ochronosis. They also showed a good correlation between the results of dermoscopy - brown-gray globular, annular, and arciform structures - and the histologic findings. In 2003 Vazquez et al demonstrated the dermoscopic features of lichen planus lesions. They recognized three dermoscopic pigmented patterns. (a) diffuse brownish lesions (b) dotted lesions demonstrating fine or coarse grey-blue or brown dots or globules; (c): mixed (diffuse areas with dotted structures). With their study they also suggested that dermoscopy has a potential value in determining the prognosis since blue dots seemed to be more persistent.

**FIGURE 2:** a: Generalized light to dark brown coloured pigmented lesions showing a regularly meshed pigment network with regular, dark brown coloured dots and globules. b: Around the flexor regions, the reticular pigment network disappeared and replaced by clusters of dots and globules.

**FIGURE 3:** The dermatoscopy of fingernails revealed only reddish, longitudinal bands on the affected nails.
The major feature was the delicate pigment network with regularly distributed dots and globules, in our case. This pigment network was disappearing and clusters of dots and globules were replacing them in dark coloured flexural areas. Besides, it is important to note that there was no melanocytic lesion on nail examination.

By means of this case report, the dermoscopic features of the pigmented lesions in DC are described for the first time. Although at this level, it is not possible to assume that these findings are specific or characteristic for this syndrome; future reports highlighting the dermoscopic features may simplify and be helpful for the clinical diagnosis of DC.

REFERENCES