Dear Editor,

A 23-year-old male presented to our clinic with complaints of progressive roughening, cracking, and fragility of finger and toenails since the age of 9 years. Examination revealed longitudinal ridging and splitting of all finger nails with mild dystrophy and onychoschizia of some nails [Figure 1a]. The toenails were partially involved. A closer cutaneous evaluation revealed tan-brown mottled to reticulate pigmentation on the anterior aspect of the neck and V area of the chest [Figure 1b]. The oral cavity showed asymptomatic leukokeratosis of the tongue [Figure 1c]. The skin over the palms and soles was dyspigmented and thickened with the absence of dermatoglyphics [Figure 1d]. Rest of the mucocutaneous examination was normal. Considering the constellation of findings, history was probed further. He was born at term, in good health out of non-consanguineous marriage, through unassisted normal vaginal delivery. The antenatal, perinatal, and postnatal period were uneventful, and he had normal developmental milestones, on par with his age. There was no significant illness or drug intake in the mother during pregnancy or lactation. The patient disclosed that his sister had aplastic anemia and died at the age of 15. His parents were unaffected; there were no relevant findings in his cousins or grandparents. He denied consumption of tobacco, and there was no history suggestive of photosensitivity, aggravation of pigmentation on sun exposure, or any other systemic complaints.

On onychoscopy, nail surface abnormalities were seen in the form of several parallel longitudinal ridges [Figure 2a], longitudinal splits forming deep furrows [Figure 2b], horizontal splits at the free distal edges resulting in jagged edges, and splinter hemorrhage [Figure 2c].

Dermoscopic examination revealed pigmented lines consisting of brown dots and globules arranged as a netlike pattern on the V area of the neck [Figure 3a] and the absence of dermatoglyphics [Figure 3b]. Blood analysis revealed pancytopenia (hemoglobin—7.1 g/dL, white blood cells—1600 cells/mL, platelets—46,000 cells/mL, and erythrocyte sedimentation rate—27 mm/h).

Based on the clinical presentation, significant family history, and deranged hematological parameters, a diagnosis of dyskeratosis congenita (DKC) was thought of. Fanconi’s anemia was excluded because of the presence of nail changes and leucoplakia. Genetic disorders of photosensitivity and reticulate pigmentary abnormalities were excluded on the basis of absence of photosensitivity and the presence of hematological abnormalities, respectively.

The patient was treated with oral pyridoxine, folic acid, danazol and with injectable erythropoietin under the guidance of the hematologist and is under regular follow-up.

DKC is an inherited multisystem bone marrow failure syndrome displaying marked clinical and genetic diversity. The mode of inheritance is highly variable with several germline mutations affecting the telomerase complex components (e.g., DKC1, TINF2, TERC, and TERT) that cause defective telomere maintenance, leading to prematurely shortened telomeres which negatively affect the proliferative potential of stem cells.[3] First described by Zinsser and later by Engman and by Cole et al[2] this rare genodermatosis in a classical case is characterized by the development of a triad of ectodermal dysplastic features: reticulate hyperpigmentation, oral leucoplakia, and nail dystrophy by the age of 10 years. The cardinal pigmentary changes present as hypo- or hyperpigmented macules and patches in a reticulate or mottled pattern chiefly involving the sun-exposed areas such as the upper trunk, V area of the chest, and face. Atrophy and telangiectasia resulting in poikilodermatus changes, alopecia of the eyebrows, eyelashes, and scalp, canities, palmoplantar hyperkeratosis, hyperhidrosis, and adermatoglyphia are other cutaneous findings.[3]

Mucosal involvement in the form of black patches or white patches presenting as leucoplakia is a pathognomonic feature and occurs in nearly 80% of patients. It may occur at birth or within the first few months and typically involves tongue and buccal mucosa. The oropharynx, gums, and hard palate are less frequently affected.[3]
Nail involvement, an essential component of the diagnostic triad, is seen in approximately 90% of patients with DKC. In most cases, fingernails are more commonly and severely affected and also involved earlier than toenails. Ridding, longitudinal splitting, onychoschizia, and brittleness are the preliminary nail abnormalities to develop and may manifest as early as in the first few months after birth or can occur in adolescence. With disease progression, in due course of time, nail dystrophy sets in, resulting in pterygium formation and progressive loss of a distal portion of the nail, and at times, even complete nail loss can occur. Mild to severe clubbing, secondary to lung infection, black nails, and fingertip atrophy may arise rarely.

In addition to the typical ectodermal dysplastic aberrations, an array of non-mucocutaneous abnormalities involving dental, gastrointestinal, genitourinary, neurological, ophthalmic, pulmonary, and skeletal systems arise with time. Potentially fatal extracutaneous manifestations include pulmonary fibrosis, pulmonary vasculature abnormalities, myelodysplastic syndrome, acute myeloid leukemia, solid tumors, and pancytopenia with bone marrow failure. Pancytopenia involving at least two of the cell lineages affects 76% of the patients by 20 years of age, and the resulting bone marrow failure is the cause of premature mortality by the third decade in the majority of cases (80–90%). There is also a heightened propensity to develop malignant mucosal neoplasms in the third decade, particularly squamous cell carcinoma occurring within sites of leukoplakia. Cutaneous squamous cell carcinoma, Hodgkin’s lymphoma, adenocarcinoma of the gastrointestinal tract, and bronchial and laryngeal carcinoma are other neoplasms reported.

The diagnosis of DKC is usually established based on the history and cluster of clinical findings after excluding photosensitive genodermatoses, bone marrow failure syndromes, reticulate pigmentary disorders (dermatopathia pigmentosa reticularis, dyschromatosis symmetrica hereditaria, reticulate acropigmentation of Kitamura, Dowling-Degos disease, macular amyloidosis, amyloidosis cutis dyschromia), and dermatological and non-dermatological causes of trachyonychia and nail dystrophy (lichen planus, psoriasis, alopecia areata, Darrier’s disease, and pachyonychia congenita). The dermoscopic and onychoscopic differentiating features of DKC from the common differentials are listed [Tables 1 and 2].

Although the onset of DKC is in early childhood, there is wide variability concerning the age at presentation, clinical features, progression, as well as severity, and they may not manifest in a predictable pattern in each patient. The clinical findings are expressed progressively at different stages of life, and the diagnosis is often delayed or missed.

Figure 2: (a) Onychoscopy demonstrating longitudinal ridging. DermLite DL4N, polarized mode x10 (black arrow). (b) Onychoscopy demonstrating longitudinal splits forming deep furrows—DermLite DL4N, polarized mode x 10 (blue arrow). (c) Onychoscopy demonstrating longitudinal splits, furrows, with splinter hemorrhage (orange circle), and distal lamellar splitting (yellow arrow). DermLite DL4N, polarized mode x10

Figure 3: (a) Dermoscopic examination demonstrating pigmented lines consisting of brown dots and globules arranged as a netlike pattern—DermLite DL4N, polarized mode x10 (black circle). (b) Dermoscopic image demonstrating adermatoglyphia—DermLite DL4N, polarized mode x10 (blue circle)
Dermoscopic and TERT and TERC mutations detected in cryptic Brown or white hub in the center

Onychoscopic findings
1. Pitting, fuzzy lunula, and leukonychia

Table 1: Dermoscopic features of reticulate pigmentary disorders

| Reticulate pigmentary disorder | Dermoscopic findings |
|--------------------------------|----------------------|
| DKC                            | Pigmented lines consisting of brown dots and globules arranged as netlike pattern on the V area of the neck |
| Reticulate acropigmentation of Kitamura | Absence of dermatoglyphics |
| Dyschromatosis symmetrica hereditaria | Black dots on dark-brown or tan background and white scales |
| Dowling-Degos disease | Conspicuous interruptions in the dermatoglyphics |
| Macular amyloidosis | Reticulate (net-like) and/or uniform hyperpigmented and/or hypopigmented spots |

Table 2: Onychoscopic features of common disorders causing trachyonychia and nail dystrophy

| Dermatoses | Onychoscopic findings |
|------------|-----------------------|
| Lichen planus | Onychorrhexis, onycholysis, splinter hemorrhages, longitudinal erythronychia |
| Psoriasis | Multiple leukonychia, punctate leukonychia, melanonychia, onychomadesis |
| Darier’s disease | Distal splitting, prominent hyponychial vascular structures, and distal short longitudinal white lines |
| Nail plate thickening and crumbling, red lunula, lamellar splitting, Beau’s lines, dystrophy, onychomadesis, dilated capillaries, onychoysis, splinter hemorrhage, scales, oil spots, brown discoloration, white streaks |

The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship
Nil.

Conflicts of interest
There are no conflicts of interest.

Resham Vasani, Kavya Baddireddy
Bhojani Clinic, Mumbai, Maharashtra, Government Hospital for Mental Care, King George Hospital, Visakhapatnam, Andhra Pradesh, India

Address for correspondence:
Dr. Kavya Baddireddy,
5, Ocean Drive Layout, Gitam College Post, Visakhapatnam - 530 045, Andhra Pradesh, India.
E-mail: kavyabaddireddy@gmail.com

References
1. Terada K, Miyake K, Yamaguchi H, Miyake N, Yamanaka K, Kojima S, et al. TERT and TERC mutations detected in cryptic dyskeratosis congenita suppress telomerase activity. Int J Lab Hematol 2020;42:316–21.
2. Cole HN, Rauschkolb J, Toomey J. Dyskeratosis congenita with pigmentation, dystrophia unguis and leukokeratosis oris. Arch Dermatol Syphiligraphie. 1930;21:71–95.
3. Ward SC, Savage SA, Giri N, Alter BP, Rosenberg PS, Pichard DC, et al. Beyond the triad: Inheritance, mucocutaneous phenotype, and mortality in a cohort of patients with dyskeratosis congenita. J Am Acad Dermatol 2018;78:804–6.
4. Alsabbagh MM. Dyskeratosis congenita: A literature review. J Dtsch Dermatol Ges 2020;18:943–67.
5. Baykal C, Kavak A, Gülcen P, Büyükbabani N. Dyskeratosis congenita associated with three malignancies. J Eur Acad Dermatol Venereol 2003;17:216–8.

This is an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.

How to cite this article: Vasani R, Baddireddy K. Dermoscopic and onychoscopic features of dyskeratosis congenita. Indian Dermatol Online J 2022;13:786-8.

Received: 12-Jan-2022. Revised: 11-Apr-2022. Accepted: 22-Apr-2022. Published: 21-Oct-2022.

© 2022 Indian Dermatology Online Journal | Published by Wolters Kluwer - Medknow