MAYO CLINIC

Dyskeratosis Congenita

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A 14-year-old girl with dyskeratosis congenita (DC) was evaluated for widespread symptomatic oral lesions that developed shortly after undergoing allogeneic hematopoietic stem cell transplant (allo-HSCT) at the age of 1 year. Physical examination revealed generalized reticulate pigmentation of the skin and nail dystrophy consistent with the DC phenotype (Figures 1 and 2) Intraoral examination revealed generalized Wickham striae and erythema of the bilateral buccal mucosa with a linear pseudomembranous ulcer on the right buccal mucosa (Figure 3). Histopathologic analysis of the lesion revealed lichenoid mucositis without evidence of dysplasia and cumulative findings were consistent with oral chronic graft-versus-host disease (cGVHD). The patient was

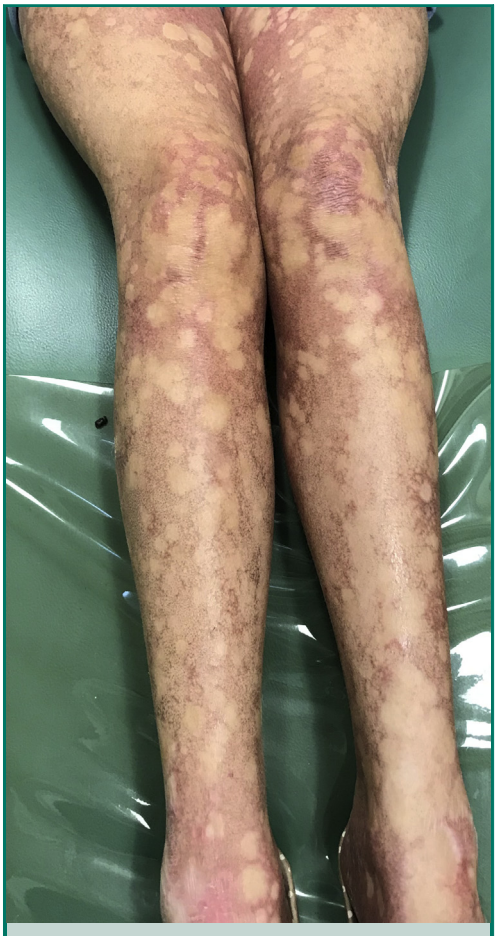

FIGURE 1. Dyskeratosis congenita skin phenotype demonstrating characteristic reticular skin pigmentation.

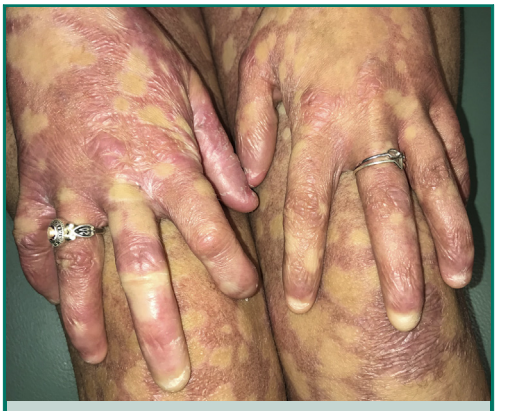

FIGURE 2. Dyskeratosis congenita nail phenotype demonstrating characteristic nail dystrophy.

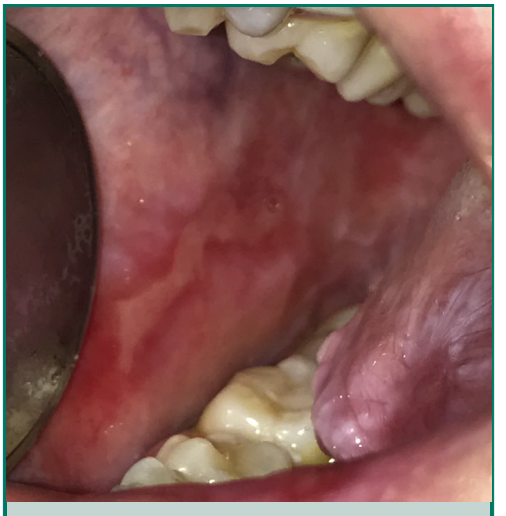

FIGURE 3. Oral lesion in a patient with dyskeratosis congenita demonstrating a linear pseudomembranous ulcer with surrounding striae on the right buccal mucosa.

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prescribed fluocinonide gel 0.05% to oral lesions twice daily and dexamethasone solution 0.5 mg/5 mL, 10-mL swish and spit twice daily for symptom management, and nystatin rinse 100,000 U, 5-mL swish and spit three times daily for antifungal prophylaxis.

DC is a rare, inherited bone marrow failure syndrome characterized by dystrophic nails, reticular skin pigmentation, and oral leukoplakia.[1] This condition is associated with several telomerase-shortening genes with multiple inheritance patterns, including X-linked (*DKC1*), autosomal dominant (*TERT*, *TINF2*), or autosomal recessive (*NOP10*, *NHP2*, *TCAB1*).[1] Bone marrow failure is the principal cause of morbidity and mortality in patients with DC, and allo-HSCT is the only curative treatment.[1] Survivors of allo-HSCT are at increased risk of developing second solid malignancies, including cutaneous and oral squamous cell carcinoma (SCC).[2]

Patients with DC are also at increased risk of developing malignancies, including head and neck SCC, cervical SCC, and non-Hodgkin lymphoma.[1,3] Oral leukoplakia has been observed in up to 80% of patients with DC, with a reported 1000-fold increased risk of malignant transformation to oral SCC compared with the general population.[3] Oral cGVHD, a common complication of allo-HSCT, is also considered a potential risk factor for development of oral SCC.[2] Patients with oral cGVHD or DC must undergo continuous surveillance because of the increased risk of oral and systemic malignancies associated with both conditions.

Abbreviations and Acronyms: **allo-HSCT** = allogeneic hematopoietic stem cell transplant; **cGVHD** = chronic graft-versus-host disease; **DC** = dyskeratosis congenita; **SCC** = squamous cell carcinoma

Potential Competing Interests: Dr Stoopler is employed by the University of Pennsylvania. Dr Shanti reports no competing interests.

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1. Fernández García MS, Teruya-Feldstein J. The diagnosis and treatment of dyskeratosis congenita: a review. *J Blood Med*. 2014;5:157-167.
2. Mawardi H, Elad S, Correa ME, et al. Oral epithelial dysplasia and squamous cell carcinoma following allogeneic hematopoietic stem cell transplantation: clinical presentation and treatment outcomes. *Bone Marrow Transplant*. 2011;46(6):884-891.
3. Bongiorno M, Rivard S, Hammer D, Kentosh J. Malignant transformation of oral leukoplakia in a patient with dyskeratosis congenita. *Oral Surg Oral Med Oral Pathol Oral Radiol*. 2017;124(4):e239-e242.