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Abstract/Case Report:

A 12-year-old female presented for evaluation of persistent nail abnormalities present since early childhood. Examination demonstrated bilateral anonychia and hyponychia of the fingernails, sparing the right first and fifth digits and the left fifth digit. The patient reported that the fingernails spontaneously detach as they grow distally along the nail plate. The toenails were present bilaterally but appeared dystrophic. In addition, the patient endorsed gradually progressive pigmentary changes involving the forehead, chest, back, and arm.

Further cutaneous assessment revealed patchy hyperpigmented and hypopigmented macules involving the upper chest and lower neck, along with mild oral leukoplakia of the buccal mucosa. Genetic evaluation was recommended at this time but not pursued by the patient. Soon after, the patient was evaluated by hematology/oncology for chronically elevated ferritin and additional evaluation revealed a history of easy bruising. Lab evaluation demonstrated pancytopenia with macrocytic anemia. Given the longstanding nail dystrophy, mucocutaneous dyspigmentation, and oral manifestations, an underlying genodermatosis was suspected.

Subsequent genetic analysis identified a pathogenic mutation in the *TINF2* gene, confirming a diagnosis of Dyskeratosis Congenita (DKC). While the patient's bone marrow evaluation showed no evidence of malignancy, *TINF2* mutations are associated with early-onset disease and a high risk of progressive bone marrow failure. Accordingly, the patient's near-complete anonychia, dystrophic toenails, and diffuse hyper- and hypopigmented patches served as clinical clues, prompting multidisciplinary evaluation before progression to transfusion dependence, malignancy, or advanced marrow failure. This case highlights that early detection of nail and pigmentary findings concerning for DKC can lead to a diagnosis during a clinically stable phase, allowing early surveillance before progression to advanced systemic disease.