

Rothmund–Thomson Syndrome Type 2 Misdiagnosed as Dyskeratosis Congenita

Rothmund-Thomson Sendromu Tip 2'nin Diskeratozis Konjenita Olarak Yanlış Teşhisi

Yılmaz Orulluoğlu E. and Aslan D.: Rothmund-Thomson Syndrome Type 2

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08.03.2026
23.06.2026

A 13-year-old girl presented with anemia and thrombocytopenia. Given her striking phenotype, a bone marrow aspiration was performed to exclude inherited bone marrow failure syndromes; the marrow was normocellular for her age, with no dysplastic changes or cytogenetic abnormalities, and blood counts subsequently normalized following hematinic treatment. Physical examination revealed features suggestive of dyskeratosis congenita (DC), including reticular hyperpigmentation, alopecia, and nail dystrophy. Although initial genetic screening for all known DC-associated genes at that time at an international DC registry was negative and telomere length was within normal limits for her age-adjusted deciles, the patient was enrolled in the registry as a "gene-negative" DC case. She was monitored under this diagnosis until she was lost to follow-up.

Nineteen years later, the patient returned to our clinic to seek genetic counseling regarding the potential hereditary risks for her offspring following a successful and spontaneous pregnancy, which proceeded without any hematologic or obstetric complications. Current clinical findings remained consistent with her initial presentation, as shown in Figure 1: Panel A (reticular hyperpigmentation), Panel B (alopecia), and Panel C (nail dystrophy). Whole-exome sequencing identified a homozygous mutation in *RECQL4* (c.2344G>A; p.Asp782Asn), confirming Rothmund–Thomson syndrome (RTS) type 2.

While RTS and DC share overlapping clinical features, normal telomere length may serve as a helpful discriminator to support differentiation between these entities [1,2,3,4,5]. Precise molecular diagnosis is vital for clinical management: while DC requires vigilance for pulmonary and hepatic complications, RTS necessitates lifelong surveillance for osteosarcoma and skin malignancies [2,3,5]. Furthermore, the successful pregnancy in this patient highlights a unique clinical intersection, as fertility potential is typically significantly reduced in RTS -in contrast to DC- and reported pregnancies are rarely uncomplicated. This case emphasizes that RTS should be considered in the differential diagnosis of patients presenting with a DC-like phenotype but preserved telomere length.

Keywords: Rothmund–Thomson syndrome, dyskeratosis congenita, telomere length

Anahtar Kelimeler: Rothmund-Thomson sendromu, konjenital diskertoiz, telomer uzunluğu

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Figure 1. Clinical features of the patient at age 32, demonstrating persistent phenotypic findings consistent with her initial presentation at age 13. (A) Reticular hyperpigmentation, (B) Alopecia, (C) Nail dystrophy.