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Rothmund–Thomson Syndrome: Proof That Rare is Always in Fashion

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Abstract

Rothmund–Thomson syndrome (RTS) is a rare autosomal recessive genodermatosis characterized by photosensitivity, poikiloderma, growth retardation, ocular abnormalities, and an increased risk of malignancies. The disease typically presents in early infancy with photosensitive erythema and edema that gradually evolves into chronic poikiloderma involving both exposed and non-exposed areas. We report a case of a 30-year-old male who presented with exfoliative erythematous lesions over the hands and feet, generalized poikiloderma, nail dystrophy, and characteristic bird-like facies. Cutaneous manifestations had their onset at three months of age and progressed over time. Histopathological examination showed features consistent with poikiloderma vasculare atrophicans. Based on the characteristic clinical evolution, histopathology, and exclusion of close differentials such as xeroderma pigmentosum and Kindler syndrome, a diagnosis of Rothmund Thomson syndrome was made. The patient was managed with strict photoprotection and emollients and advised lifelong surveillance for early detection of malignancies and ocular complications. This case is reported due to the rarity of the disorder, its classical clinical evolution from infancy to adulthood, and to emphasize the

importance of early diagnosis and long-term multidisciplinary follow-up.

Keywords: Rothmund–Thomson syndrome; Poikiloderma; Photosensitivity; Genodermatoses; RECQL4; Rare disease

Introduction

Rothmund–Thomson syndrome (RTS) is a rare autosomal recessive genodermatosis characterized by early-onset poikiloderma, photosensitivity, growth retardation, skeletal abnormalities, ocular involvement, and predisposition to malignancies[1]. The condition was first described by Rothmund in 1868 and later further delineated by Thomson[2].

The etiopathogenesis of RTS is attributed to mutations in the *RECQL4* gene, which encodes a DNA helicase involved in DNA replication and repair. Defective DNA repair mechanisms result in genomic instability, explaining the photosensitivity and increased susceptibility to cutaneous and extracutaneous malignancies[3–5].

RTS is extremely rare, with an estimated prevalence of less than 1 per 1,000,000 population and no clear gender or ethnic predilection[6]. Cutaneous manifestations usually begin between 3 and 6 months of age, initially affecting photo-exposed areas before progressing to generalized poikiloderma[7]. Owing to its rarity and phenotypic overlap with other photosensitive genodermatoses, RTS is frequently underdiagnosed. This report is special as it demonstrates the classical evolution of RTS from infancy into adulthood with supportive histopathological findings, reinforcing the need for early recognition and lifelong surveillance.

Case Report

Chief Complaint - Skin peeling with redness over both hands and feet for the past 2 years.

History - A 30-year-old unmarried male presented with exfoliative erythematous lesions over the dorsum of both hands and feet. He was apparently normal until 3 months of age, when he developed erythema followed by blistering over photo-exposed areas, predominantly involving the “V” area of the neck and distal extremities. With increasing age, the lesions gradually evolved into dull brown hyperpigmentation interspersed with hypopigmented areas and later involved covered parts of the body.

He received intermittent treatment until 9 years of age, after which follow-up was discontinued. At 14 years, he underwent surgical repair for ventricular septal defect. At 22 years, he was diagnosed with pulmonary tuberculosis and completed six months of antitubercular therapy, during which he was also diagnosed with ulcerative colitis. He was born out of a non-consanguineous marriage with no similar illness in the family.

Clinical Features

The patient was ill-built and undernourished. General physical examination revealed clubbing, with absence of pallor, icterus, cyanosis, lymphadenopathy, and edema. Vital parameters were within normal limits.

Cutaneous examination

Revealed multiple discrete hyperpigmented and hypopigmented macules and patches over the trunk and extremities. Ill-defined erythematous plaques with exfoliation and marked atrophy were present over the dorsum of both hands. Well-defined erythematous to hyperpigmented plaques with exfoliation and erosions were seen over both feet. Multiple ill- to well-defined

atrophic plaques were present over the anterior aspects of both legs. Nails showed dystrophy with subungual hyperkeratosis and distal onycholysis.

Facial examination revealed characteristic bird-like facies with a prominent forehead and beaked nose.

Investigations

Histopathological examination showed hyperkeratosis, focal parakeratosis, epidermotropism of atypical lymphocytes without Pautrier microabscess formation, vacuolar alteration of the basal layer, and a band-like lymphocytic infiltrate with dilated capillaries and wiry collagen bundles in the upper dermis, consistent with poikiloderma vasculare atrophicans.

Diagnosis

Rothmund–Thomson syndrome

Treatment Plan and Prognosis

The patient was managed with strict photoprotection, photoprotective clothing, and regular emollients. He was advised lifelong dermatological and ophthalmological follow-up for early detection of cutaneous malignancies and ocular complications. Genetic counselling was recommended. Prognosis depends on vigilant long-term surveillance and early management of complications.

Discussion

RTS is a rare genodermatosis with multisystem involvement, predominantly affecting the skin and eyes[8]. The underlying *RECQL4* mutation results in impaired DNA repair following ultraviolet exposure, accounting for photosensitivity and increased malignancy risk[4,5]. The classical biphasic cutaneous progression from infantile photosensitive erythema to chronic poikiloderma, as seen in this patient, is a hallmark feature[7,9].

Differential diagnoses such as xeroderma pigmentosum and Kindler syndrome were considered but excluded based on clinical course, lack of severe neurological involvement, and histopathological findings[10,11]. The presence of poikiloderma vasculare atrophicans on histopathology further supports the diagnosis. Long-term follow-up is essential due to the documented association of RTS with squamous cell carcinoma, basal cell carcinoma, and osteosarcoma[12]. Reporting such cases enhances awareness and contributes to improved diagnosis and management strategies.

Conclusions

This case highlights the classical clinical evolution and histopathological features of Rothmund–Thomson syndrome. Early diagnosis, strict photoprotection, genetic counselling, and lifelong multidisciplinary follow-up are crucial to reduce morbidity and improve patient outcomes. Reporting rare cases such as this strengthens existing literature and facilitates timely recognition of this uncommon genodermatosis.

References

1. Vennos EM, James WD. Rothmund–Thomson syndrome. Dermatol Clin. 1995;13(1):143-150.
2. Rothmund A. Über Cataracten in Verbindung mit einer eigentümlichen Hautdegeneration. Albrecht von Graefes Arch Ophthalmol. 1868;14:159-182.
3. Kitao S, Shimamoto A, Goto M, et al. Mutations in RECQL4 cause a subset of cases of Rothmund–Thomson syndrome. Nat Genet. 1999;22:82-84.
4. Siitonen HA, Kopra O, Kääriäinen H, et al. Molecular defect of RAPADILINO syndrome expands the phenotype spectrum of RECQL diseases. Hum Mol Genet. 2003;12:2837-2844.
5. Larizza L, Roversi G, Volpi L. Rothmund–Thomson syndrome. Orphanet J Rare Dis. 2010;5:2.

6. Wang LL. Clinical manifestations and genetic basis of Rothmund–Thomson syndrome. Clin Genet. 2003;63:1-7.
7. Inamadar AC, Palit A, Athalikar SB, et al. Rothmund–Thomson syndrome: Report of three cases. Indian J Dermatol Venereol Leprol. 2003; 69: 67-69.
8. Hallman N, Patiala R. Congenital poikiloderma atrophicans vasculare in a mother and her son. Acta Derm Venereol. 1951;31:401-406.
9. Wang LL, Levy ML, Lewis RA, et al. Clinical manifestations in a cohort of patients with Rothmund–Thomson syndrome. Am J Med Genet. 2001;102:11-17.
10. Lehmann AR, McGibbon D, Stefanini M. Xeroderma pigmentosum. Orphanet J Rare Dis. 2011;6:70.
11. Kindler T. Congenital poikiloderma with traumatic blistering and progressive cutaneous atrophy. Br J Dermatol. 1954;66:104-111.
12. Wang LL, Gannavarapu A, Kozinetz CA, et al. Association between osteosarcoma and Rothmund–Thomson syndrome. J Natl Cancer Inst. 2001; 93:435-441.

Abbreviations

Rothmund–Thomson syndrome (RTS)

RecQ-like helicase 4 (RECQL4)

Legend Figures

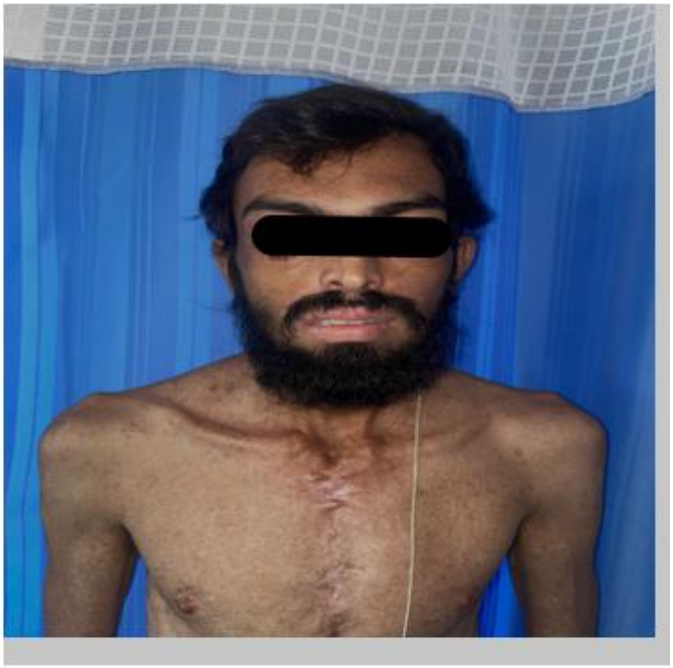

Figure 1: Typical birdlike facies that is prominent forhead & beaked nose

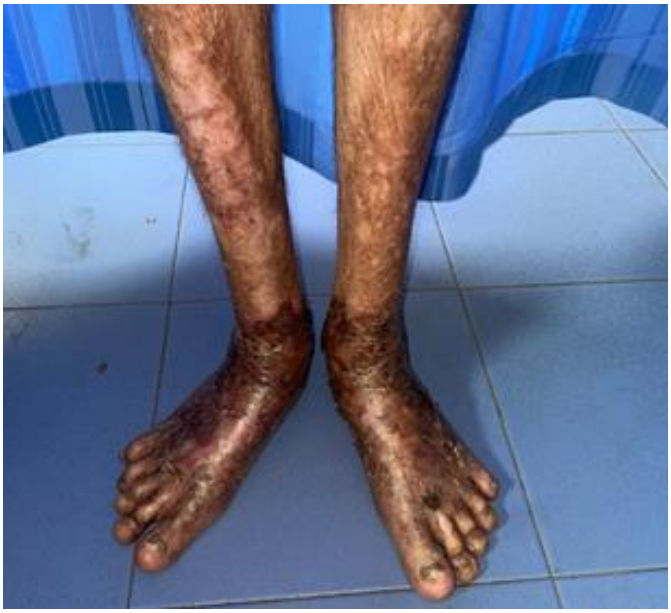

Figure 2: Well defined erythematous to hyper pigmented plaque with exfoliation & erosion (+) over bilateral foot. Multiple ill to well defined atrophic plaque present over the bilateral anterior aspect of leg. Nails – dystrophic nails, subungual hyperkeratosis and distal onycholysis.

Figure 3: Ill defined erythematous plaque with exfoliation present over dorsum of bilateral hands. Presence of marked atrophy.

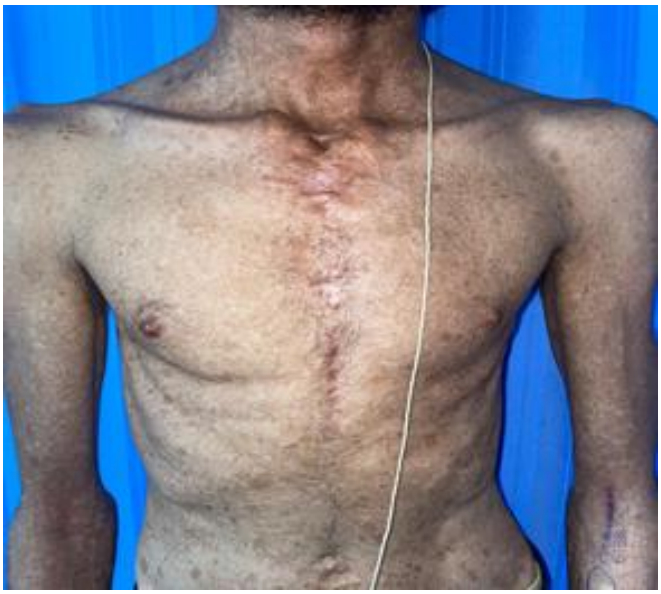

Figure 4: Multiple discrete hyper pigmented & hypo pigmented macules & patches + over the trunk, upper limb and lower limb.