ROTHMUND THOMSON SYNDROME

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SUMMARY

A-7-year-old female child presented with mottled hypopigmented and telangiectatic lesions over the face, arms, legs, and buttocks. The child is short statured with very short thumbs. The hair is sparse, short, and thin with loss of eye lashes and eye brows. There is no cataract or other extracutaneous features. To the best of our knowledge, this is the first case of R.T.S reported in Libya. Our patient was diagnosed on clinical features and histopathological findings.

INTRODUCTION

Rothmund Thomson syndrome (RTS) is a genetic disorder usually presents in early life as poikiloderma and a variety of extra cutaneous defects including cataract and skeletal abnormalities,1,2 photosensitivity, hypogonadism, and defective dentition.

Case Report

A. A. is a female Libyan child referred to us for the first time at the age of one year from the pediatrician as a case of atopic infantile eczema that started at the age of 3 months. The baby was born after uneventful full term pregnancy. There was no family history of a similar illness. However, the patient had a sister who died at the age of 8 months because of congenital anomalies in the heart. Patient's parents are first degree cousins.

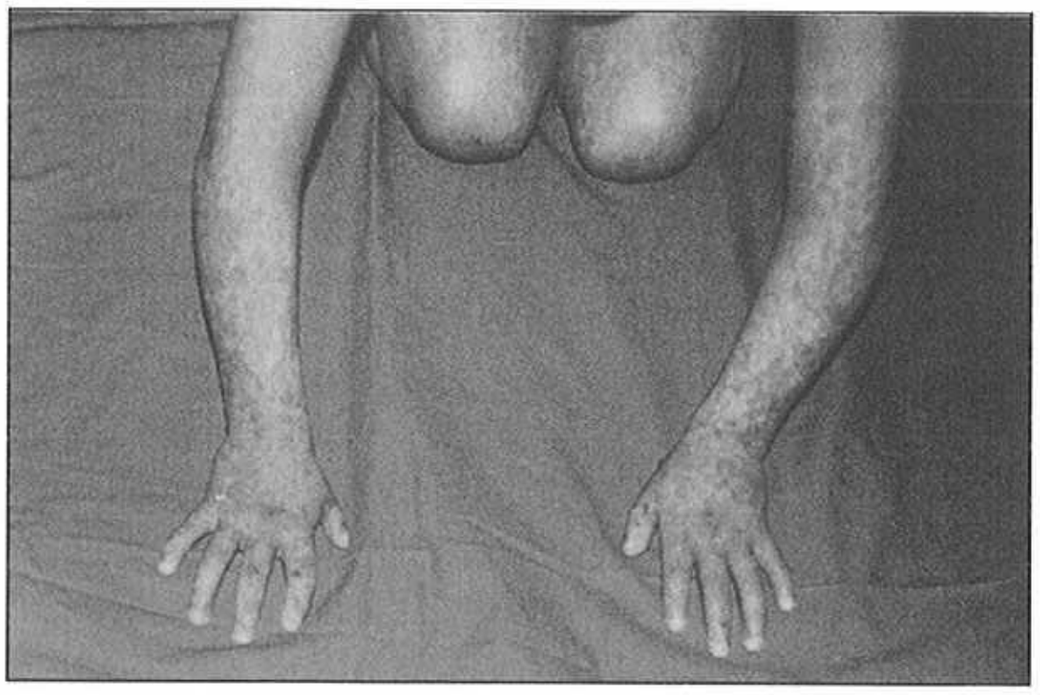

Figure 1: Rothmund Thomson syndrome. Very short thumbs.

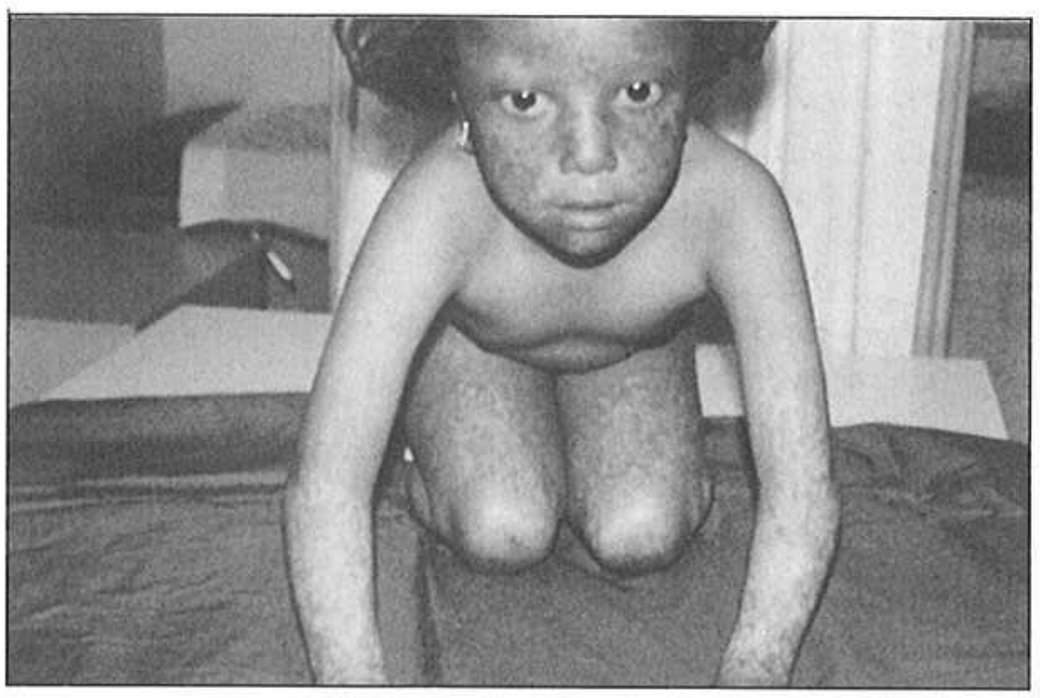

Figure 2: Rothmund Thomson syndrome. Absence of eye brows and lashes.

At the time of presentation, there was macular erythematous telangiectatic and pigmented lesions over cheeks and forehead, back, and buttocks. The child did not show up until the age of 7 years. Then represented to us at the age of 7 years with the same type of lesions extending to the arms, legs, and buttocks. In addition, the child developed papular warty lesions over dorsa of hands and feet. She was small for her age with very short thumbs (Figure 1). Her motor and mental developments were normal. There were no cataracts, and the hair appeared short and thin. Both eye brows and lashes were absent (Figure 2).

Skin biopsy showed thinned epidermis with hydropic degeneration of the basal layer with histiocytic lymphocytic infiltration in the upper dermis.

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DISCUSSION

Poikiloderma congenitale (R.T.S.) is inherited in an autosomal recessive pattern. It appears few months after birth with erythema on the face and subsequently extends to the dorsa of hands and feet and occasionally to the arms, and legs, and buttocks[1,2,3,4]. Palmoplantar hyperkeratosis and calcinosis cutis are associated features[5]. The hair is sparse and nail dystrophy is present in 25% of the patients[4,6]. There is increased sensitivity to sun light reported in some patients, and development of skin tumors has been also reported[5-7].

The extracutaneous features of this syndrome include zonular cataracts in about 50% of patients[2,6,8] which usually occurs before the age of 7, and skeletal abnormalities with small stature, saddle nose, short hands, joint sublaxation, kyphoscoliosis, and cysts in the long bones[1,6,9]. Metaphyseal chondrodysplasia and osteogenic sarcoma has been also reported in some cases[10,11,12].

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