Case report

Rothmund–Thomson syndrome in a young man without cataract involvement

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Introduction

Rothmund–Thomson syndrome is a rare disorder that is inherited as an autosomal recessive trait occurring predominantly in females at the age of 3–6 months or later [*1*]. It is characterized by light sensitivity and some degrees of poikiloderma. The disease has a worldwide distribution. One review of the literature showed 200 cases up to 1992 [*2*].

Rothmund–Thomson syndrome was probably first described by Rothmund, an ophthalmologist, in 1868. All reported cases suffered from premature cataracts [*3*]. Later, Thomson described sisters with similar skin changes in 1923 and subsequently the disorder was labelled as poikiloderma congenita [*4*]. Cataracts were not a fundamental finding in them. Taylor reviewed all the literature and proposed the eponym of Rothmund–Thomson syndrome [*5*].

We report a case of the syndrome in a young man without any problems with vision or premature cataracts or malignant transformation. Facial photosensitivity and involvement of his skin, such as sparse scalp and eyebrow hair, with involvement of the fingernails were also noted. We review some of the relevant literature.

Case report

An 18-year-old man was seen with photosensitivity of the face, neck and ears, with warty papules on his extremities. A bird-like face and poikilodermic skin were prominent signs in this case. In the physical examination, he was slightly short in stature, but small hands and feet were noted.

Pigmentation, telangiectasia and reticulate erythema were seen all over the face except on the upper lids, under the nose, lower lip and dorsum of both ears. Erythema had appeared when he was 6 months old. The eyebrows were thin with loss of bilateral end parts. Hypo- and hyperpigmentation areas, with partial plaques of keratosis pilaris, were seen on the neck, trunk and distal parts of the limbs. Bilateral partial ill defined brown scaly plaques were found in both axillae. Warty hyperkeratotic papules were seen on the palms, soles, and the back of his hands and feet. Papyrus scars were present on his knees.

He had a history of right forearm fracture at the age of 16 years that fused after 2 years. No history of ocular problems or convulsion was reported. Genital examination revealed normal development, and hypopigmented macules were seen on the

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penile shaft. His fingernails were thin, but mucous membranes were normal in appearance except for mild gingivitis.

In the laboratory evaluation, fasting blood sugar, complete blood cell count, and liver and renal function tests were normal. The serum levels of sodium, calcium and phosphorus were normal. The serum alkaline phosphatase level was raised at 368 IU/L (normal range 100–290 IU/L).

Radiographic reports of the pelvis, hands, skull, chest, forearms and the lower extremities were normal, without any patchy sclerosis, cystic changes or osteolytic lesions except mild osteoporosis in the bones of the right hand.

Discussion

Rothmund–Thomson syndrome is a rare, inherited disease [*1*]. The disease is an autosomal recessive disorder and a worldwide review of literature in 1992 showed that 200 cases had been published up to that time [*2*].

Heterozygous carriers are virtually normal but may be identifiable by a minor sign such as light sensitivity. The parents of our case suffer from mild photosensitivity without any other signs of the syndrome. The genodermatosis of Rothmund–Thomson syndrome can be diagnosed by clinical findings such as short stature, especially in the limbs, light sensitivity and poikiloderma. Children are usually normal at birth and have minimal findings in the first 3 months of life [*6*]. Cataract was not seen in this case. Bilateral cataracts can develop between the fourth and seventh year, usually in about 40% of reported cases, but they are more frequent in some families than in others [*2,5*].

The diagnosis of Rothmund–Thomson syndrome is made on clinical grounds as no consistent laboratory test has been identified [*7*]. The essential features in the differential diagnosis are the age of onset, the distribution of the lesions, and the combination of atrophy, telangiectasia and mottled pigmentation, most intense on light exposed skin but not necessarily confined to it [*8*]. Short stature with photosensitivity and radiodermatitis of the facial skin were seen; skin signs such as loss of scalp and eyebrow hair with nail involvement were seen in this case. Scalp hair is often sparse and fine, and may be absent. Eyebrows, eyelashes, and pubic and axillary hairs are often sparse or absent. Nails are normal, or small and dystrophic.

Teeth are often normal, but microdontia and early caries have been reported [*3,7*]. Short limbs without any malignant transformation were prominent features in our case. Squamous cell carcinoma may develop in the keratosis or in the surrounding atrophic skin. After cutaneous epithelioma, osteosarcoma is the most frequent malignancy [*1*].

Thus patients with Rothmund–Thomson syndrome need a careful survey [*3*]. Physical development is frequently retarded; most patients are of small stature and some are dwarfs. The dwarfism is proportionate, with slender delicate limbs, small hands and feet, and short stubby fingers. The skull may be small and bird like, sometimes with a saddle nose. Aminoaciduria has occasionally been reported but has not been a consistent finding [*1*]. One report described osteogenesis imperfecta in a patient [*9*].

Hypogonadism and the incidence of hyperparathyroidism also appear to be increased [*10*]. Mental development is usually normal, but may be retarded. Neurological examination of our case showed no abnormality, and genital organs also had

normal development, but hypopigmented macules were detected on his penile shaft. In excised biopsy specimens, flattening of the epidermis with dermoepidermal junction oedema were seen. Acrogeria, Kindler syndrome, dyskeratosis congenita, xeroderma pigmentosa, and Bloom and Cockayne syndromes must be differentiated from Rothmund–Thomson syndrome [*1*].

Light sensitivity is a common feature in patients with Rothmund–Thomson syndrome [*1,2,3,8*]. High sun protective sunscreen is thus recommended for all patients with this syndrome.

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