Rothmund-Thomson Syndrome with Malignant Fibrous Histiocytoma: Report of a Case and Review of Literature

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Received: July 12, 2009
Accepted: April 28, 2010

Abstract

Rothmund–Thomson syndrome is a rare autosomal recessive genodermatosis characterized by early onset of poikiloderma and several other cutaneous and organ involvements. We are going to report a 14-year-old girl who has been diagnosed with Rothmund–Thomson syndrome since she was 3 years old and has been suffering from pain and swelling of the right elbow and forearm for about 6 months. There are few previous reports on Rothmund–Thomson syndrome associated with malignant fibrous histiocytoma. We decided to report this case as another supporting document for this association.
(Iran J Dermatol 2010;13: 27-29)

Keywords: poikiloderma, malignant fibrous histocytoma, Rothmund–Thomson syndrome

Case Report

Our case is a 14-year-old girl who is a known case of Rothmund–Thomson syndrome from the age two. She was admitted because of a swelled tender erythematous mass on her right elbow that had appeared one year ago and enlarged gradually.

On physical examination, she had a bird-like appearance. Scalp hair, eyebrows and lashes was sparse (Figure1).

Conjunctiva was pale. The patient had angular cheilitis and her tongue and buccal mocousa was pigmented.The first metacarps and metatarses were absent and forearms were shortened. There were ulcers and warty lesions on her digits, palms and soles (Figure2).

Her skin was poikilodermatous and pigmentation, depigmentation, atrophy and telangiectasia were seen on her face, trunk and limbs.

Physical growth was stunted but the patient was mentally normal. There were hypogonadism and signs of puberty such as pubic and axillary hairs and menstruation were not observed. Nails and teeth were normal and other systems had no problems.

Blood count showed mild normochromic and normocytic anemia. Blood biochemistry was normal. Chest X-Ray showed numerous opacities suggestive of lung metastasis. X ray study of the affected limb revealed soft tissue swelling and a pathological fracture. Magnetic Resonance Imaging showed a large mass with expansion and destruction of the proximal third of ulna causing widening with necrotic areas extended to the arthicular surface of the elbow without joint effusion, consistent with a mostly benign process, specially chondral, in origin (Figure 3).

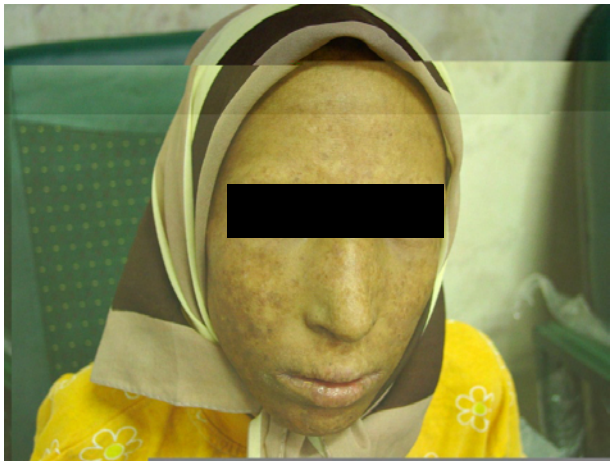

Figure 1. *Bird-like appearance. Scalp hair, eyebrows and lashes are sparse.*

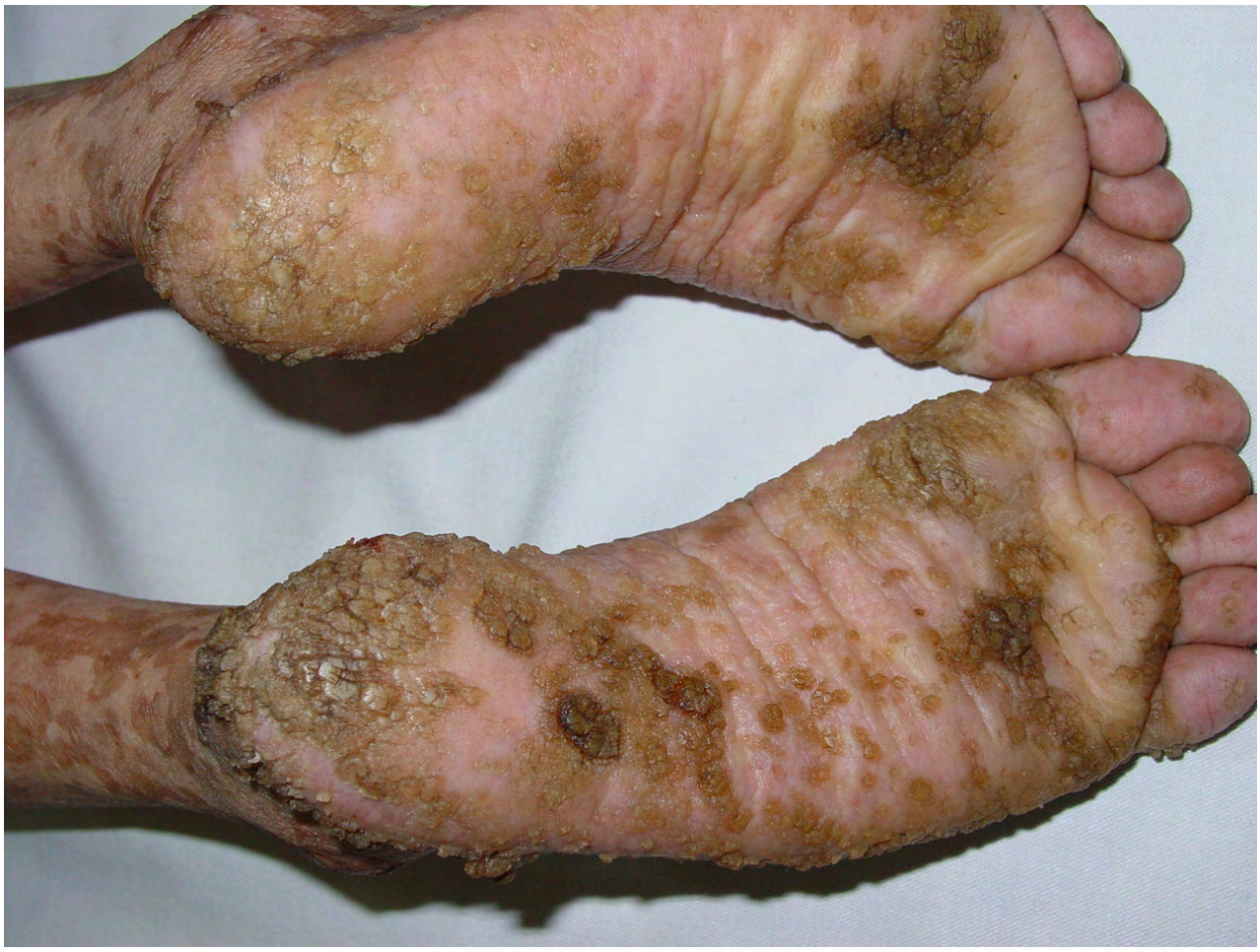

Figure2. *Ulcers and warty lesions on digits and soles*

Histopathology revealed a malignant neoplastic tissue mostly with a storiform pattern composed of pleomorphic spindle cells with a high N/C ratio, vesicular nuclei and prominent nucleoulei, associated with multinucleated giant tumoral cells. There were areas of necrosis and foci of hyalinized matrix, especially in a storiform pattern (Figure 4). No malignant osteoid formation was seen. Thus, histology was consistent with high grade sarcoma with malignant fibrous histiocytoma.

The patient was referred to the orthopedic department and despite chemotherapy and amputation of the involved elbow, the patient died after 2 months.

Discussion

Rothmund–Thomson syndrome is a rare autosomal recessive disorder occurring predominantly in females [1]. There have been several reports of various karyotypic abnormalities including trisomy 8 mosaism, reduced DNA repair capacity and increased sensitivity to UVC [2,3]. The earliest lesions develop between three and six months of age.

Plaques of erythema and oedema are succeeded by poikiloderma. Dull brown pigmentation develops later than atrophy and telangiectasia. Cheeks are first and most severely involved but the forehead, chin and ears seldom escape. The hands, forearms and lower legs are next affected and the buttocks and thighs are frequently involved. Light sensitivity tend to diminish after early childhood, but may persist into adult life [4-6].

In many cases, keratoses develop on the exposed skin from adolescence onward. Squamous cell carcinoma may develop in the keratoses or in the surrounding atrophic skin and large warty keratoses of hands, wrists, feet and ankles may restrict patients' activities [7-8]. Scalp hair is often sparse and fine or may be absent. Eyebrows and lashes, pubic and axillary hairs are often sparse or absent. Nails are normal or small and dystrophic. Teeth are often normal. Most patients are of short stature. The skull may be small and facial features are bird-like.

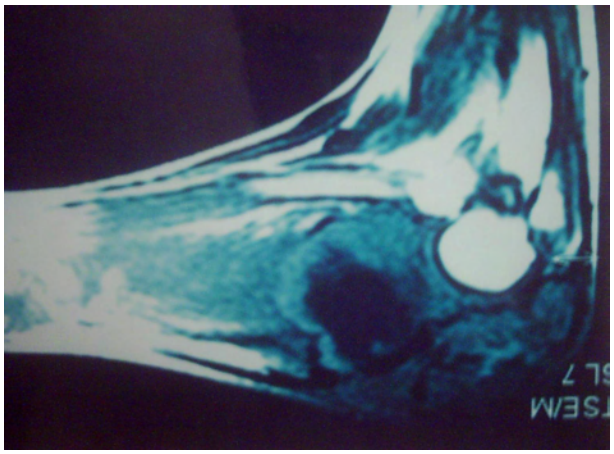

Figure 3. *MR Imaging shows a mass with expansion and destruction of proximal third of ulna causing widening with necrotic area extended to the arthicular surface of the elbow without joint effusion.*

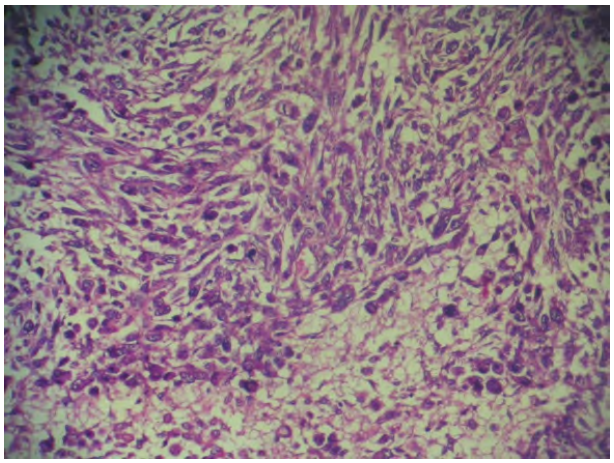

Figure 4. *Pleomorphic spindle cells with a of tumor high N/C ratio, vesicular nuclei and prominent nucleoulei and multinucleated giant tumoral cells . (H&E*40)*

Hypogonadism and hyperparathyroidism are frequent [9]. An association between. Skeletal abnormalities include a radial X-Ray defect which may present with thumb hypoplasia with an abnormal radial head or complete absence of radius [10]. There is a recognized risk of osteosarcoma, especially in the bones of the lower limbs, which can present in childhood [11,12]. Moreover, there is few report of its association with malignant eccrine poroma and malignant fibrous histiocytoma [13-14].

Individuals with Rothmund–Thomson syndrome are usually of normal intelligence. Life expectancy is normal but depends on the development of associated malignancies. In this report , a high grade sarcoma were found in association with Rothmund–Thomson syndrome that led to patient death . Therefore, it could recommended to keep in mind the considerable possibility of malignant neoplasm in the setting of Rothmund–Thomson syndrome that may be very helpful to diagnose them earlier and prolong the patients' survival.

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