Clinical Approach to a child with Poikiloderma: A case report

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Abstract

Rothmund-Thomson Syndrome is a rare autosomal recessive condition presenting usually in infancy that can be diagnosed based on time of onset, spreading and appearance of the poikiloderma.The purpose of reporting this case is to highlight the clinical approach to a child who presents with the features of poikiloderma.

Introduction

Rothmund-Thomson Syndrome(RTS) is a rare autosomal recessive disorder, usually resulting from mutation in RecQL helicase gene that functions during DNA repair.[1] It presents in infancy with erythematous macule or plaque often associated with blisters and edema that progress to develop poikiloderma initially involving face and later to other sun-exposed sites. The characteristic onset and progression of poikiloderma helps to differentiate it from other causes of early onset poikiloderma.[2] Further it is characterized by heterogenous features like short stature, skeletal abnormalities, cataract, premature ageing, abnormalities of hair, nail and teeth.[2] It is associated with increased risk of osteosarcoma in childhood, cutaneous epithelial neoplasm and hematological malignancies in adults.[2]

Case Report

A 4-year-old girl presented with rash on her face that was noticed by her mother since 1 month following birth. She was 3rdof the 5 children born at full term via normal vaginal delivery at home to consanguineous parents. Initially, the mother noticed erythematous macules and plaques on bilateral cheek that gradually developed areas of hypopigmentation and telangiectasia within 6 months (Fig 1a). Lesions with similar morphology and progression appeared to involve shoulder (Fig 1b) followed by V-area of neck after a year that was aggravated on exposure to sunlight. There was no history of hair loss, recurrent respiratory infection, oral ulcer, feeding problems, dental, ocular or neurological symptoms. On family history (as presented by pedigree in Fig 2) there was demise of a 4th child at birth and history of similar skin lesion on 8 months old 5thchild who developed the lesions at 2 months following birth (Fig. 1a).

On cutaneous examination, there were multiple ill-defined erythematous plaques on bilateral cheek, dorsum of nose, V-area of neck and right shoulder with areas of dyspigmentation, atrophy and telangiectasia. Height of the child was less than 3rd percentile for age and weight was at 10th percentile for age (as per WHO growth chart). There was no abnormalities detected on systemic examination. Baseline complete blood count, Anti-nuclear antibody, Comprehensive metabolic panel and X-ray of bilateral hand was normal. Karyotype analysis and gene sequencing was not done due to unavailability. RTS was diagnosed, and the patient was advised for sun-protective behaviors and annual evaluation for the eyes, skin and bone.

DISCUSSION

RTS presents at three to six months' age with erythematous macule or plaque over face, sometimes associated with blisters and edema which gradually progress to involve extremities and gluteal regions sparing trunk and abdomen. [1] Gradually, poikiloderma develops over the initial lesion that persist throughout the life.

In few of the cases, there might be systemic involvement that includes dental abnormalities (microdontia, conical teeth, frequent caries, loss of teeth), ocular involvement (bilateral subscapular cataract) and bone abnormalities (absent or deformed radii, deformed ulnar bone, short hands and feet, aplasia of thumb, delayed bone age and osteoporosis)[1]. There is increased risk of osteosarcoma in childhood and epithelial neoplasms in adult.[2] Patients have a normal life span unless associated with malignancy. Strict photoprotection and routine screening for malignancy are the mainstay of management.

Patients in our case had characteristic rash of RTS developing at one month's age, initially acute features of rash which developed into poikiloderma that was persistent with typical distribution pattern and had delayed growth. However other systemic features of RTS were absent.

Detailed history and careful examination may aid in differentiating other causes of childhood poikiloderma. Acrogeria manifests at the time of birth or shortly afterwards with poikiloderma limited to acral parts[3]. Hereditary sclerosing poikiloderma presents in childhood with generalized poikiloderma with accentuation in flexures and extensor surfaces along with sclerodermatous plaques on palms and soles[4]. Dyskeratosis congenita is characterized by triad of severe nail involvement, poikiloderma conspicuous over neck and leucoplakia at a later age[5]. Kindler syndrome is characterized by poikiloderma that develops at the age of 2 to 3 years in photo-exposed sites along with acral blisters and mucosal stenosis[2]. Patients with Cockayne's syndrome develops poikiloderma that spreads centripetally starting from distal limbs along with typical facies, limb abnormalities, wasting and neurological manifestations[6]. Rare disorders like Bloom's syndrome, Fanconi's anemia and Ataxia telangiectasia present with prominent telangiectasia and characteristic facies[2].

Reference:

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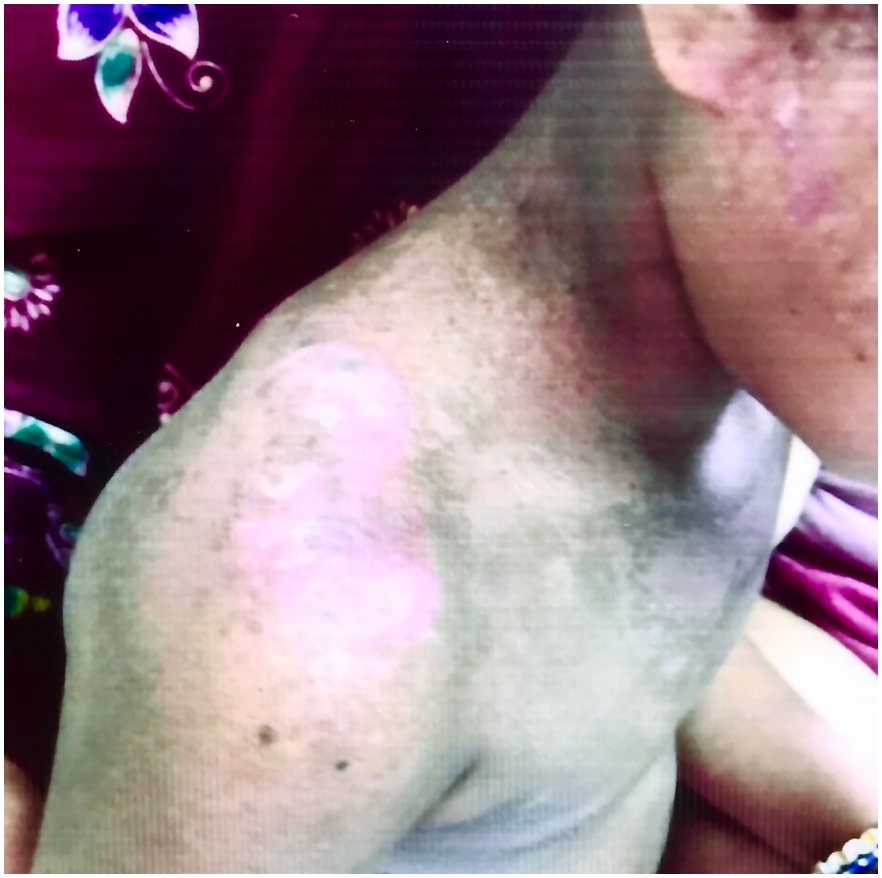
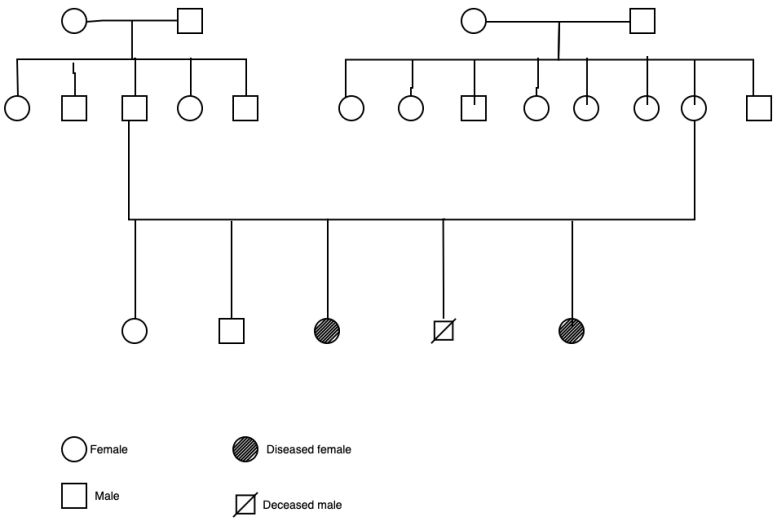
[Figure 1a showing erythematous plaques on bilateral cheek, bridge of nose, V-area of neck in both the siblings]



[Fig. 1 b showing erythematous plaques along with atrophy, dyspigmentation and telangiectasia on right shoulder]



[Fig. 2 showing pedigree of the child]