OPEN ACCESS
Saudi Journal of Medicine
Abbreviated Key Title: Saudi J Med
ISSN 2518-3389 (Print) |ISSN 2518-3397 (Online)
Scholars Middle East Publishers, Dubai, United Arab Emirates
Journal homepage: https://saudijournals.com/journal/sjm/home

Case Report

The Rothmund Thomson Syndrome

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| **Received:** 16.02.2019 | **Accepted:** 26.01.2019 | **Published:** 28.02.2019
DOI: 10.36348/sjm.2019.v04i02.011

Abstract

We report the case of a girl of 2 years and a half, without a notion of consanguinity, admitted for erythema of both cheeks which appeared 40 days after birth. In view of the notion of photosensitivity and the appearance of poikiloderma found in the clinical examination, the diagnosis of Rothmund Thomson syndrome was retained. Rothmund Thomson syndrome is a rare genetic disorder with autosomal recessive inheritance. It is characterized by the early onset of poikiloderma associated with various cutaneous and extra-cutaneous abnormalities with an abnormally high incidence of cutaneous cancers. There is no treatment of the disease, the care is limited to its consequences.

Keywords: Rothmund Thomson, genetic disease, poikiloderma, skin cancer, osteosarcoma, RECQL4 gene.

INTRODUCTION

Described for the first time in 1868, Rothmund Thomson syndrome is a rare genetic disease with autosomal recessive inheritance, characterized by the early onset of poikiloderma and may affect several parts of the body.

OBSERVATIONS

We report the case of a girl of 2 years and a half, 4 th of her four siblings, with a similar case of the older brother aged 14, without the notion of consanguinity in the parents.

Admitted for erythema of both cheeks, appeared 40 days after birth, and treated as atopic dermatitis for a period of two years without any improvement.

The examination found an erythema of the cheeks with poikilodermia appearance, and the notion of photosensitivity also affecting the backs of the hands and forearms. There was no notion of photophobia, Phannian disorders, or dentition (Figure 1 & 2).

In view of this aspect of recent poikilodermie of the photo-exposed zones, the diagnosis of Rothmund Thomson syndrome was retained.

The therapeutic decision was a photo-eviction and photo-protection of clothing and chemicals with surveillance.

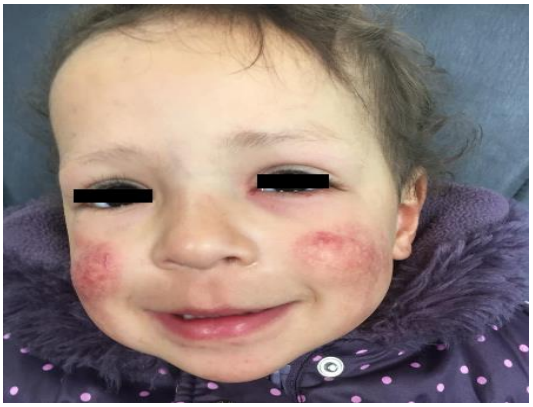

Fig-1: Poikiloderma cheeks (front view)

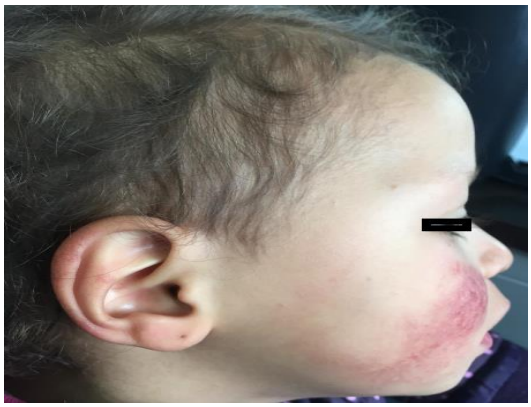

Fig-2: Poikiloderma cheeks (side view)

DISCUSSION

Rothmund-Thomson syndrome is a rare genetic disease whose prevalence is unknown [1].

It is very polymorphic clinically, and is characterized by the early onset of poikilodermie associated with various skin, phanerial, bone, ocular, endocrine and dental abnormalities, with abnormally high incidence of skin and bone cancers; 5% and 30% respectively [2].

Mutations in the RECQL4 gene have been described in 2/3 of the cases and would be correlated with an increased risk of osteosarcoma formation. In the other 1/3 of the cases, the gene in question has not yet been identified [1, 3].

Because of their multiplicity, the management of these skin cancers occurring in these patients proves difficult, some authors have demonstrated the interest of dynamic phototherapy in the management of these cutaneous tumors, and which could be an excellent alternative in the tolerance plan [4].

The prognosis is variable: life expectancy is normal if there is no cancer, while the evolution of patients with malignant diseases depends on the quality and frequency of cancer screening and treatment.

CONCLUSION

The diagnosis of Rothmund-Thomson syndrome is clinically oriented and confirmed by molecular study.

There is no treatment of the disease; the care is limited to its consequences. Genetic counseling is offered to patients and their families.

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