

The Rothmund Thomson Syndrome

Abdessamad Sakkah^{1*}, Jalal El Benaye², Ihssane Hallab¹, Youness El Khachine¹, Abderrazak Jakar³, Mohamed El Haouri⁴

¹Resident, Department of Dermatology, Moulay Ismail Military Hospital, Meknes, Morocco

²Associate Professor, Department of Dermatology, Moulay Ismail Military Hospital, Meknes, Morocco

³Dermatology Specialist, Moulay Ismail Military Hospital, Meknes, Morocco

⁴Professor, Department of Dermatology, Moulay Ismail Military Hospital, Meknes, Morocco

*Corresponding author: Abdessamad Sakkah

| Received: 16.02.2019 | Accepted: 26.01.2019 | Published: 28.02.2019

DOI: [10.21276/sjm.2019.4.2.11](https://doi.org/10.21276/sjm.2019.4.2.11)

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.

Copyright @ 2019: This is an open-access article distributed under the terms of the Creative Commons Attribution license which permits unrestricted use, distribution, and reproduction in any medium for non-commercial use (NonCommercial, or CC-BY-NC) provided the original author and source are credited.

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 poikiloderma 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.

REFERENCES

1. Elhadri, S., Elguendouz, F., Chahbi, Z., & Baizri, H. (2015, September). Syndrome de Rothmund Thomson avec retard statural par rupture de la tige pituitaire: à propos d'un cas. In *Annales d'Endocrinologie*, 76(4), 405. Elsevier Masson.
2. Sahel, H., Zemouri, F. Z., Otsmane, F., & Bouadjar, B. (2013, April). Syndrome de Rothmund Thomson familial chez trois frères et sœurs dont deux avec ostéosarcome. In *Annales de Dermatologie et de Vénérologie*, 140(1), s45-s46. Elsevier Masson.
3. Denis-Thely, L., Bost, M., Reymond, J. L., Maitre, S., Touraine, R., & Cambazard, F. (2005). Syndrome de Rothmund-Thomson et déficit en hormone de croissance. In *Annales De Dermatologie Et De Venereologie*, 132(3), 168-169. Sppif-Masson Service.
4. Modiano, P., Wantz, M., Durand, F., Van Lerberghe, L., Lasek, A., & Lebas, D. (2016, December). Intérêt de la photothérapie dynamique dans le syndrome de Rothmund-Thomson. In *Annales de Dermatologie et de Vénérologie*, 143(12), S383-S384. Elsevier Masson.