Case report

DERMABRASION IN A GIRL WITH CONGENITAL POIKILODERMA (Syndrome Rothmund-Thomson)

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SUMMARY

A case of congenital poikiloderma in a 6-year-old girl is reported. The lesions on the face, ears and gluteal area appeared during her first year. Pathohistology of a biopsy taken from the face revealed an atrophic epidermis, dilated capillaries in the papillary dermis and a mild perivascular infiltrate composed of lymphocytes, histiocytes and a few melanophages. A test-dermabrasion was carried out in an area of fully developed lesions on the face. Six months later no lesions could be observed in the dermabraded area, a second biopsy displayed a normal histologic structure of the skin including hair follicules, sebaceous and sweat glands.

In view of such observations the author raises the question at what time in the embrional development do the malformations start in this condition.

KEY WORDS

congenital poikiloderma, syndrome Rothmund-Thomson, dermabrasion, 6-year-old girl

INTRODUCTION

Poikilodermas may be considered as a special group inside the broader concept of congenital atrophies. They appear soon after birth or during the early infancy, the main symptoms being skin atrophy, hyper- and hypopigmentation, teleangiectasia and occasionally also a pityriasiform scaling. Congenital poikiloderma or Thomson's syndrome was described in 1923 and later in 1936 (1). It is inherited in an autosomal recessive mode, the familial occurrence being rare, consanguinity of the parents is mostly not detectable. The sites of predilection are the face, ears, extremities and the gluteal region. The lesions usually start to appear during the first year of life as a patchy erythema of the face. Hypotrichosis, a triangular shape of the face with a high front, hypertelorism and a small chin may also be expressed.

Certain authors believe that the syndromes of Thomson and Rothmund represent the same pathologic entity, although some signs may be missing (2). Rothmund who described the symptom in 1868 mentioned additionally to the skin symptoms, consanguinity of parents, a juvenile cataract, shortened extremities, photosensitivity, hypoplasia of the genitals as well as occurrence of malignancies (1,3,4,5,6,7).

Treatment includes the avoidance of skin irritants, photoprotection and application of neutral creams.

CASE REPORT

The patient was a 6-year-old girl. Her family history was uneventful except that her father's mother and grandmother suffered from diabetes. Consanguinity of her parents and grandparents was excluded.

The delivery was normal, her weight at birth was 2730 grams and length 49 centimeters. She was not stilled and occasionally suffered from bronchitis.

At the age of 7 months her mother noticed reddened and swollen cheeks, later on dilated capillaries and an irregular pigmentation became apparent. Two years later teleangiectatic vessels and erythema were observed also on the lower ear-lobes and in the gluteal region. On the face striae distensae-like lesions were also present. She was treated with various topical preparations however with poor result. Every exposure to the sun was followed by an impairment of her skin condition.

Her physical and psychical development was in accordance with her age. Except for caries of her anterior teeth and slightly enlarged tonsils her general condition was considered to be normal.

The skin of both cheeks was moderately atrophic, partially similar to striae distensae and partially in form of small spots. Numerous dilatated capillaries were visible. In the skin of the lower ear-lobes and of the gluteal region no atrophies but dilated capillaries in a net-like fashion were expressed.

Routine laboratory tests were within the normal limits except for increased values of alkaline phosphatase which findings are normal at this age.

Histopathologic analysis of a biopsy revealed an atrophic epidermis, dilated capillaries in the papillary dermis as well as a mild inflammatory infiltrate including a few melanophages.

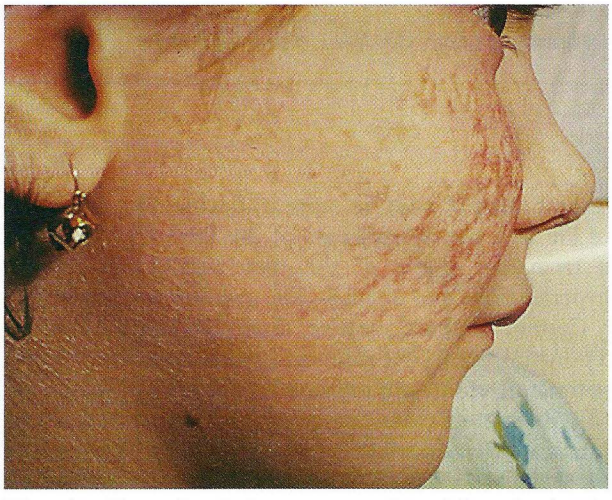

Fig. 1. The abraded area on the right cheek six months after dermabrasion.

TREATMENT

As the topical treatment with various ointments was unsuccessful, the decision was made to carry out a test-dermabrasion in local anesthesia with xylocain. An area of 3 x 4 cm on her right cheek was abraded. After the bleeding was stopped the eroded surface was covered with an antibiotic powder. The next day a crust was formed which resolved after 10 days, resulting in a mildly pinkish colored skin. Six months after the treatment the excellent result persisted (Fig. 1). A new biopsy revealed a normal structure of the epidermis and dermis.

A more extensive dermabrasion was proposed, unfortunately, adverse war events prevented the patient from undergoing further treatment.

DISCUSSION

The number of patients with the Rothmund-Thomson syndrome reported is to small to decide whether these are two separate nosologic entities or just the same, often oligosymptomatic condition. The girl under observation was not affected by cataract, dwarfism or other symptoms which are characteristics of the Rothmund syndrome. Only skin lesions of the face, ears and buttocks as well as a photosensitivity were noted. The causative pathogenetic mechanism remains so far unknown, an increased sensitivity to the effects of gamma rays by fibroblasts including a reduced synthesis of DNA, was reported (8).

In view of the extremely encouraging effects of dermabrasion in our case, this treatment modality deserves a short discussion. Only otherwise normal patients with just skin lesions should be selected for dermabrasion. The fact that the regeneration of skin in the abraded area starts from normal structures, e.g. sebaceous and sweat glands and hair follicules, poses the question at what time of the embryonal development do the abnormalities start to appear. It can be assumed that this happens at a later stage when the anlage of sebaceous and sweat glands are already expressed. Contrary to the effect of dermabrasion in congenital poikiloderma no such favorable

result was observed in ichthyoses. From these observations the author draws the conclusion that the skin lesions in congenital poikiloderma result from a deficiency in the embryonal development, whereas the skin lesions in ichthyosis are primarily genetically determined.

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