

POIKILODERMA CONGENITALE—AN EARLY CASE OF ROTHMUND-THOMSON'S SYNDROME

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Abstract. A case of congenital poikiloderma is presented. Two types of congenital poikiloderma can be differentiated on the basis of onset of symptoms and configuration of skin lesions, i.e. Rothmund-Thomson's type and Wodniansky's type. The case presented here is of the Rothmund-Thomson type, but showed only minor abnormalities apart from skin changes and hair loss. A slightly elevated concentration of lysine and cystine was found in the urine of our patient.

Key words: Poikiloderma congenitale; Rothmund-Thomson's syndrome; Congenital malformations

In its full development the Rothmund-Thomson's syndrome consists of poikilodermatous skin changes, bilateral cataracts, hair loss, bone defects, defects of the teeth and nails and disturbance in the hormonal balance.

The present case showed only minor abnormalities apart from the skin changes.

CASE HISTORY

The patient, a first child, was born after a full-term pregnancy. The birthweight was 2 550 g. Apart from emesis gravidarum treated with pyridoxine, phenobarbital and promazine, and oedema treated with chlorthiazide, there was no evidence of abnormalities during the pregnancy. The parents were not related and there were no further cases in the family.

The skin appeared normal at birth, but from the age of 3 months the patient developed skin changes beginning on the cheeks spreading to the ears, the forehead and the extremities. The patient was first seen at the age of 2½ and was further investigated at the age of three. During this time the skin changes had covered new areas (Fig. 1). Originally the changes were teleangiectasias, but at the latest visit to hospital some of the areas had changed character, becoming scaly, dry, and with loss of lanugo hair.

At present the patient has teleangiectasias on the cheeks (Fig. 2), forehead, ears, ulnar sides of arms, on the front of thighs and on the buttocks. On the arms, especially the distal parts, and on the ears, the changes now have the appearance of poikiloderma. The skin of the affected parts is dry and atrophic but shows no evidence of hyperkeratosis. There is

no lanugo hair on the affected parts, and small areas of alopecia have developed on the scalp. The teeth show evidence of heavy early caries. The ears have a slightly abnormal shape, with atrophy of the lower part of the helix. The nails are normal. No abnormalities have been found by X-ray examination of the total skeleton and no eye symptoms could be demonstrated. Height and weight at 2½ years were 14 kg and 89 cm, and at 3 years, 14 kg and 95 cm.

Laboratory examinations. Chromosomal analysis: normal (46XX). Urine examination for porphyrines, urobilinogen and porphobilinogen: negative. Serum electrophoresis: normal. Wassermann reaction: negative. Serum-concentrations of vitamin A and C: normal. The chromatographic examination of amino acids in the urine shows a slight elevation in the concentration of cystine and lysine.

Skin biopsy from the lower forearm showed atrophy of the epidermis, but no hyperkeratosis. The dermal connective tissue was somewhat sclerotic. There was some dilatation and an increase in the number of small vessels with slight perivascular oedema. There were a few perivascular lymphocytes and pigment-carrying histiocytes. The number of elastic fibres was considered normal. Immunofluorescence studies gave negative results.

COMMENTS

The present case demonstrates skin changes associated with the Rothmund-Thomson type of congenital poikiloderma (4, 6). The first lesions developed at the age of 3 months, with a steady progression visible during the observation period, which is in contrast to the Wodniansky-type where symptoms are present at birth (8). No sign of cataract has developed, though this symptom most often develops between the age of four and seven (3, 5). Apart from the skin lesions, minor abnormalities of the outer ear, alopecia and pronounced caries were noted.

The chromosomal analysis was normal, which is in accordance with previous investigation (2, 7). Increased light sensitivity has been reported in 35% of the published cases (5), but has until now

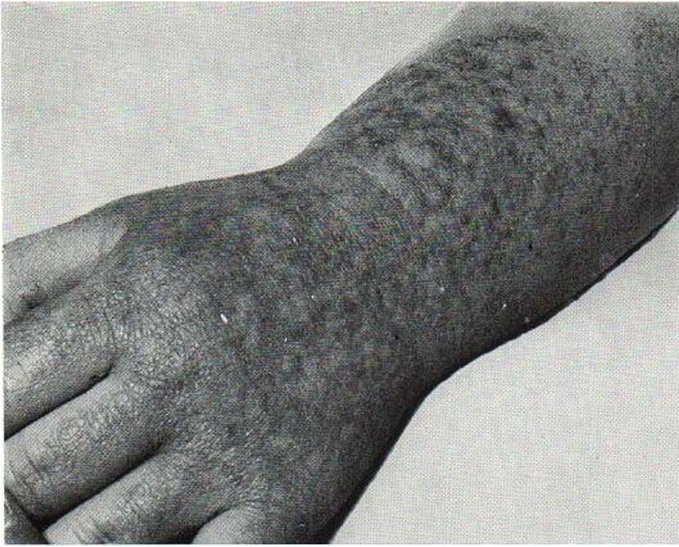


Fig. 1. Skin of forearm at the age of three.

not been found in our patient. The diagnosis of congenital poikiloderma is easy, due to the characteristic skin changes.

Subgrouping depends on the character of the skin anomaly and the associated defects in other organ systems (8). Our patient, we believe, belongs to the Rothmund-Thomson group on the basis of a combination of skin changes, hair loss and dental defects, together with a typical onset and progression.

The biochemical defects found in this patient, i.e. the slightly increased discharge of lysine and cystine, does not relate the patients symptoms to Hartnup's disease or homocystinuria (1).



Fig. 2. Face of patient at the age of three.

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