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Case report

Congenital poikiloderma with unusual hypopigmentation and acral blistering at birth

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Abstract

Congenital poikiloderma is an uncommon hereditary disorder. It has been reported in association with various syndromes. No case has previously been reported from this environment. We report a case of congenital poikiloderma in a two and a half year old female Nigerian associated with unusual generalised hypopigmentation and acral blisters at birth. The child subsequently developed macular hyperpigmentation on an erythematous background and atrophy of the skin. Although she had some features which were suggestive of Rothmund–Thomson syndrome (RTS), the presence of nypopigmentation at birth, along with acral blistering, was noted to be peculiar to this child. We, therefore, feel that this case presents a distinct variant of congenital poikiloderma that has not been described previously.

Keywords: Congenital poikiloderma; Hypopigmentation; Acral blistering

1. Introduction

Congenital poikiloderma is uncommon but has been described as part of distinct hereditary syndromes. We recently saw a two and a half year old Nigerian female with bizarre poikiloderma, i.e. hyperpigmentation, scarring, telangiectasia on hypopigmented skin that did not conform completely with any previously described syndromes associated with poikiloderma in the literature. Both her parents are dark skinned but she was hypopigmented (white) at birth and she also had blisters on both ankles and the upper back at birth. The blisters healed shortly after birth and never recurred. Three months later she developed poikiloderma that was progressive and became generalised involving most parts of the body but excluding the palms and soles. She had scanty scalp hair, eyebrows, and one of her nails was dystrophic. She was noticed to have some developmental delay. This appears to be an undescribed variant of congenital poikiloderma.

2. Case report

A two and a half year old girl was brought to the

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dermatology clinic with complaints of bizarre pigmentation. Both parents were Nigerians. The mother had had an uneventful pregnancy for 9 months with spontaneous vaginal delivery. At birth, the baby was described as white. She also had a few blisters and raw surfaces on both ankles and on the upper back. All other systems appeared normal. The weeping lesions healed within 4 weeks.

When the child was 3 months old, the mother noticed an unusual redness on her face, scalp and upper trunk. Subsequently the girl developed spotty brown pigmentation on the exposed parts of her body, and later, on the back, chest, and nappy areas. The mother noticed unusual scarring of the skin in the same areas. The child's hair was scanty and light brown in colour and the mother noticed some developmental delay.

She had neck control at 4 months, sat with support at 8 months, started crawling at 2 years of age, and at presentation was still unable to stand unaided. There was no history of seizures. There had been no undue sensitivity to sun exposure and no abnormal colour of her urine. There was no family history of a similar disorder or of any blistering illness. She was the second child of her parents.

On examination, there was a young girl who was small for her age, measuring 78 cm in length; she had a head circumference of 43 cm and weighed 8 kg. The

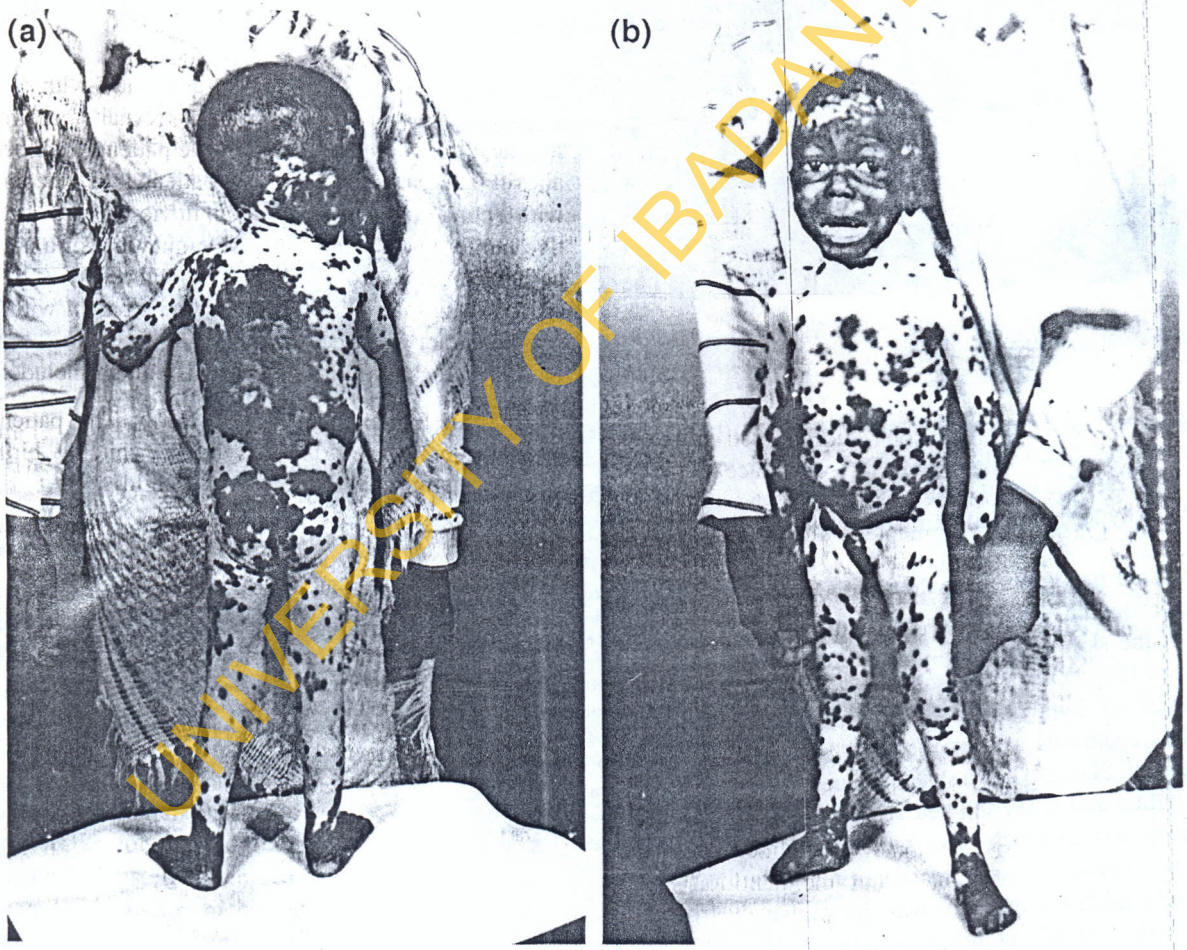

Fig. 1. (a) Macular hyperpigmentation on depigmented skin with scarring on the back and scalp. (b) Macular areas of hyperpigmentation on hypopigmented skin.

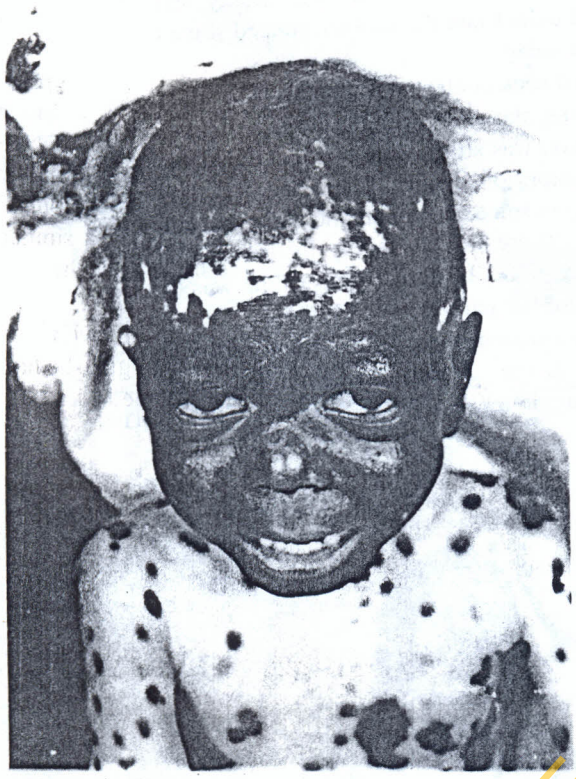

Fig. 2. Scanty hypopigmented hair and scarring on the scalp. There are scanty eyebrows and a saddle shaped nose.

main findings were cutaneous; the exposed parts of her body were pigmented, i.e. the scalp, face, dorsum of the hands, and feet. She had macular areas of hyperpigmentation on hypopigmented skin over the trunk and limbs.

There was some background erythema and telangiectasia visible on the hypopigmented skin. There was atrophy of the skin and marked scarring on the pigmented areas (Fig. 1a,b). The scalp was scarred with some areas of hypopigmentation and scanty brown hair. The eyebrows were scanty but the eyelashes were normal. She also had a saddle shaped nose (Fig. 2). The palms and soles showed some hyperlinearity but were soft to touch. The mucous membranes were not involved, and the dentition was normal. The right middle finger nail was dystrophic. There was no evidence of inflammation around the nail.

The central nervous system examination showed mild hypotonia in the upper limbs and delayed mental development; the mental age was 10 months with chronological age at 30 months. Ophthalmological examination revealed large blood vessels but no abnormal vasculature, and some areas of melanosis on both conjunctiva. There were no abnormalities in the cardiovascular, respiratory, gastrointestinal and genitourinary systems.

Histology of the skin taken from hypopigmented areas revealed atrophy of the epidermis with a mild round cell inflammatory infiltrate in the upper dermis. In addition there was atrophy of the dermal appendages. These features were in keeping with poikiloderma atrophicans.

3. Comments

A literature search of disorders associated with congenital poikiloderma has failed to reveal a clearly identical disorder to that seen in the patient described above. The early onset in this patient is in keeping with a congenital disorder which, in view of the negative family history may suggest a mutation, or a non-heritable skin disorder, or an autosomal recessive disorder or she may be the offspring of a parent with a non-penetrant disease.

The characteristic findings in this patient include:

1. Unusual hypopigmentation at birth. The patient was actually described as being white at birth which is unusual for a Nigerian child.

Table 1

Comparison of our case with previously reported cases of RTS

Features of RTS	Our case
Poikiloderma	+
Onset at infancy	+
Short stature	+
Absence of or sparseness of eyelashes and scalp hair	+
Family history	–
Cataract (juvenile)	–
Bone defects	–
Small hands	–
Sensitivity to light	?
Hypogonadism	–
Defective dentition	–
Nail dystrophy	+

2. She had vesicles and sore areas on both ankles and upper back at birth which did not reoccur after birth.
3. She developed progressive poikiloderma, affecting the whole skin surface but sparing the palms and soles.
4. She had sparse golden brown scalp hair, scanty eyebrows and normal eyelashes.
5. Small stature.
6. Dystrophic nail changes.
7. Delayed mental development.

These above findings do not fit completely into any already described poikilodermatous disorder and a few differential diagnoses were therefore considered.

3.1. Congenital poikiloderma/Rothmund–Thompson syndrome (RTS) [1]

This is an autosomal recessive condition occurring predominantly in females. The skin is normal at birth and there is early onset of poikiloderma that starts as early as 3–6 months and is more marked on the photosensitive areas. Other features of RTS include short stature, absence of or scanty hair on the scalp, eyebrows and eyelashes, juvenile cataract, and nail dystrophy.

Below, we compare our case with previously reported cases of RTS in the English language literature [2–4] (Table 1).

The salient features to be noted are as follows. Our patient presented with unusual hypopigmentation and had blisters and sore areas on both ankles and on the upper trunk at birth but these features have not been previously reported in RTS; previous reports of RTS in a black child had been without hypopigmentation at birth [5]; vesicles and bullae have been reported in RTS as a result of photosensitivity [6] and although pigmentation was more in the sun exposed areas in our patient, vesicles did not develop.

There are some other features of RTS however that cannot be commented on as being absent in our patient because they tend to occur at a later age: (1) juvenile cataract, which tends to occur between the 4th and 7th year of life, and (2) hypogonadism, which manifests much later in life.

Other differential diagnoses include:

3.2. Congenital poikiloderma with features of hereditary acrokeratotic poikiloderma [6]

This was described in a 14 year old boy with bizarre progressive poikiloderma and acral blistering at birth. The blistering lesions were recurrent. The scalp, face, palm, and soles were unaffected throughout the duration of the illness in that patient. The generalised involvement of our patient's skin and the occurrence of blisters only at birth are not in support of this diagnosis.

3.3. Xeroderma pigmentosum (XP) [7]

This is an autosomal recessive disorder in which patients also present with poikiloderma, mostly occurring in the sun exposed areas of the body, due to an inability to repair ultraviolet damaged DNA. Unlike patients with XP, our patient's poikiloderma was generalised although with slight accentuation in the sun exposed areas of her body; she also had blisters at birth with scanty scalp hair and eyebrows, which are not associated with this syndrome.

3.4. Dyskeratosis congenita [8]

This is an X linked recessive disorder with full expression in males. Nail changes occur early in dyskeratosis congenita, between the first and second decades of life. They may be one of the first signs of dyskeratosis congenita. They then develop a reticulate pattern of poikiloderma and mucosal leukokeratosis. Females have been reported with mainly pigmentary abnormalities [9]. Our patient had only one dystrophic nail with generalised poikiloderma that started in the first decade of life.

3.5. Cockayne's syndrome [10]

Individuals with this syndrome present with early onset poikiloderma with marked photosensitivity, but unlike our patient they have a characteristic facies, viz. protruding ears, long limbs, and large hands and feet which were not present in our patient.

3.6. Ectodermal dysplasia [11]

In view of the involvement of the hair and nails the

ectodermal dysplasias where also considered, especially the group I-III described by Freire-Maia and Pinheiro [11]. However, the presence of this type of poikiloderma which happens to be the obvious abnormality in this child, has not been previously reported in ectodermal dysplasia within any of these three groups.

Other congenital blistering disorders excluded were the porphyrias and epidermolysis bullosa. Although a history suggestive of congenital hypopigmentation was obtained, this presentation of the patient had excluded such disorders.

The patient presented here has some similarities to RTS. However, we believe that this is a distinct, previously undescribed variant of congenital poikiloderma which is worthy of note.

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