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Case Report

Congenital skin aplasia associated with unilateral focal dermal hypoplasia

Imane Couissi, Zakia Douhi, Noura Kalmi, Meryem Soughi, Sara El Loudi, Hanane BayBay, Fatima Zahra Mernissi

Department of Dermatology, University Hospital Hassan II Fès, Morocco

Corresponding author: Imane Couissi, MD, E-mail: imane.couissi@usmba.ac.ma

ABSTRACT

Congenital skin aplasia (CCA) is a rare congenital anomaly involving variable layers of the skin, most commonly affecting the scalp yet may be seen on the trunk and limbs as well. It is most often seen as solitary lesions or as part of a heterogeneous group of syndromes, such as Goltz syndrome and focal dermal hypoplasia. Herein, we report a newborn of one day of life, who presented multiple, well-limited, reddish-orange, ulcerated patches with irregular contours and a clean surface, as well as hyperpigmented atrophic macules with a linear distribution along the Blaschko's lines along the left hemisphere. We observed syndactyly of the left second and third toes with hypoplasia of the left great lip. Goltz syndrome is a rare congenital skin characterized by a unique clinical presentation, which is unilateral focal dermal hypoplasia (FDH). Looking for other associated features is important. The recognition of these characteristic features will permit early appropriate genetic counseling and treatment.

Key words: Congenital skin aplasia; Syndrome; Unilateral focal dermal hypoplasia

INTRODUCTION

Congenital skin aplasia (CCA) is a rare congenital anomaly involving variable layers of the skin, most commonly affecting the scalp yet may be seen on the trunk and limbs as well [1,2]. CCA is most often seen as solitary lesions or as part of a heterogeneous group of syndromes, such as Goltz syndrome and focal dermal hypoplasia.

CASE REPORT

This was a newborn of one day of life from the non-consanguineous marriage of a 29-year-old mother. Well-monitored pregnancy was carried to term with a vaginal delivery. No specific medication, smoking, alcoholism, or toxin intake were present. No similar case in the siblings and no notion of autoimmune bullous dermatosis in the family was present as well.

Our opinion was sought for the congenital absence of skin on the left hemisphere. A general examination found a pink-toned and responsive newborn, HD, and respiratorily stable.

A dermatological examination revealed multiple, well-limited, reddish-orange, ulcerated patches with irregular contours and a clean surface (Figs. 1 and 2), as well as hyperpigmented atrophic macules with a linear distribution along the Blaschko's lines along the left half of the body (Figs. 3a and 3b).

The rest of the examination revealed syndactyly of the left second and third toes with hypoplasia of the left great lip. Trans-fontanelle and cardiac ultrasound returned without any particularities. Biological tests were normal. The diagnosis of focal dermal hypoplasia was retained.

DISCUSSION

Cordon first described CCA in 1767 and, since then, over 500 cases have been reported [2].

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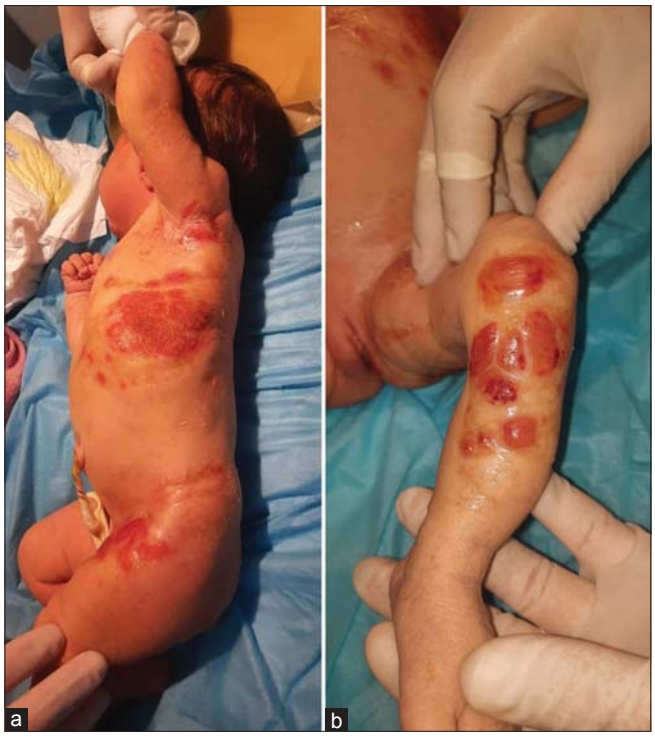

Figure 1: (a and b) Congenital patchy skin aplasia on the left half of the body.

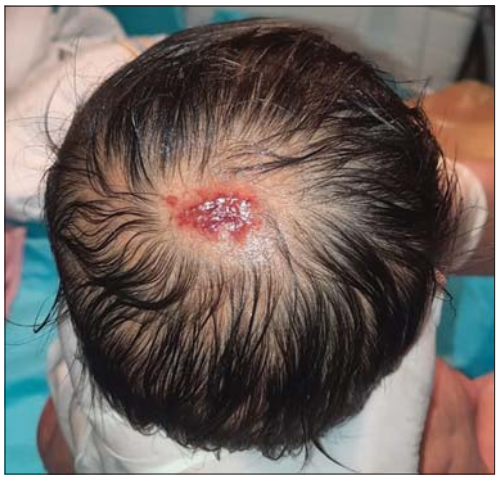

Figure 2: Congenital patchy skin aplasia on the scalp.

Eighty percent of cases of CCA occur on the scalp, while rarely being associated with a malformation syndrome. Some cases have been reported in the literature associated with trisomy 13, Wolf–Hirschhorn syndrome, ectodermal dysplasia, and Goltz syndrome (focal dermal hypoplasia).

According to the literature, more than 250 cases of unilateral focal dermal hypoplasia (FDH) have been reported worldwide [3]. Among these reports, only nine cases had unilateral or nearly unilateral FDH. Our case was one of the few cases of HPF with unilateral manifestations associated with congenital skin aplasia.

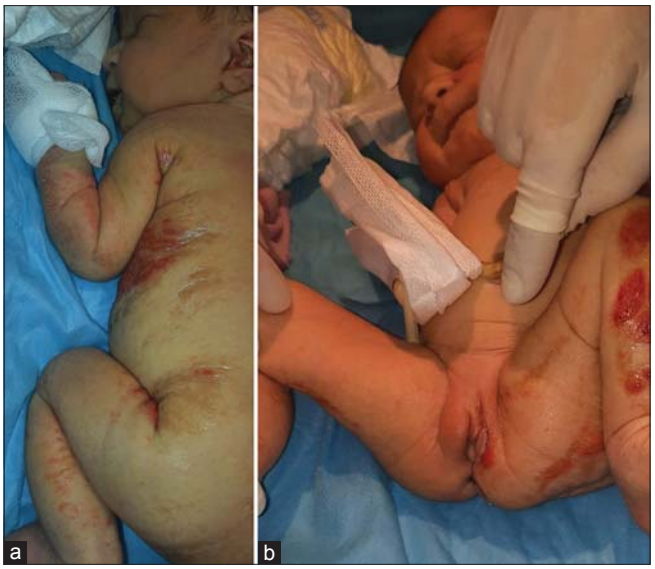

Figure 3: (a and b) Hyperpigmented atrophic macules with a linear distribution following a Blaschko's lines distribution along the left half of the body.

While HPF is an X-linked disease, as expected, 9 of 10 patients with unilateral HPF were female. However, only one male patient was reported with unilateral HPF. The right side of the body was predominantly affected in 70% of the patients, in contrast to our case, in which the left side was affected.

All patients had the classic presentation of pigmented atrophic skin, which follows Blaschko's lines associated with lesions of congenital cutaneous aplasia in hemicorporeal plaques. However, half of the patients (including our case) did not have fat herniation represented by fat nodules in the dermis, nor did they have scalp or dental involvement.

Musculoskeletal abnormalities were involved in 70% of the cases reported. Our case presented syndactyly of the second and third toe [4]. Ocular and nail involvement has been described in about 30% of the patients. It is somewhat surprising that internal organ involvement is mentioned in only one case report, published in 1984.

The diagnosis of unilateral focal dermal hypoplasia was usually based on the clinical skin presentation and associated symptoms. However, molecular genetic testing may be employed to confirm the diagnosis in cases where the clinic is inconclusive.

CONCLUSION

In conclusion, Goltz syndrome is a rare congenital skin characterized by a unique clinical presentation, which is FDH unilateral focal dermal hypoplasia.

Looking for other associated features is important. The close examination of the extremities is recommended. The recognition of these characteristic features will permit early treatment.

Consent

The examination of the patient was conducted according to the principles of the Declaration of Helsinki.

The authors certify that they have obtained all appropriate patient consent forms, in which the patients gave their consent for images and other clinical information to be included in the journal. The patients understand that their names and initials will not be published and due effort will be made to conceal their identity, but that anonymity cannot be guaranteed.

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