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Congenital absence of the skin: a case series report

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Abstract

Background: Extracranial congenital skin defects, defined as aplasia cutis congenita (ACC), are a rare clinical entity. The form of presentation varies among patients according to the causal agent and the point during pregnancy at which the defect occurs. We present a series of cases of noncranial ACC, analysing its possible etiopathogenesis and the evolution of the lesions.

Materials and methods: This is a retrospective review of cases of extracranial ACC at a tertiary hospital.

Case presentation: Patient 1 is a full-term newborn of a monochorionic diamniotic gestation with the death of one foetus at 14 weeks of gestational age. Physical examination revealed two skin defects located on the posterior aspect of both thoraco-abdominal flanks. Patient 2 is a full-term newborn presenting with an ulcer on the nasal tip and cephalic extension on a follow-up pregnancy ultrasound. Patient 3 is a full-term newborn of a monochorionic diamniotic gestation with twin feticide due to encephalocele, presenting an ulcer on the lateral aspect of the proximal third of her right leg. The gestation associates severe oligohydramnios due to premature rupture of the membranes and breech presentation. Patient 4 is a full-term newborn with an ulcer on the left antecubital fossa with no relevant gestational history.

Conclusion: The analysis of the obstetric history and the clinical presentation of the lesions helps to guide the aetiopathogenic diagnosis of congenital skin defects. The increased intracompartment pressure in the limbs is related to the increased severity of skin lesions and musculoskeletal and nerve involvement. Conservative treatment is related to a good prognosis for the lesions.

Background

Localized congenital skin defects are a rare entity that is defined in the literature as aplasia cutis congenita (ACC). The most frequent location is the scalp, where it presents as a sporadically occurring, isolated lesion; however, other parts of the body can also be involved [1].

According to the Frieden [2] classification, isolated involvement of the extremities without other associated anomalies is classified as type VII. Differential diagnosis with congenital Volkmann ischaemic contracture syndrome (CVICS) is difficult, since the two entities present overlapping clinical characteristics. Thus, there are authors who argue that CVICS constitutes a subtype of ACC type VII [3].

We present a series of ACC and CVICS cases with an extracranial location, and we analyse the possible aetiological agents according to the characteristics of the lesions and obstetric history, as well as the treatment and subsequent evolution.

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Material and methods

This is a retrospective analysis of a series of cases of congenital skin defects with an extracranial location that was diagnosed at a tertiary hospital, including a review of obstetric history and maternal-foetal risk factors, as well as the postnatal evolution according to treatment. Informed consent to publication was obtained directly from the patients' parents. Given that the study was retrospective, approval was not requested from the hospital ethics committee. The authors declare that they are free of conflicts of interest.

Case presentation

Clinical case 1

Full-term female newborn, 38 weeks of gestational age, weighing 3080 g. Daughter of a grand multiparous mother (7 previous pregnancies) with a personal history of type II diabetes mellitus that was first treated with metformin and then with insulin during the 15th week of pregnancy. She was pregnant with monochorionic diamniotic twins, with the death of one foetus at 14 weeks of gestational age. Vaginal delivery occurred without incident.

At our consultation at 8 days of life, the physical examination revealed two skin defects located on the posterior aspect of both thoraco-abdominal flanks. The left lesion was slightly larger than the right. Both defects had erythematous margins, were well defined, and were star-shaped. The absence of tissue was superficial, and healed areas could be observed on both sides.

Treatment was conservative and involved the application of occlusive dressing with Linitul® (Alfasigma) and bandaging. Progressive epithelialization was observed, with complete healing at 1 month of life (Fig. 1).

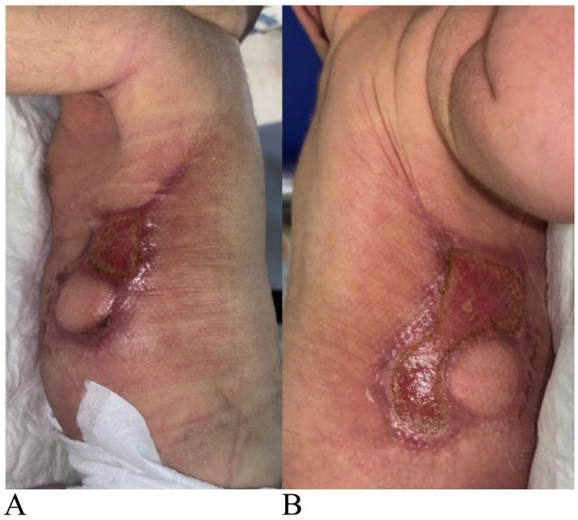


Fig. 1 Patient 1. Skin defects located on the posterior aspect of both thoraco-abdominal flanks (8 days of life). **A** Left side. **B** Right side

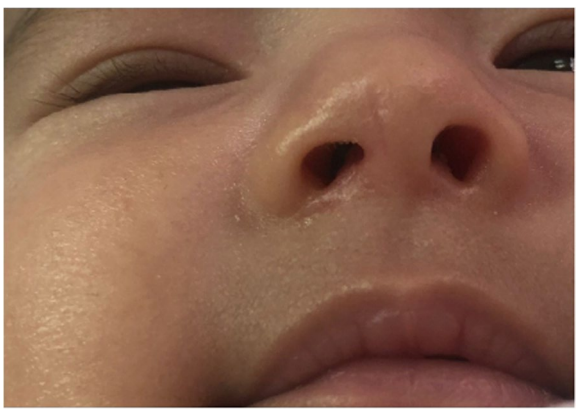


Fig. 2 Patient 2. Residual scars on the columella and alar base, resulting in minimal stenosis of the right nostril

Clinical case 2

Full-term female newborn, 40 weeks of gestational age, weighing 2800 g. Daughter of a biparous mother with a history of spontaneous abortion during week 10 that required the subsequent release of uterine synechia at the level of the internal os of the cervix. Single pregnancy by in vitro fertilization. Controlled pregnancy without incident. Third-trimester ultrasound showed a cephalic position with a tendency towards head extension. Delivery by caesarean section due to suspected loss of foetal well-being after a 22-h-long rupture of the membranes.

At birth, an ulcer was diagnosed on the nasal tip, with exposed cartilage and collapsed right ala that led to the deviation of the columella to the left side.

Treatment was performed with chlorhexidine-impregnated dressings. The alar cartilage recovered its natural anatomy without a need for nasal shapers. At discharge from the neonatal unit (when the infant was 15 days old), the wound was completely healed, with two residual scars on the columella and alar base, resulting in minimal stenosis of the right nostril. The patient did not require any further treatment (Fig. 2).

Clinical case 3

Female newborn, 33 + 3 weeks gestational age at birth, weighing 1640 g. Daughter of a 35-year-old primiparous mother with no clinical history of interest. Pregnancy with monochorionic diamniotic twins was achieved by in vitro fertilization. At week 16 of gestation, occipital encephalocele was observed in one of the twins. At week 22, a feticide was performed by extra-amniotic laser

photocoagulation of the placental insertion of the umbilical cord of the affected foetus. Premature rupture of the membranes at week 26 led to oligohydramnios. Caesarean delivery at 33 + 3 weeks was performed due to suspected chorioamnionitis and breech presentation.

At birth, an ulcer was observed on the lateral aspect of the proximal third of the right leg, with muscle exposure. In the lesion bed, bone splinter was evident; therefore, a fibula fracture was assumed. The edges of the lesion were well defined. The distal muscles of the leg were hypotrophic, and ipsilateral clubfoot without dorsal flexion movements and eversion of the ankle were present. Distal pulses were retained, with good capillary filling.

Occlusive dressings with Aquacel® (ConvaTec) and Prontosan® (B.Braun) were applied to the wound. When the infant was 1 week old, closure of the edges with direct tensionless suture was attempted after debridement, but dehiscence occurred after 5 days. Treatment with local dressings was maintained to facilitate a second attempt at closure (cleaning with chlorhexidine and closure of the edges with Steri-Strips).

In terms of trauma, X-rays taken at birth revealed a peroneal fracture at the level of the neck. As an initial treatment, an ankle foot orthosis was placed for suspected involvement of the external popliteal sciatic nerve. A bone scan was performed that showed no abnormality in any other bone, and a phospho-calcium metabolism analysis showed normal results. Prior to discharge, at 1 month of life, X-rays were repeated and showed that the fracture was in the process of consolidation, with bone callus formation. As of 9 months of follow-up, the patient is continuing treatment with Ponseti boots and shows improvement in the active mobility of the ankle and toes, which maintain some medial deviation (Fig. 3).

Clinical case 4

Full-term female newborn, 39 + 4 weeks of gestational age, weighing 3310 g at birth. Daughter of a biparous mother with no clinical history of interest. Gestation of a single foetus by in vitro fertilization, with controlled pregnancy without incident. Prenatal ultrasound without described anomalies. Eutocic vaginal delivery without incident.

At birth, two ulcers were evident in the left antecubital fossa; they had a necrotic bed and muscular exposure and were associated with wrist drop of the left hand and mild dorsal atrophy. The infant presented restricted pronation/supination and extension of the wrist, with palpable distal pulses and good capillary filling. An ultrasound showed atrophy of all muscle groups of the left forearm.

Local dressings with chlorhexidine were applied to both ulcers, and rehabilitation treatment of the hand has led to progressive improvement in wrist function. As of 3 years of follow-up, the resulting scars are well-formed and do not limit the mobility of the elbow. The range of motion of the wrist is unrestricted (Fig. 4).

Discussion

ACC constitutes a heterogeneous group of congenital defects characterized by the localized absence of skin. The first case was described by Cordon in 1767 [4]. The most frequent form of presentation is sporadic. Hereditary forms have also been described, including autosomal

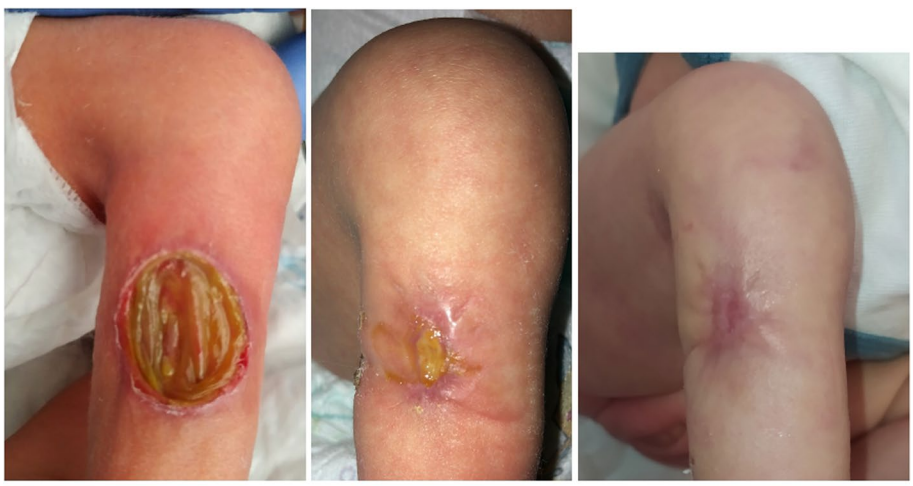
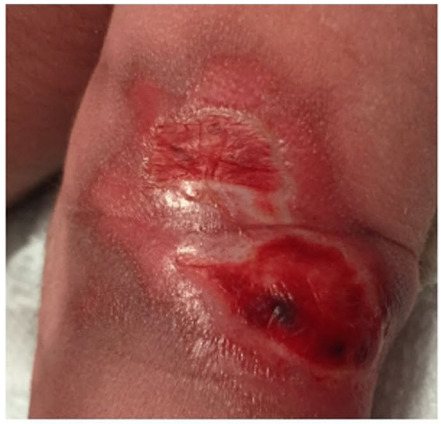


Fig. 3 Patient 3. **A** Skin defect on the lateral aspect of the lower limb at birth. **B** Skin defect appearance after a month of follow-up. **C** Immature scar after 2 months



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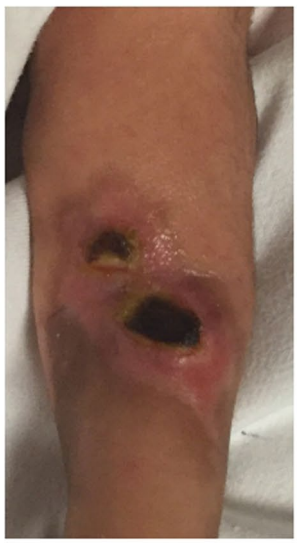

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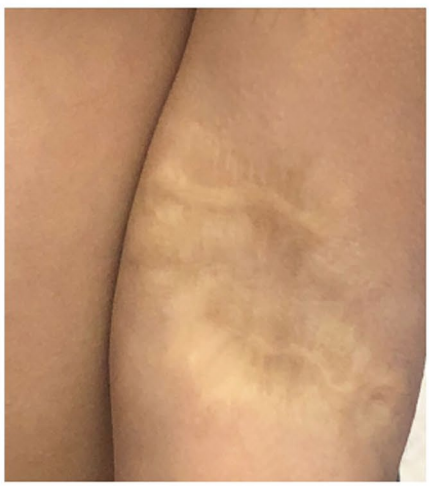

C

Fig. 4 Patient 4. **A** Skin defects in the antecubital fossa present at birth. **B** Progression 1 month after birth. **C** Mature scars after 3 years of follow-up

dominant forms with low penetrance and autosomal recessive forms [5].

Extracranial involvement is rare but has been described in the literature, especially on the trunk and upper extremities [6]. The clinical presentation in such cases is highly varied, and its evolution is intimately linked to the aetiology of the defect and to point pregnancy when it occurs [7]. Different studies have observed clear differences in the mode of foetal healing when the wound occurs during the first two trimesters of pregnancy compared to the third trimester. Before 24 weeks of gestation, the foetus is able to perform regenerative healing without fibrous scar formation. In the last stage of pregnancy, the process is similar to that during extrauterine life and is generally associated with more adverse effects [8].

At present, the exact mechanism that causes the characteristic absence of skin in ACC is not yet known. Theories have been proposed regarding the clinical presentation of lesions and the obstetric history. Classically, 5 different theories have been proposed: foetus-foetus transfusion, vascularization and coagulation disorders, amniotic membrane adhesions, biomechanical alterations, and external teratogenic agents [9].

Abnormalities in placental vascularization, thrombosis/placental infarction, and changes in circulatory dynamics are postulated as the main theories in cases of ACC associated with a foetus papyraceous (type V of the Frieden classification). The clinical presentation tends to be bilateral and symmetrical, with predominant involvement of the trunk and a stellate appearance of the lesions [10, 11]. The history of a deceased foetus during the first trimester of pregnancy is present in case 1 and constitutes the main aetiological hypothesis in this case. The observed lesions are also consistent with this ACC subtype. Metformin and insulin, drugs that the patient's mother took during pregnancy, have not been associated with significant teratogenesis, and their role as an aetiological factor is rejected.

In case 2, the position of the foetal head on the control ultrasound (a tendency towards extension) would justify the decubitus of the nose against the uterine walls, which would condition not only the loss of substance but the deviation of the associated columella. These biomechanical alterations would justify the appearance of the lesion in this specific area but cannot rule out the presence of adhesions to the amniotic membrane that would explain the disruption of the skin.

Cases 3 and 4 showed lesions of the extremities that are examples of ACC type VII according to the Frieden classification. However, both cases show deep skin involvement, with eschar-like lesions affecting deep tissues. In addition, the patients presented distal atrophy of the affected limbs and some movement restrictions, which led to a consideration of nerve involvement. This type of lesion has been described in the literature as CVICS, a syndrome characterized by muscle necrosis and nerve paralysis secondary to an increase in intracompartment pressure [12, 13].

The difference between CVICS and ACC is not clear, so much that some authors have proposed that ACC subtype VII includes lesions described as ischaemic contractures. In reality, the distinction is rather conceptual: ACC constitutes a clinical diagnosis, while CVICS describes the pathophysiological mechanism [12].

In most cases of CVICS described in the literature, the lesions tend to be located in the upper limbs [14, 15]; however, it can also affect the lower limbs. In the third patient in our series, premature membrane rupture with associated severe oligohydramnios, as well as an alteration in the normal position of the foetus, was a risk factor associated with an increase in intracompartment pressure [14, 16]. In addition, the location of the lesion at the level of the head of the fibula is a risk factor in itself, since it is an area with little soft tissue coverage and therefore increased vulnerability to pressure. Similarly, the scarcity of amniotic fluid conditions a state of foetal immobility that predisposes patients to lower bone mineralization, which favours stress fractures [17].

In this specific case, the antecedent of an invasive antenatal procedure was also considered a possible cause of the injury. However, this does not seem like a very plausible explanation, since, in addition to the fact that the procedure was performed without entering the amniotic cavity, the characteristics of the wound, including the complete absence of scarring at birth, suggest prolonged aggravation over time rather than development late in pregnancy.

Regarding the patient in case 2, lesions related to the CVICS have been described more frequently in the volar aspect of the upper limb, with variable extension. At birth, the ulcers did not present the characteristic yellowing of lesions caused by increased intracompartment pressure; however, hypotrophy of the arm was clear, as was nerve paralysis. A careful examination of the affected limb showed that the intrauterine lesion had been more extensive but that some of the skin had already healed. Although there was no clear evidence, the authors postulated that the lesion could have occurred early in pregnancy and that most of the skin regeneration had occurred in utero.

The differential diagnosis of the latter cases included entities such as neonatal gangrene, necrotizing fasciitis or congenital varicella. However, the Doppler study of the extremities did not show vascular involvement, and the maternal history did not suggest a congenital infection. The absence of blisters ruled out an association with epidermolysis bullosa. In case 4, an important differential diagnosis to take into account would be amniotic band syndrome; however, in this pathology, it is common to find other associated defects in the affected limb due to the effect of constrictive bands. In addition, expression of the lesions as ulcers is infrequent; instead, they usually manifest as deep circumferential depressions [12].

Regarding wound healing, it is important to take into account the regenerative capacity during the early neonatal period, during which there is less production of fibrous scar tissue than in later stages [18]. This fact makes conservative treatment the first choice, since in our experience, the generated scar will be well-formed and will not be associated with limitations in function. Similarly, when the extremities are, we believe that it is of utmost importance to initiate rehabilitation treatment early. The effects of increased intracompartment pressure are not limited to the skin but can include muscle hypotrophy, ligamentous contracture and nerve paralysis that would benefit from early musculoskeletal stimulation.

The limitations of the study lie in its retrospective nature and the fact that the case series was limited to a single centre. In addition, to complete the aetiological diagnosis, it would have been useful to have pathological studies showing the degree and type of skin involvement. However, few cases with these characteristics are presented in the literature, and therefore, it is important to contribute to the study of this condition by detailing the experience at each centre.

Conclusion

Extracranial congenital skin loss is an infrequent finding with a presentation that varies greatly depending on the aetiology and point in pregnancy at which the damage occurs. CVICS constitutes an aetiopathogenic mechanism of skin lesions extending to deep planes that are associated with an increase in intracompartment pressure. Its distinction from ACC is difficult, and the two entities are closely related. Conservative treatment is the gold standard for skin regeneration, while muscle atrophy and nerve paralysis benefit from early rehabilitation.

Authors' contributions

Jesús Vicente Redondo Sedano conceived and designed the analysis, performed the analysis, and wrote the paper. María Dolores Delgado Muñoz conceived and designed the analysis and contributed data or analysis tools.

Alicia Gómez Sánchez contributed data or analysis tools and collected the data. Sara Monserrat Proaño Landazuri contributed data or analysis tools and collected the data. María López Maestro contributed data or analysis tools. Jesús Rodríguez Calvo contributed data or analysis tools. Cecilia Castellano Yañez collected the data. Andrés Gómez Fraile contributed data or analysis tools. María Eunate Marti Carrera conceived and designed the analysis and contributed data or analysis tools. The authors read and approved the final manuscript.

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Availability of data and materials
The authors confirm that the data supporting the findings of this study are available within the article and/or its supplementary material.

Declarations

Ethics approval and consent to participate
All procedures performed in our study were in accordance with the ethical standards of the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.
Informed consent was obtained from all legal guardians of the children included in the study.

Consent for publication
All authors agree in publishing this original article.

Competing interests
The authors declare that they have no competing interests.

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