

## Aplasia cutis congenita: a rare extensive bilateral case of extremities

Majdouline Boujoual<sup>1</sup>, Fatima Jabouirik<sup>2</sup>, Jaouad Kouach<sup>3</sup>, Driss Moussaoui<sup>3</sup>, and Mohammed Dehayni<sup>3</sup>

<sup>1</sup>Department of Gynecology-Obstetric,  
University of medicine Tangier, Military Training Hospital Med V,  
Rabat, Morocco

<sup>2</sup>Department of Pediatrics,  
Ibn Sina Hospital Center,  
Rabat, Morocco

<sup>3</sup>Department of Gynecology-Obstetric,  
Military Training Hospital Med V,  
Rabat, Morocco

---

Copyright © 2014 ISSR Journals. This is an open access article distributed under the *Creative Commons Attribution License*, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.

**ABSTRACT:** Aplasia cutis congenital is a rare congenital absence of skin, exceptionally involves extremities. Most cases are sporadic but familial autosomal dominant pattern can be found. The etiology remains unclear, both genetic and environmental causes have been implicated. This lesion has primarily a clinical diagnosis. Imaging studies may be used to evaluate underlying tissues and investigate other comorbid abnormalities. Treatment modalities depend upon the extent defect and involvement of underlying tissues. Genetic counseling is useful in cases of hereditary form. We report a new case of extensive bilateral aplasia cutis congenital of extremities in a new born, occurred as an isolated sporadic case without family history, no teratogenic medication and no evidence for an amniotic band or infection in pregnancy.

**KEYWORDS:** Aplasia cutis congenital, Diagnosis, Etiology, Prognosis, Management.

### 1 INTRODUCTION

Aplasia cutis congenita (ACC) is a rare congenital absence of skin (1-3/2000-10000), first described as a lesion on an extremity by Cordon in 1767 and after on the scalp by Campbell in 1826. Most cases are sporadic but a familial autosomal dominant pattern of inheritance can occur [1]. Lesions can be multiple on different surfaces of the body but mostly seen on the scalp and rarely on extremities [2]. ACC is primarily a clinical diagnosis, with no specific histologic alterations. In the new born, ACC poses diagnosis, evolutionary and therapeutic difficulties: it could be mistaken by obstetricians for damage inflicted by spiral electrodes or Willet's forceps [3], or associated with other congenital malformations, intrauterine infections, chromosomal anomalies, ectodermal dysplasia or epidermolysis bullosa [4] and expose to risk of mortality in approximately 21 %. Also management is still controversial may be conservative, surgical or combination of both [5].

### 2 OBSERVATION

A 3 day old mal baby, delivered at term by cesarean section with good extra-uterine life adaptation. He was born of a consanguineous marriage with no family history of congenital anomalies, no history of medications or disease during pregnancy and no birth iatrogenic trauma.

On examination at birth, the baby weighed 3.2 Kg, had normal neonatal reflexes and was found to have a left extensive lower limb defect characterized by a well demarcated erythematous superficially eroded skin lesion extended from the thigh

to the foot, dotted with ulcerative hemorrhagic closets, sometimes covered by a thin, translucent oozing membrane and associated with an epidermolysis more pronounced at the foot, while surrounding skin was normal (figure 1). Symmetric lesions were seen but less marked on right lower limb (figure 2). Systemic examination was unremarkable with no clinical stigmata of any other congenital abnormality. A clinical diagnosis of ACC was made. No X-ray defects had been found. Laboratory examinations including complete blood count, biochemistry, parameters for liver and kidney functions were all within normal limits. Echocardiography and abdominal ultrasonography didn't show any other abnormalities.

Local wound care was provided based on gentle cleansing of the denuded area with saline-diluted bovidine iodine lotion and then covering it with antibiotic-impregnated non-adherent dressing to promote healing in a moist environment. After two weeks, new born was transferred to pediatric plastic surgeon for plastic reconstructive surgery because of his extensive lesions. The parents were informed of recurrence risk in future pregnancy if a hereditary form is involved following an autosomal dominant pattern.



***Figure 1: left lower limb aplasia cutis congenita extended from the thigh to the foot***



***Figure 2: right lower limb less marked lesion of aplasia cutis congenita***

### **3 DISCUSSION**

#### **3.1 DEFINITION**

ACC is a heterogeneous group of disorders characterized by well circumscribed focal absence of epidermis, dermis and occasionally subcutis at birth. Non-scalp lesions may involve the trunk and/or extremities and are usually bilaterally symmetric [6]. The lesion is non-inflammatory and well demarcated, may be round, oval, linear or stellate [7], involving less commonly underlying periosteum defect [8], it can be superficially eroded, deeply ulcerated, completely scarred or covered with a membranous epithelium filled with serous fluid resembling a blister such us our case [9].

### 3.2 DIAGNOSIS

Antenatal diagnosis can be suspected in presence of raised  $\alpha$  fetoprotein level in amniotic fluid and maternal blood or a positive acetyl - cholinesterase test [10]. However, the diagnosis is usually made at birth based on clinical criteria [1]. Histopathological features include absent thin epidermis, weak dermis consisting of loosely arranged collagen bundles and thin sub-cutis. Dermal papillae and elastic fibers are absent and blood vessels are mal-developed [10]. Imaging studies may be used to evaluate underlying tissues, to investigate other comorbid abnormalities and help to establish pre-surgical planning [8].

Our case presented bilateral symmetric defect of extremities occurred as an isolated sporadic case without family history, no teratogenic medication and no evidence for an amniotic band or infection in pregnancy.

### 3.3 ETIOLOGY

The etiology remains unclear; both genetic and environmental causes have been implicated: Vascular abnormalities, including disruption of vascular blood supply, arrest of midline embryological development, defects in closure of the neural tube, viral infections, involution of an intrauterine hemangioma, amniotic adherence have been attributed as the cause [7], [8].

A few case reports are linked to certain teratogens such as Methimazole, carbimazole, misoprostol, valproic acid, diclofenac, angiotensin-converting enzyme inhibitors, benzodiazepines, low molecular weight heparin [9], [10], [11]. ACC can also be associated with other physical anomalies [2]. The most common are limb anomalies: amelia, syndactyly, limb reduction defects, polydactyly, Volkmann's ischemic contracture and clubbing of the hands and feet. Bowel atresia may also be associated. Other less common associations include cleft lip and palate, pencephaly, hydrocephalus, neural tube defects, epidermolysis bullosa, ectodermal dysplasias and chromosomal anomalies such as trisomy.

### 3.4 DIFFERENTIAL DIAGNOSES

Differential diagnoses include epidermolysis bullosa, focal dermal hypoplasia syndrome, neonatal herpes, fetal varicella, transient bullous dermolysis of the newborn and lesions attributed to birth trauma secondary to vacuum extraction, forceps or fetal scalp monitor electrodes [6], [10].

### 3.5 PROGNOSTIC

Early death during pregnancy is usually associated with ischemic complications manifesting as absence of structure, whereas late mortality in the second or third trimester tends to be associated with embolic phenomena and coagulopathy, leading to neurological problems in the survivor [1]. At born, large lesions cause death secondary to infection or hemorrhage which can lead to mortality of 20-30% [2], [10]

### 3.6 MANAGEMENT

Management necessitates interdisciplinary cooperation. Treatment modalities depend upon the extent and involvement of underlying tissues defect: small isolated cutaneous involvement can decrease in size via secondary epithelialization from surrounding healthy tissue, requiring a conservative approach avoiding potential operative risk to the infant. However, the possible complications of conservative management are: wound infection, fatal hemorrhage and biochemical abnormalities due to sulphadiazine dressings who may lead to increased potassium absorption and cause hyperkalemia [3], [5], [6], [8], [12]. Therefore, involvement of vital organs would warrant early surgery [11], the same as large lesions who may necessitate surgical interference with skin grafts or local skin flaps [6]. Genetic counseling is useful when ACC is inherited [12].

## 4 CONCLUSION

Aplasia cutis is a rare dermatological disorder of neonates which may rarely have an extensive bilaterally symmetrical presentation of extremities. Medical history, physical examination, imaging and genetic study should be made to rule out any other associated anomalies. The management involves a multidisciplinary approach including both conservative and surgical treatment in order to minimize complications and optimize outcomes.

**REFERENCES**

- [1] C. Lückoff, J. McDougall, M. Nelle, K. Kernland Lang and H.J. Altermatt, "Aplasia cutis congenita in a surviving twin", *Swiss Society of Neonatology*, 2002. [Online] Available: [http://www.neonet.ch/en/03\\_Case\\_of\\_the\\_month/archive/11\\_dermatologic\\_disorders/2002\\_10.asp](http://www.neonet.ch/en/03_Case_of_the_month/archive/11_dermatologic_disorders/2002_10.asp)
- [2] M.B. Yilmaz, C. Aydin , E. Ergun, H. Nurata and M.K. Baykander, "Aplasia Cutis Congenita Associated with Multiple Congenital Anomalies", *Turk Neurosurg*, Vol: 20, No: 1, 66-68, 2010.
- [3] R. Izhar and I. Ghani, " Aplasia Cutis Congenita and Antithyroid Drugs", *J Pak Med Assoc*, 52(11): 526-8, 2002.
- [4] E. Mihçi, S. Erişir, Ş.Taçoy, G. Lüleci, E. Alpsoy and N. Oygür, "Aplasia cutis congenita: three cases with three different underlying etiologies", *The Turk J of Pediatr* , 51: 510-514, 2009.
- [5] Tahmeedullah, Naseem ul Haq, M. Bilal and N. Laiq, "Aplasia cutis congenita scalp : management options", *JPostgrad Med Inst*, 24 (1): 36-40, 2010.
- [6] T.A. Abulezz and M.A. Shalkamy, " Aplasia cutis congenita: Two cases of non-scalp lesions", *Indian J Plast Surg*. Jul-Dec; 42(2): 261–264, 2009.
- [7] L. Tasin, S. Belli and E. Chiodini, " Aplasia cutis congenita and methimazole. A case report and literature review", *Eur. J. Pediat. Dermatol.* 15, 117-20, 2005.
- [8] Steven M. Levine, Derek D. Reformat and Charles H. Thorne, "Cutis Aplasia: Perioperative Management and Case Report", *Am J Crit Care*, 21:212-215, 2012.
- [9] J.F Chen, S.C Chen and C.P Chiang, "Aplasia Cutis Congenita Associated with Cutis Marmorata Telangiectatica Congenita, Atrial Septal Defect, and Epilepsy: A Newly Recognized Syndrome? A Case Report", *Dermatol Sinica*, 157-164, 2008.
- [10] S.A. Salati, "Familial aplasia cutis congenital", *Journal of Pakistan Association of Dermatologists*, 23 (2):221-224, 2013.
- [11] S. Ramachandran, K. Mishra and V.V.Batra, "Aplasia Cutis Congenita: A Rare Case with Extensive Symmetrically Distributed Lesions", *Journal of the association of physicians of india*, Vol. 62: 274-276, 2014.
- [12] S. K. Shivakumar, S. Dwarakanath, G. Swaroop and N. K. Venkataramana, "Aplasia cutis congenita of the scalp: Therapeutic modalities" *Neurology India*, Vol 54, Issue 3: 312-313, 2006.