SOMATO PUBLICATIONS ISSN: 2688-1071

Archives of Clinical Case Reports

Case Report

Aplasia Cutis Congenita on the Lower Limbs in A Full-Term Infant

Yanrong Wang*, Lu Chang and Hong Li

Neonatal Department of Neonatology, Third People's Hospital of Shenzhen, Shenzhen, China

*Address for Correspondence: Yanrong Wang, Department of Neonatology, Third People's Hospital of Shenzhen, Shenzhen, China, E-mail: 123rong@sohu.com

Received: 01 December 2021; Accepted: 19 December 2021; Published: 22 December 2021

Citation of this article: Wang, Y., Chang, L., Li, H. (2021) Aplasia Cutis Congenita on the Lower Limbs in A Full-Term Infant. Arch Clin Case Rep, 4(2): 10-12.

Introduction

Aplasia cutis congenita (ACC) presents the congenital absence of skin, which may occur anywhere on the body and was first reported in 1767 by Cordon [1]. It is a rare congenital abnormality involving variable layers of the skin and is devastating malformations to parents. The exact etiopathogenesis of ACC is not known and may be multifactorial in association with genetics, environmental and exogenous causes.

Intrauterine conditions may play an important role, e.g. placental dysfunction, embryological malformations, epidermal dysplasias, chromosomal aberrations, infections (mainly viral infections), or hypoxia. Sporadic cases have been reported as autosomal dominant or autosomal recessive [2.3] and the disease occurs with an incidence rate of 1-3 cases per 10 000 live births, regardless of race or sex. The protocol of ACC and the timing of surgery attribute to the risks involved with both conservative and surgical approaches. The conservative therapy exposes the patient to the risk of desiccation and necrosis of the ACC tenuous wound bed and related morbidity, such as hemorrhage, infection, superior sagittal sinus thrombosis and slow wound healing. In contrast, surgical intervention carries the risks of anesthesia, infection, massive hemorrhage, skin graft loss, scalp flap necrosis and donor-site morbidity. Here we report our experience with the treatment of newborns suffering from ACC.

Case Presentation

We presented a male infant from a single pregnancy, with a birth weight of 3230 g, head circumference of 34 cm and body length of 50 cm, born after 39 full weeks of gestation, delivered by Caesarean section because of impending fetal asphyxia at a hospital. It was evaluated at 9/10/10 points on the Apgar score respectively on 1/5/10 minutes of life. The mother was a 35-year-old woman, second pregnancy, first birth. During two weeks before birth the mother had elevated serum creatinine (92umol/L), and decreased glomerular filtration rate (60ml/min). There were no fetal abnormalities at the 12-and 38-week ultrasonography scans. The histologic feature of the placenta indicated chorioamnionitis. The first pregnancy was Ectopic pregnancy in the 10th week. The mother had no history of herpes simplex virus or varicella zoster virus infection during pregnancy. No congenital disorder was found

among the family's medical history.

On examination, there was focal skin defect on the lower extremities (Figure 1). There were no cutaneous blisters or vesicles, no digital or limb length discrepancies, and no nail, scalp or oral abnormalities. The rest of the neonatal examination was unremarkable.

Ultrasounds of brain's underlying structures, heart and abdomen were normal. Skeletal survey, hearing and ophthalmological examination were normal. Both infammatory markers and blood cell counts were normal. Microbiological swabs were taken from these lesions and baby's larynx, which were found to be aseptic. Chromosomal study and histology of skin defects was not performed because the parents did not consent to the procedure.

The baby was treated with antibacterial cream for skin defects and a dry-adhesive circular bandage was used and changed every two to three days. After 14 days, the defects were re-epithelialised. Baby was discharged home after 21 days of admission.

Further consultation at 5 weeks of age showed evidence of complete healing, leaving hyperpigmentation on the lower extremities. The baby did not present with any functional problem. Weight gain was appropriate for age. A follow up after 9 months showed a favorable outcome without scar formation and functional impairment of the legs.

Discussion and Conclusion

ACC is a rare congenital heterogeneous disorder [4] that are associated with either skin degeneration or failure of the skin to fully develop [5]. ACC is classified into nine groups on the basis of presence of associated abnormalities, mode of inheritance, location and pattern of the skin defect (Table 1). Till 2017, there had been reports of approximately 500 cases. The exact mechanism is not fully understood, but many risk factors, such as intrauterine trauma, chromosomal abnormalities and amniotic defects have been implicated [6]. ACC can occur on any parts of the body and may involve all layers including the skull, a condition that can be found in 20% of patients. According to Frieden's [5]classification system for ACC consisting of nine groups as the baby presented lesions on the lower limbs without other complications and screening for involvement of other systems such as brain, ear, eye, heart, and abdominal organs was normal, the baby was diagnosed as type VII ACC.

Unfortunately, the histology of skin defects could not be done due to parents' consent.

Being a rare disease, the optimal treatment protocol for ACC has not been established [7]. Since 1970, surgical and conservative management have been attempted [8]. Small lesions may heal spontaneously without or with an atrophic scar that needs surgical intervention. Our patient was treated with conservative therapy (antibacterial cream) to prevent secondary infection and the result was favorable. No severe complications such as infection or hemorrhage were found which may be attributable to the fact that the patient had relatively mild forms of disease that did not require more surgical intervention.

Previous reports emphasized intrauterine vascular ischemia, am-

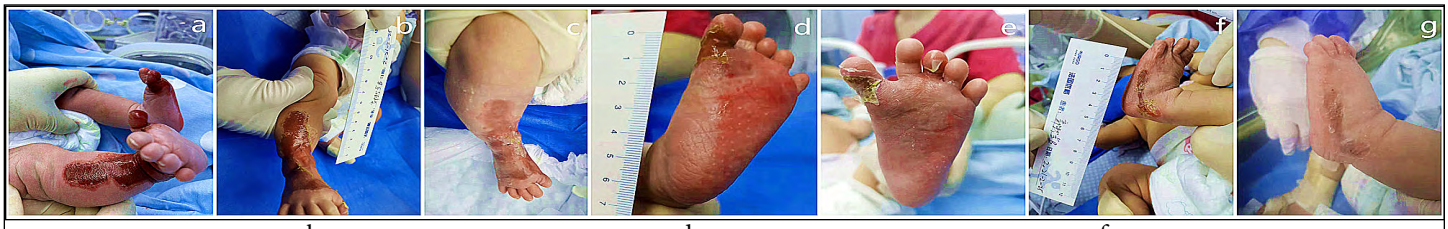

Figure 1: Clinical presentation of aplasia cutis congenita (ACC)

(a).Skin defects of aplasia cutis congenita on day 1; (b) view of the right lower limb on day 14; (c) view of the right lower limb on day 35; (d) view of the left plantar surface on day 14; (e)view of the left plantar surface on day 35; (f) lateral surface of left foot on day 14; (g) lateral surface of left foot on day 35

Table 1: Frieden's classification of ACC	
Type I	ACC of the scalp without multiple abnormalities
Type II	ACC of the scalp with associated limb abnormalities
Type III	ACC of the scalp with associated epidermal and organoid nevi
Type IV	ACC overlying embryologic malformations
Type V	ACC associated with fetus papyraceus or placental infarcts
Type VI	ACC associated with epidermolysis bullosa
Type VII	ACC localized to the extremities without blistering and without associated abnormalities
Type VIII	ACC caused by specific teratogens
Type IX	ACC associated with malformation syndromes

niotic adherences, and viral infections may attribute to ACC and associated anomalies [9,10]. The mother had no history of infections, exposure to any teratogenic substances such as alcohol or other chemicals, or radiation during pregnancy. The mother had elevated serum creatinine and decreased glomerular filtration rate and the pathologic change of placenta was observed. The family's medical history had no congenital disorders. In spite of the absence of chromosomal study and histology of skin, we speculate that mother's health condition may play a role in ACC.

ACC of the lower extremities can involve some critical areas proximal to a joint. Conservative therapy is recommended. The etiology is elusive and mother's health condition may attribute to ACC. More cases and cytogenetic techniques are needed to better understand the exact etiology of Aplasia cutis congenita.

Declaration of interest

The authors report no conflicts of interest.

References

1. Cordon, M. (1767) Extrait d'une lettre au sujet de trois enfants de la même mère nés avec partie des extrémités dénuée de peau. J Med Chir Pharm, 26: 556-557.
2. Lei, GF., Zhang, J., Wang. XB., You, XL., Gao, JY., Li, XM., et al. (2019) Treating aplasia cutis congenita in a newborn with the combination of ionic silver dressing and moist exposed burn ointment: a case report. World J Clin Cases, 7(17): 2611-2616.
3. Marneros, AG. (2013) BMS1 is mutated in aplasia cutis congenita. PLoS Genet, 9(6): e1003573.
4. Verhelle, NA., Heymans, O., Deleuze, JP., Fabre, G., Vranckx, JJ., Van den, hof B. (2004) Abdominal aplasia cutis congenita: case report and review of the literature. J Pediatr Surg, 39(2): 237-239.
5. Frieden, IJ. (1986) Aplasia cutis congenita: a clinical review and proposal for classification. J Am Acad Dermatol, 14(4): 646-660.
6. Brzezinski, P., Pinteala, T., Chiriac, AE., Foia, L., Chiriac, A. (2015) Aplasia cutis congenita of the Scalp-what are the steps to be followed? Case report and review of the literature. An Bras Dermatol, 90:100-103.
7. Browning, JC. (2013) Aplasia cutis congenita: approach to evaluation and management. Dermatol Ther, 26(6): 439-444.
8. Belkhou, A., François, C., Bennis, Y., Duquennoy-Martinot, V., Guerreschi, P. (2016) Aplasia cutis congenita: Update and management. Ann Chir Plast Esthet, 61(5): 450-461.
9. Duan, X., Yang, GE., Yu, D., Yu, C., Wang, B., Guo, Y. (2015) Aplasia cutis congenita: A case report and literature review. Exp Ther Med, 10(5): 1893-1895.
10. Schierz, I., Giuffrè, M., Vecchio, A., Antona, V., Corsello, G., Piro, E. (2021) Recognizable neonatal clinical features of aplasia cutis congenita. Italian Journal of Pediatrics, 46(4): 25-29.