

Case Report

Aplasia cutis congenita associated with epidermolysis bullosa in a Nigerian child

Sunday O. Ochigbo^{1*}, Anachunna I. Kingsley², Cobham E. Antigha², Udo J. Jacob¹

Department of Paediatrics, ¹University of Calabar, ²University of Calabar Teaching Hospital, Calabar, Cross River State, Nigeria

Received: 20 October 2017

Revised: 13 November 2017

Accepted: 14 November 2017

***Correspondence:**

Dr. Sunday O. Ochigbo,

E-mail: ochigbosunny@gmail.com

Copyright: © the author(s), publisher and licensee Medip Academy. This is an open-access article distributed under the terms of the Creative Commons Attribution Non-Commercial License, which permits unrestricted non-commercial use, distribution, and reproduction in any medium, provided the original work is properly cited.

ABSTRACT

Aplasia cutis congenita (ACC) associated with epidermolysis bullosa is a rare heterogeneous congenital cutaneous lesion characterized by absence of skin mostly involving the epidermis and dermis. ACC commonly affect the scalp in 75% of cases but can affect other sites like the trunk and limbs. It is highly heterogeneous and inherited either as autosomal dominant, recessive or caused by a new mutation. The exact cause is unknown although some intrauterine conditions may play a role in the etiology. Anticonvulsants and antithyroid drugs have also been implicated. The condition can coexist with other syndromes. Our patient was 2.95 kg female baby delivered via emergency caesarean section (CS) due to previous CS and cephalopelvic disproportion (CPD) at gestational age (GA) of 42 weeks. After delivery, we observed skin defects which measured 6 cm×4 cm and 2 cm×2.5 cm on the shin and dorsum of the right foot respectively. There were loose fragment of skin and blisters around the hypopigmented edges of the lesion. Milia were observed on the dorsum of the foot; hence patient was classified into group VI Friedens classification of ACC. The lesion was conservatively managed and dressed daily using gauze-impregnated with honey. The lesion healed within 2 weeks and 4 weeks review showed no residual scar. We recommend conservative management with honey in the absence of deep tissue loss as observed in our patient, as well as educate parents on the need to avoid aggravating factors.

Keywords: Newborn, Heterogeneous, Aplasia cutis, Skin

INTRODUCTION

Aplasia cutis congenita (ACC) is an inherited rare disorder of the newborn, which commonly affect the scalp in 75% of cases, can also affect other sites like the trunk and the limbs. The condition is characterized by the absence of the skin mostly involving the epidermis and dermis.¹ ACC is highly heterogeneous, it can be inherited either as autosomal dominant, recessive or caused by a new mutation. It can coexist with other congenital malformations. The absence of family history does not exclude the possibility of the condition. The exact cause

is unknown; some intrauterine conditions may play a role in the etiology.² Using the Friedens classification ACC is grouped into 9, type VI lesion affects the limbs, associated with Epidermolysis bullosa.³ We herein report a rare case of Aplasia Cutis Congenita associated with epidermolysis bullosa

CASE REPORT

Baby RJU was a female baby delivered by 25-year-old applicant through emergency caesarian section at gestational age (GA) of 42 weeks at University of

Calabar Teaching Hospital (UCTH), Nigeria. The indication was previous C/S with cephalopelvic disproportion (CPD) and postdate. She had been in labor for more than 10 hours prior to arrival. The mother was a gravida, 2 para, 2 both alive (one male, one female). Pregnancy was booked at GA of 26 weeks and antenatal period was uneventful. No risk factors for sepsis were observed. A 2.95 kg female baby was delivered with Apgar scores of 5, 4 and 8 at 1, 5 and 10 minutes respectively. All necessary resuscitative measures were instituted and baby remained stable after 5 minutes. Other anthropometric measurements were occipito-frontal circumference (OFC) 34 cms and length 48 cms.

On examination we observed skin defect affecting the right lower limb (leg and foot).



Figure 1: Skin defect at birth.

The skin defect on the shin measured 6 cm × 4 cm and dorsum of the foot 2 cm × 2.5 cm, other systemic examinations were essentially normal and there were no dysmorphic features or physical signs suggestive of post-date. The older sibling did not have similar defect and there was no history of such in the family. The father is a 35 year old secondary school teacher. The baby was admitted into the neonatal intensive care unit (NICU - Inborn unit) and managed as moderate birth asphyxia with aplasia congenita cutis.

The following investigations were requested with results; full blood count; PCV 40%, platelets $101 \times 10^9/l$, WBC 4.4×10^9 , neutrophil 22%, lymphocytes 78%; eosinophil 1% and platelets 300×10^9 . The wound swab was sterile and abdominal scan was essentially normal. X-ray of the right leg showed no evidence of fracture.

Baby was managed for moderate asphyxia according to the standard protocol of the unit. The lesion was dressed daily using gauze-impregnated with honey. The burns and plastic unit of the hospital was invited and upheld the line of management. Baby showed remarkable improvement within 2 weeks and was discharged home. Follow up by the 4th week showed completely healed lesion with only residual pigmentation Figure 1-3.



Figure 2: 2 weeks after treatment.



Figure 3: 4 weeks after treatment.

DISCUSSION

This is the first reported case of aplasia cutis congenita associated with epidermolysis bullosa presenting at our center, and to the best of the authors' knowledge this is the first from South-South region of Nigeria.

The index patient is classified into group 6 Friedens classification because of the loose fragment of skin around the edges of the lesion and blisters at the distal edge.³ The observed hypopigmentation may indicate a healed prenatal lesion that broke down. Milia were also observed on the dorsum of the foot. The above findings are therefore consistent with ACC associated with epidermolysis bullosa probably the dominant dystrophic type (DDEB). This was originally called Bart's syndrome. In view of the heterogeneity of ACC we could not completely establish if the condition is Autosomal dominant or recessive, though the parents claimed there was no history of such condition in the family. The lack of family history does not exclude the possibility of ACC

because it may occur as a result of new mutation. The mother did not use any antithyroid drugs, anticonvulsants or other drugs implicated in the etiology of ACC.^{2,4} In addition, the absence of viral infections and other inherited abnormalities, made us hypothesize that perinatal hypoxia may play a role in the pathogenesis of the disease. Involvement of the lower limbs are usually bilaterally, but asymmetric distribution has also been reported. Maya and Yakubu reported the first case in Jos, Nigeria with symmetric ulcers affecting both upper limbs.^{5,6} Our patient's x-ray did not reveal abnormalities of the bone beneath the ulcer. It has been reported that ACC in some cases are quite variable, beyond the loss of skin there can be complete absence of bones beneath the lesion.⁷ Rajabin and Aghaei reported the presence of congenital bone defect, syndactyly and brachydactyly of the toes of 9-year-old girl with ACC.⁸ The swab from the ulcer did not grow any organism, that is not surprising because the lesions are usually sterile. Hence the use of antibiotics was not necessary except if secondarily infected by bacteria. Laboratory results from our patient were all within normal limits. This is not unexpected because there is no specific laboratory abnormalities that have been documented to be consistent with this condition.⁹ Our diagnosis was based on the clinical presentation, as we could not demonstrate histological absence of skin due to diagnostic challenges. The lesion was dressed daily with natural honey. By the 2nd week the baby showed remarkable improvement and complete healing occurred by the 4th week of treatment. Baby did not require surgical therapy involving skin graft or local skin flap because there was no deep tissue loss.

CONCLUSION

ACC with associated epidermolysis bullosa is a rare unique heterogeneous presentation with skin fragility that responded to conservative treatment with honey within 4 weeks without scarring. We therefore recommend follow-up of patients with such conditions as well as educate their parents on the need to avoid aggravating factors.

Funding: No funding sources

Conflict of interest: None declared

Ethical approval: Not required

REFERENCES

1. Joseph G. Morelli. Cutaneous Defects. Robert M. Kleigman. Nelson Textbook of Pediatrics. Chapter 647. 18th edition. Philadelphia, PA: Saunders Elsevier; 2007.
2. Agata P, Anna S, Dorota P, Barbara K. Congenital skin aplasia on the lower limb in a premature infant with ELBW - case report. Italian J Pediatr. 2014;40:88
3. Frieden IJ. Aplasia cutis congenita: clinical review and proposal for classification. J Am Acad Dermatol 1986;14(4):646-60.
4. İçcimen A, Yardımcı G. Aplasia cutis congenita. J Turk Acad Dermatol. 2010;4(2):04201
5. Atik B, Tan O, Bayram I, Tuncer O, Kirimi E. Asymmetrical non-scalp aplasia cutis congenita: a case report. J Dermatol. 2004;31:923-6.
6. Maya, Y, Yakubu AM, Aplasia Cutis in a Nigerian child. case report. Niger J Paediatr. 2017;44(1):32-4.
7. Kruk-Jeromin, J, Janik, J. and Rykała, J. Aplasia Cutis Congenita of the Scalp. Dermatologic Surg. 1998;24:549-53.
8. Rajabian MH, Aghaei S. Adams-Oliver syndrome and isolated aplasia cutis congenita in two siblings. Dermatol online J. 2006;12(6):17.
9. Moros PM, Labay MM, Valle SS. Aplasia cutis congenita in a newborn: aetiopathogenic review and diagnostic approach. Ann Esp Paediatr. 2000;52(5):453-6.

Cite this article as: Ochigbo SO, Kingsley AI, Antigha CE, Jacob UJ. Aplasia cutis congenita associated with epidermolysis bullosa in a Nigerian child. Int J Res Dermatol 2018;4:80-2.