# Bullous aplasia cutis congenita: A rare presentation of a rare disease

Lauren Watchmaker[1] BA, Jacqueline Watchmaker[2] MD

Affiliations: [1]University of Wisconsin School of Medicine and Public Health, 750 Highland Avenue, Madison, Wisconsin, USA, [2]Department of Dermatology, Boston University, 609 Albany Street, Boston, Massachusetts, USA

*Corresponding Author: Jacqueline Watchmaker, 41 East Springfield Street, Boston MA 02118, Tel: 262-391-7464, Email: Jacqueline.watchmaker@gmail.com*

## Abstract

Aplasia cutis congenita (ACC) is a congenital disorder characterized by localized or generalized absence of skin. Bullous aplasia cutis congenita (BACC) is a rare clinical subtype that has few documented reports in the literature. Herein, we present a new case of BACC in which the bulla was unruptured at birth.

*Keywords: bullous aplasia cutis congenita, scalp lesions, bulla, pediatric dermatology*

## Introduction

Aplasia cutis congenita (ACC) is a rare congenital disorder characterized by localized or generalized absence of skin. It represents a heterogeneous group of disorders that have been classified on the basis of inheritance patterns and associated physical abnormalities [1]. Although ACC can present anywhere on the body, the majority of reported cases present as isolated scalp lesions [2]. Bullous aplasia cutis congenita (BACC) is a rare clinical subtype that has less than 20 published cases in the literature [3]. Herein, we present a case of bullous aplasia cutis in which the bulla was unruptured at birth.

## Case Synopsis

Dermatology consultation was requested to evaluate a lesion on the vertex scalp of a one-day-old, healthy, full-term, female infant born by cesarean section. The lesion was an 8mm circular, tense bulla filled with serous fluid with a focal area of hemorrhagic crust (**Figure 1**). The bulla was present at birth and remained stable in size over the first 24 hours. The patient had no other physical examination abnormalities. Given the negative maternal history of herpes simplex virus (HSV) and overall health of the infant, HSV was not suspected. The dermatology consultant recommended an ultrasound to assess for potential underlying malformations. The ultrasound revealed a fluid collection in the skin that extended only to the



**Figure 1**. *8mm circular, tense bulla filled with serous fluid with a focal area of hemorrhagic crust.*

subcutaneous fat. The diagnosis of BACC was made and conservative treatment with wound care was recommended.

## Case Discussion

Bullous aplasia cutis congenita is rarely seen in its initial form; most cases present as a flat scar after the bulla has ruptured in utero. In our case, we suspect the bulla was unruptured owing to caesarean birth rather than vaginal birth. The exact pathogenesis of BACC is unknown. It has been theorized that BACC is a form fruste of a neural tube defect [4], which could relate to genetic factors or environmental exposures. We recommend ultrasound for these lesions to evaluate for underlying abnormalities. If ultrasound cannot be performed because of the bullous nature of the lesion, MRI could be considered. Management is controversial but can include nonintervention, conservative treatments, and/or surgical intervention. Conservative treatment is typically recommended for small lesions especially if no underlying defects are found. There have also been reports of successful conservative management of large scalp defects with or without involving bones, though bone defects >5cm are unlikely to resolve on their own [5].

## Conclusion

Bullous aplasia cutis congenita is a rare subtype of aplasia cutis congenita with few documented cases in the literature. Dermatologists should consider BACC in their differential diagnosis for vesicobullous scalp lesions in newborns.

## Potential conflicts of interest

The authors declare no conflicts of interests.

## References

1. Frieden IJ. Aplasia cutis congenita: a clinical review and proposal of classification. *J Am Acad Dermatol*. 1986;14:646. [PMID: 3514708].
2. Demmel U. Clinical aspects of congenital skin defects. I. Congenital skin defects on the head of the newborn. *Eur J Pediatr*. 1975;121:21. [PMID: 765133].
3. Colon-Fontanez F, Fallon-Friedlander S, Newbury R, Eichenfield LF. Bullous aplasia cutis congenita. *J Am Acad Dermatol*. 2003;48:S95–8. [PMID: 12734490].
4. Drolet B, Prendiville J, Golden J, Enjolras O, Esterly NB. Membranous aplasia cutis with hair collars: congenital absence of skin or neuroectodermal defect? *Arch Dermatol.* 1995;131:1427-3. [PMID: 7492133].
5. Bharti G, Groves L, David LR, et al. Aplasia cutis congenita: clinical management of a rare congenital anomaly. *J Craniofac Surg*. 2011;22:159. [PMID: 21187755].