

**Case Report**

## APLASIA CUTIS CONGENITA: UNCOMMON FINDING OF TWO CASES OCCURRING IN ONE PATIENT IN TWO SUCCESSIVE DELIVERIES.

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**Abstract**

**Background:** Aplasia cutis congenita is a heterogenous group of disorders characterised by the absence of a portion of skin, with or without the absence of underlying structures such as bone, in a localized or widespread area at birth.

**Case presentation:** An uncommon case of delivery of a female baby with aplasia cutis congenita occurring in a patient who had a female baby with aplasia cutis congenita in her immediate past confinement. Both babies died within one week of delivery from suspected congenital cardiac abnormality.

**Conclusion:** Aplasia cutis congenita is an uncommon congenital abnormality, often times with good prognosis. It has a poor prognosis when there are other congenital abnormalities associated with it.

**Keywords:** Aplasia cutis congenita, Congenital cardiac abnormality, Prognosis.

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**INTRODUCTION**

Aplasia cutis congenita is a heterogenous group of disorders characterised by the absence of a portion of skin, with or without the absence of underlying structures such as bone, in a localized or widespread area at birth.<sup>1,2</sup> It was first reported by Cordon in 1767.<sup>3</sup> It most commonly affects the scalp, but can also affect any part of the body, including the trunk and limbs.<sup>3</sup> Aplasia cutis congenita is most often a benign isolated defect, but some people may have other congenital abnormalities involving the cardiovascular, gastrointestinal, genitourinary and central nervous systems.<sup>1</sup>

Aplasia cutis congenita is an uncommon abnormality of the newborn.<sup>3</sup> The incidence is approximately 3 in 10,000 births.<sup>3</sup> The cause of this condition is unclear and appears to be

Multifactorial.<sup>2</sup> There is no racial or sexual predilection for aplasia cutis congenita.

The prognosis is dependent on the severity of the associated abnormalities, as underlying or associated defects may significantly affect mortality and morbidity.<sup>3</sup>

**CASE PRESENTATION**

A female neonate was delivered at the labour ward of the Federal Medical Centre, Yenagoa at 38 weeks gestational age with absence of skin on parts of the right lower limb, exposing the subcutaneous tissues on those parts of the limbs. The neonate was immediately transferred to the special care baby unit (SCBU) of the facility for expert care. She had good Apgar scores and birth weight was 3 kg.

The mother was a 31-year-old primary school teacher with tertiary level of education. She was a

P5<sup>+2</sup> (2 alive). Pregnancy was booked for antenatal care at 15 weeks' gestational age, and was uneventful until delivery. The results of her investigations were all within normal limits. She did not take any other medications apart from her routine haematinics. She did not have varicella or herpes simplex viral infection during pregnancy. There was no family member that had similar features or had a child with similar features. In her last confinement in 2016, she was delivered of a live female baby who also had absence of skin on parts of the lower limbs. A diagnosis of aplasia cutis congenita was made then, and the baby died within the first week of life. Her second confinement in 2013 was complicated with intrauterine foetal death for which she had urgent Caesarean section due to positional cephalopelvic disproportion. She also had two premarital terminations of pregnancy at private clinics through manual vacuum aspiration at 6 weeks and 8 weeks respectively. She did not have known medical condition.

Musculoskeletal examination revealed well-demarcated ulcerations around the umbilicus and at the right lower limb extending from the mid-thigh to the foot (Figures 1 – 3). It involved the epidermis and dermis of the skin, which exposed the blood vessels in the subcutaneous tissue. A diagnosis of aplasia cutis congenita was made. Antibiotic ointment was applied on the lesion and covered with crepe bandage. About one hour after delivery, the neonate was noticed to be cyanosed with tachypnoea and tachycardia. A cyanotic congenital heart disease was suspected. Intranasal oxygen was commenced and the parents were counselled on the development. Arrangements were made to do an echocardiography for him. The neonate however died at the 12th hour of life at the SCBU. The parents declined autopsy despite counselling.



**Figure 1: Aplasia Cutis Congenita; showing the exposed subcutaneous tissue and blood vessels.**



**Figure 2: Aplasia Cutis Congenita; another view showing the exposed subcutaneous tissue and blood vessels.**



**Figure 3: Aplasia Cutis Congenita; another view.**

## DISCUSSION

The lesions of aplasia cutis congenita are noninflammatory and well-demarcated, ranging widely in size.<sup>3</sup> They may be circular, oval, linear, or stellate in configuration.<sup>3</sup> At birth, the lesions may have already healed with scarring or may remain superficially eroded to deeply ulcerated, occasionally involving the dura or the meninges.<sup>3</sup> The membranous type of aplasia cutis congenita is most common.<sup>3</sup> The defect may involve only the epidermis and dermis, which may result in mild scarring, or it may extend into subcutaneous tissue, or rarely periosteum, skull, and dura.<sup>3</sup> In this case, there were well-demarcated ulcerations around the umbilicus and at the right lower limb extending from the mid-thigh to the foot (Figures 1 - 3). It involved the epidermis and dermis of the skin, which exposed the blood vessels in the subcutaneous tissue.

The exact aetiology of aplasia cutis congenita is unclear. The condition is thought to be multifactorial.<sup>2,3</sup> The factors that may contribute to this condition include genetic factors; teratogens such as methimazole, carbimazole, misoprostol, and valproic acid; compromised vasculature to the skin; and trauma.<sup>2,3</sup> The mother of this baby was not on any of the above medications. The proximity of scalp aplasia cutis congenita to the scalp hair whorl, which is thought to be the point of maximum tensile force during rapid brain growth, has led to the hypothesis that tension-induced disruption of the overlying skin occurs at 10 – 15 weeks of gestation when hair direction, patterning, and rapid brain growth occur. This may also explain the increased incidence of aplasia cutis congenita on the vertex scalp.<sup>3</sup>

Early rupture of the amniotic membranes, forming amniotic bands, has also been implicated in the cause of aplasia cutis congenita in many cases.<sup>3</sup> The bullous or membranous variants of aplasia cutis congenita reveal a distinct histologic pattern identical to those in encephalocoeles and meningocoeles.<sup>3</sup> These types of aplasia cutis congenita may represent the incomplete or unusual forms of neural tube closure defect.<sup>3</sup> Aplasia cutis

congenita is typically sporadic; however, autosomal dominant and less commonly autosomal recessive cases have also been reported.<sup>3</sup>

The diagnosis of aplasia cutis congenita is made on physical examination.<sup>3,5</sup> The findings are indicative of an in-utero disruption of skin development.<sup>3</sup> The lesions usually occur on the scalp lateral to the midline.<sup>3,5</sup> They may also occur on the face, trunk, or limbs.<sup>3,5</sup> The lesions are well demarcated.<sup>3,5</sup> If they form early in gestation, they may heal before delivery and appear as an atrophic, membranous or fibrotic scar.<sup>3</sup> If they form later in gestation, they may appear as well-demarcated areas devoid of epidermis and dermis, thereby exposing the subcutaneous tissue and blood vessels.<sup>3,5</sup> There are no specific laboratory abnormalities found in aplasia cutis congenita.<sup>2,3</sup> Differential Diagnoses include epidermolysis bullosa and focal dermal hypoplasia syndrome.<sup>3</sup>

Treatment of aplasia cutis congenita varies depending on the condition of the infant. Conservative treatment is preferred.<sup>4</sup> Small areas usually heal spontaneously over time. Gentle cleansing and application of bland ointments or silver sulfadiazine can help prevent infection. If infection occurs, antibiotics can be used.<sup>3,5</sup> Larger lesions may require surgery.<sup>3,5</sup> The decision to use medical, surgical, or both depends on the size, depth, and location of the skin defect.<sup>3</sup>

Major complications of aplasia cutis congenita are rare, but can include haemorrhage, secondary local infection, meningitis, sagittal sinus thrombosis, bone affection and death. Complications can also result from associated congenital malformations involving the cardiovascular, gastrointestinal, genitourinary and central nervous systems, when present.<sup>3</sup>

The prognosis of aplasia cutis congenita is usually good, but if it is associated with other anomalies, the prognosis is dependent on the severity of the associated abnormalities.<sup>3</sup> Full-thickness defects of the scalp, skull, and dura are associated with a mortality rate of greater than 50%.<sup>3</sup> This baby was

suspected to also have a congenital heart abnormality when he became cyanosed, with tachypnoea and tachycardia. This diagnosis would have been confirmed on autopsy, but the parents declined despite profuse counselling. Aplasia cutis congenita is uncommon. However, the mother of this baby has now had two female babies that died from this condition. She is likely to be autosomal dominant for aplasia cutis congenita. That is why all her children were not affected. She was counselled for genetic studies, but declined because she is no longer interested in conception. She is presently on a long-acting reversible contraceptive.

### CONCLUSION

Aplasia cutis congenita is an uncommon congenital abnormality, often times with good prognosis. It has a poor prognosis when there are other congenital abnormalities associated with it like in the case presented. Preconception and antenatal care will play a major role in reducing the incidence of aplasia cutis congenita.

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