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# A Rare Case of Aplasia Cutis Congenita of the Scalp with Dual Skin Defects in a Non-Syndromic Preterm Newborn

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## Abstract

Aplasia Cutis Congenita (ACC) is a rare congenital condition characterized by localized or widespread absence of skin at birth. It most commonly affects the scalp and can be classified into different subtypes based on clinical presentation and associated anomalies. The exact etiology remains unclear, with genetic predisposition, vascular compromise, teratogen exposure, and maternal factors being implicated. This case is of a preterm male neonate, born at 35 weeks gestation to a 40-year-old primigravida mother with a history of hypothyroidism and long-term thyroxine use. The mother conceived via in-vitro fertilization (IVF) after undergoing extensive fertility treatments. At birth, the neonate exhibited two well-demarcated skin defects over the left posterior parietal triangle of the scalp, without involvement of deeper structures. Clinical diagnosis of non-syndromic membranous scalp ACC was made. Conservative management with saline cleansing and topical antibiotic application was adopted. The lesions healed completely by six months with residual atrophic scarring and a 'hair collar sign'. This case highlights a rare instance of isolated ACC in a neonate with a unique maternal history of hypothyroidism, prolonged thyroxine use, and IVF-conceived pregnancy. The successful conservative management further supports its role in appropriately selected cases.

**Key Words:** Aplasia Cutis Congenita, Newborn, Scalp aplasia

## Introduction

Aplasia Cutis Congenita (ACC) is a rare congenital condition marked by the absence of skin at birth, most commonly on the scalp, though it can affect other areas of the body.[1,2] Frieden classified ACC into nine subtypes based on the location and pattern of skin absence, inheritance pattern and associated anomalies while Sybertdefined six subgroups.[1-3] Scalp ACC is typically divided into membranous and non-membranous variants.[4] The condition occurs in about 0.5 to 1.0 per 10,000 live births and shows a female predominance (7:5).[3,5] ACC is usually diagnosed at birth when lesions, often translucent and ulcerative, are observed, with varying degrees of skin loss, sometimes extending to the underlying bone or dura which may heal or result in an atrophic scar.[1,6] The condition's exact cause is unclear, though factors such as genetic predisposition, vascular issues, and teratogen exposure are thought to play a role.[1,3] ACC management may involve conservative treatments or surgical intervention, depending on the severity of the lesion.[3]This case report presents a rare instance of a non-syndromic membranous scalp ACC with a double skin defect in a preterm neonate born to an elderly primigravida mother with a history of hypothyroidism and fertility treatments, which healed completely with conservative care.

## Case Report

A male infant was born at 35 weeks gestational age (GA) as a first child of a 40-year-old mother via planned elective lower segment cesarean section (LSCS) at Nepalgunj Medical College. The mother had a history of hypothyroidism for 15 years and was on long-term

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thyroid hormone replacement therapy (Thyroxine 50 mcg). She had been attempting to conceive for the past 10 years and underwent assisted reproductive techniques, including Fresh Egg Donation (ED) and in vitro fertilization (IVF). The IVF protocol included goserelin acetate stat on day 1, norethisterone 5 mg on day 1 and day 7 and regular intake of dydrogesterone 10 mg, estradiol 2 mg, aspirin 75 mg, low-dose prednisolone 5 mg and progesterone 8% gel. Along with this, she took PPIs, multivitamins (including iron, folic acid and calcium), and protein supplements regularly. She consumed a mixed diet and had no history of alcohol consumption or cigarette smoking. The marriage was non-consanguineous, and there was no significant family history of congenital anomalies on either paternal or maternal side.

The pregnancy was confirmed by USG. She had regular ANC visits, and USG was done every 3 months. There was no suspicion of twin pregnancy, and all investigations were normal. Serum T3, T4 and TSH levels were measured regularly, and she remained euthyroid throughout the period. Anomaly scan at 22nd week revealed no significant findings as well. She didn't have any history of significant infection or trauma during pregnancy. Placental examination revealed intact placenta weighing 450 grams. The fetal surface appeared smooth and normal. The maternal surface was complete without any signs of damage. A centrally inserted 3-vessel umbilical cord was observed, and no abnormalities were detected in its structure.

The infant was born with a birth weight of 3530 grams, a length of 52 cm, and a head circumference of 33.5cm. Apgar scores at 1 minute and 5 minutes were 8/10 and 10/10, respectively. The newborn did not sustain any birth injuries, and there were no feeding difficulties or other abnormalities noted. Breast feeding was started within an hour of delivery.

Upon examination, two oval/circular skin defects were observed on the infant's scalp over the left posterior parietal triangle, measuring approximately 1.5 × 1.0 cm and 0.5 x 0.5 cm. The lesions involved only the epidermis and upper dermis, without extending into deeper structures or bone (Figure 1). Clinical confirmation was done with a magnifying lens and dermoscope, which revealed translucent, atrophic skin with absent follicular openings and prominent telangiectatic vessels. No other apparent abnormalities were noted, and the overall clinical presentation was suggestive of an isolated case of scalp ACC.

A multidisciplinary team comprising pediatricians, pediatric surgeons, and dermatologists evaluated the case. The parents were counseled on the benign nature of the condition and informed that the lesion would likely heal spontaneously, and conservative approach was adopted. The initial treatment included cleansing with normal saline and topical antibiotic ointment application. After discharge, gentle water cleansing and the application of a topical antibiotic ointment (Mupirocin 2%) to prevent secondary infections were advised. A "wait-and-watch" approach was adopted, with monthly follow-ups to monitor and assess the healing progress and rule out complications.

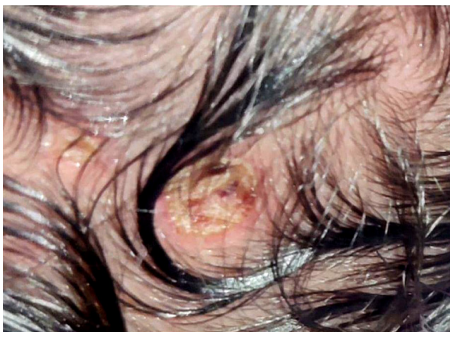

**Figure 1.** Two oval/circular skin defects were observed on the infant's scalp over the left posterior parietal triangle on 3rd day of life.

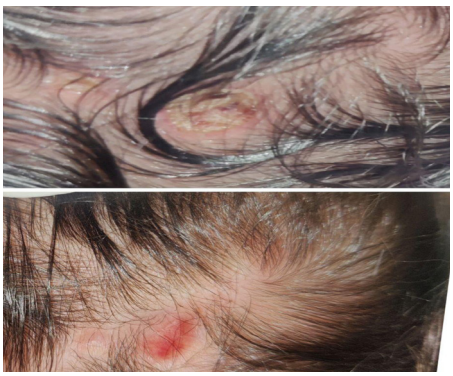

**Figure 2.** Both previous lesions had healed significantly and were covered with shiny erythematous slightly atrophic scar on the second month follow up

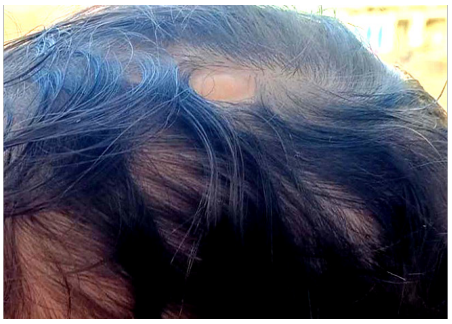

**Figure 3.** Typical 'hair collar sign' of the larger lesion on Six month follow up.

The newborn did not require intensive care and was discharged on day 3 with the mother in stable condition.

At the 2-month follow-up, both the lesions had healed significantly and were covered with shiny erythematous slightly atrophic scar, which was more evident in a larger lesion (Figure 2). Upon follow-up at 3 months, dermoscopy showed a well-defined atrophic white scar, reduced vascularity, and a persistent absence of follicular structures. By the 6-month follow-up, the larger lesion showed the typical 'hair collar sign' (Figure 3), a feature indicative of healed ACC lesion.

The child required no further medical or surgical management. During the follow-up period, no other anomalies or malformations were noted, and

anthropometric parameters along with developmental milestones were normal for his age. Given the stable clinical course and absence of alarming features, no radiological or histopathological investigations were advised.

## Discussion

ACC is a rare condition of defective skin development in utero occurring only in about 1 in every 10,000 to 20,000 live births.[3,5,7] Only about 500 cases of ACC have been mentioned in the literature so far.[8] A study conducted in Europe has shown its prevalence being as low as 5.10 per 100,000 births, which makes this condition very rare.[9] ACC is also less commonly seen among males compared to female neonates, making our case even rarer.[3,5]

This condition may coexist with other congenital anomalies and genetic syndromes or may occur as isolated defects such as our case.[1,8] The newborn was an isolated case of membranous scalp ACC and was classified as 'Group I : Scalp ACC without multiple anomalies' according to Frieden and 'Type I : ACC limited to the scalp' according to Sybert.[1,2] Scalp defect of ACC increases the risk of complications and also is considered an indicator of internal organ involvement, though in our case, the newborn showed no other defect or internal organ involvement and no complication occurred.[8,10] Nevertheless, complete effort should be made to investigate and rule out such comorbidities.

Potential causes of ACC include genetics, maternal intrapartum medications or teratogens exposure, placental infarcts, intrauterine infections and trauma, vascular compromise, amniotic membrane adhesion to the fetal skin, amniotic rupture sequence, ectodermal dysplasia, and imperfect neural tube closure. Although these factors have been deemed responsible for causing ACC, the exact pathogenesis remains unknown.[2,3,8,10-12] A unified theory is yet to be formulated. Heredity remains the only proven factor.[3] Most cases appear to be sporadic, but genetic or familial instances have also been reported with familial cases mostly showing autosomal dominant pattern of inheritance with partial penetrance and variable intrafamilial expression.[2,3,5,10] Positive family history was found only in about 2% in the Europe based study.[9] Genetic cause is also favored by the fact that other congenital anomalies or similar defects are present in twins or siblings and parents or grandparents in many cases. Advancing maternal age is found to be associated with some cases of ACC.[2]Fetal exposure to teratogens such as antithyroid drugs, cocaine, heroin, alcohol, misoprostol, methotrexate, ACE inhibitors, valproic acid, and benzodiazepines have also been hypothesized as an etiology.[3] Maternal cigarette smoking is found to be associated with occurrence of ACC in newborns.[8] There have been numerous instances linking maternal hyperthyroidism and antithyroid drugs to ACC.[11,12] More than 60 cases till date have been reported due to Methimazole or Carbimazole.[11]

In our case, mother's use of fertility medications (including goserelin acetate, norethisterone, dydrogesterone, estradiol, and progesterone) and ACC had no documented association. This draws attention to the need for further research regarding their potential effects on fetal growth and malformations. It has been suggested that maternal hyperthyroidism may itself act as a teratogen and cause congenital anomalies.[13] This, combined with the findings of our case should also raise questions regarding the potential teratogenicity of thyroxine itself. Hitherto, to our knowledge, there aren't any reports or published works associating maternal hypothyroidism and thyroxine to ACC. However, association between occurrence of ACC and maternal use of prednisolone has been reported which aligns with our case.[14]

Advancing maternal age is considered a potential risk factor for congenital anomalies, as it increases the likelihood of genetic mutations and chromosomal abnormalities.[2] Although significant association between maternal age and ACC has not been confirmed in the literature, this case favors a possible connection. Rates of congenital anomalies are also found higher among infants born to a consanguineous marriage.[15]

Management of ACC still remains controversial because of risk associated with both conservative as well as surgical approach. Conservative treatment carries the risk for delayed healing, infection and superior sagittal sinus hemorrhage or thrombosis. It is also unable to provide complete healing in bone defects with absent or abnormal dura. Contrastingly, risk of heavy blood loss, infections, scalp flap necrosis, skin graft loss, infection, persistent alopecia, anesthesia-related risks and potential brain damage remains in case of surgical treatment.[3]

Authors recommend choosing a management approach on the basis of size, location and structures at risk.[3] Conservative approach is indicated in smaller (2cm or less) superficial lesions with intact dura mater, without vascular malformations and sagittal sinus involvement. In contrast, larger, deeper lesions or association with dura and bone, CSF leakage, AV fistula and massive hemorrhage requires early surgical intervention.[3] Nonetheless, it is not true that larger lesions(>2cm) cannot be managed conservatively.[16] Harvey et. al. encountered complications among 2 out of 4 surgically managed cases and none among 13 cases managed conservatively.[16] Therefore, they recommend using conservative approach with cautious regular monitoring even for larger defects.

In this case, we adopted conservative management with regular gentle water cleansing and application of topical antibiotic ointment which aligns with recommendations mentioned in present literature for smaller lesions without involvement of deeper structures. No complication was encountered during the treatment, and the lesions had healed completely by the 6th month follow-up. The infant had also achieved all developmental milestones appropriate for his age. This case adds more evidence to the effectiveness of conservative approach, especially in

cases of smaller lesions without involvement of deeper structures or internal organs.

Scalp lesions may involve deeper structures such as bone or dura and may be associated with complications ranging from simple infections to life threatening conditions such as meningitis, hemorrhage, sagittal sinus thrombosis and even death.[5,10] So, proper evaluation of the case becomes necessary in order to instigate appropriate treatment regimens.[10] Even though our case involved only the epidermis and upper dermis, timely follow-up and careful monitoring was essential to rule out complications and evaluate the progress.

## Conclusion

We have described a rare case of Aplasia cutis congenita manifesting as two scalp defects on the left posterior parietal triangle which completely healed following conservative management. The mother's history of hypothyroidism and fertility treatments, including In-vitro fertilization (IVF), long term thyroxine use and advancing maternal age adds uniqueness to this case. ACC may occur as an isolated scalp defect. Although ACC is a rare condition whose exact pathogenesis remains unknown, it is essential to know key clinical features and management approaches. Serious complications may occur making regular follow-ups necessary. Our study underscores the effectiveness of conservative management and draws attention to the need for further research to better understand risk factors and etiology of ACC.

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