

Aplasia Cutis Congenita Mimicking as Scalp Birth Injury in a Neonate: Case Report

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Abstract

Aplasia cutis congenita (ACC) is a rare congenital skin defect. Exact aetiology of ACC is not well understood. Here, we report a case of aplasia cutis congenita mimicking as scalp injury at birth.

Keywords: aplasia cutis congenita; scalp; birth injury

Introduction

Aplasia cutis congenita (ACC) is a rare malformation. It occurs at the scalp, trunk and limbs, and in isolation or as part of group of syndromes [1]. It is a focal defect of cutaneous tissues ranging from an absence of skin to full thickness with bone and dura. Scalp lesions may associate with complications including infection, hemorrhage, thrombosis and seizures. Etiology of ACC is multi-factorial and genetics and exogenous factors possibly play role. Exogenous causes are placental infarcts, teratogens, intrauterine infections, trauma and neural tube defects. Lesions are self-limiting, but warns a workup to screen for underlying soft tissue anomalies [2].

Birth trauma is an injury to the newborn sustained during the process of birth with an incidence of 2-7 per 1000 live births [3]. It may vary from superficial injury of skin to life threatening problems [4]. In our case the newborn baby had congenital localized absence of skin over the centre of scalp region with well-defined margins. Initially it looked like as an avulsion injury by artery forceps during LSCS. It may cause an apprehension to the obstetrician, as the obstetricians are often not aware of this rare congenital skin defect.

A 29 years old female, G₃P₁L₁A₁ with previous lower segment cesarean section (LSCS) delivered, at term gestation at a tertiary care centre in Rajasthan, India by LSCS and gave birth to a 2.5 kg male baby. Antenatal period was uneventful. Though, history of anti-tubercular therapy before she conceived and of beta blockers during 3rd trimester for hypertension was there. We noticed that the newborn had a single skin lesion of about 2X2 cm diameter, oval in shape with well-defined margins involving epidermis and dermis on the scalp in the midline at lambda point of scalp (Figure 1). At first the lesion was wrongly taken as instrumental birth injury. The newborn had normal agar scores n vitals were stable. Later, clinical diagnosis of aplasia cutis congenita was made and the baby was admitted in NICU for further evaluation. There was no history of ACC causing drugs during pregnancy. As mother of our case took ATT and beta blocker, we did not find any association of these with ACC in available literature. Other associated anomalies were ruled out as ultrasonography scan was normal. As the lesion was small, less than 4 cm, local cleansing with topical anti-bacterial ointment was done and the baby was put on regular dermatology OPD follow up. Later at 6 weeks of age the lesion was completely filled without any local complication.

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Figure 1: Single skin lesion of about 2X2 cm diameter, oval in shape with well-defined margins involving epidermis and dermis on the scalp in the midline at lambda point of scalp.

Discussion

Aplasia cutis congenita (ACC) is an uncommon anomaly of newborns with an incidence of approximately 1 to 3 out of 10,000 births [5]. Gender predilection is not reported. It manifests as a solitary defect on the scalp in 70% of cases, but it may also occur as multiple lesions. Although commonly seen on the scalp yet it can affect any part of the body. Lesions are non-inflammatory and well-demarcated with variable size. They may be circular, oval and linear in shape. At birth, the lesions may have healed, may superficially erode and may deeply ulcerate. Defects in the skin that form early in gestation may heal before delivery and appear as an atrophic, membranous, bullous, or parchment like scar with associated alopecia, whereas less mature defects present as ulcerations. The membranous type of aplasia cutis congenita is most common.

The exact pathophysiology of ACC is unclear. The most commonly accepted theory focuses on the tension that prevents the skin from converging during development in utero. Proposed mechanisms include intrauterine trauma, vascular compromise, infection, and medications. Known medicines are methimazole, misoprostol and valproic acid. Aplasia cutis congenita is mostly sporadic, though autosomal dominant and recessive cases have also been reported. Mutations in the ribosomal GTPase BMS1 have been identified. Newborns with ACC need to be thoroughly examined for associated anomalies including bone and intracranial malformations. Ultrasound imaging can be used to evaluate for underlying defects [6]. Further tests may be indicated for evaluation of associated

conditions. Differential diagnoses are traumatic lesions, localized scalp infections, dermoid cyst, isolated encephalocele, meningocele and nodular neuronal heterotopia.

ACC management in neonates is based on the size of skin defect. For small lesions (<4cm), daily cleansing with topical antibacterial ointment is recommended until healing is complete [8]. Lesions typically heal within a few weeks to a few months with an atrophic hairless scar [7]. Larger lesions (>4cm) are more commonly associated with underlying defects and are at an increased risk of complications, including hemorrhage, venous thrombosis, and infection. Early surgical repair is recommended to avoid these complications. Skin grafting or flap techniques are commonly utilized as some lesions can be several centimeters in size.

The prognosis for aplasia cutis congenita (ACC) is usually excellent for small skin defects. The recovery is uneventful, with gradual epithelialization and formation of a hairless, atrophic scar over several weeks. The average length of recovery for patients treated conservatively is 27.9 days [8]. Small underlying bony defects usually close spontaneously during the first year of life. Surgical repair of large or multiple scalp defects with excision and primary closure may be considered. If aplasia cutis congenita is associated with other anomalies, the prognosis is dependent on the severity of the associated abnormalities.

Conclusion

This case report retraites the importance of antenatal history, careful examination of skin lesion, and pediatrician opinion before labelling it as birth injury.

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