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Case Report
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Aplasia Cutis Congenita of the Scalp: a Report of Three Cases

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ABSTRACT

Aplasia cutis congenita is an uncommon dermatological anomaly in which part of the skin is absent at birth due to a defect in the intrauterine skin development but usually is of unknown etiology. The most common site of the defect is the scalp but can exist in other parts of the body. In this study we would like to report 3 cases of aplasia congenita of the scalp which are seen in Tripoli, Libya hospitals. One of them is believed to be a result of premature rupture of the membranes while no cause was found for the other two cases. The main aims of this study is to highlight the presence of this condition in neonates in Tripoli Hospitals and to emphasize the importance of skin inspection at birth for early detection of the condition in order to prevent secondary infection, avoid scaring disfiguration of the lesion and to apply an early management of the skin defect when it is required.

Keywords - *Aplsia cutis congenital; Scalp; Skin defect.*

INTRODUCTION

Aplasia cutis congenita (ACC) is defined as an absence of the skin in a localized or widespread area at birth[1,2] It is most commonly presented as a solitary defect of the scalp but it may also occur in other areas of the body. Other cases may be presented as multiple lesions in the scalp and different parts of the body such as the face, the trunk and the limbs. Lesions are usually non-inflammatory and well demarcated.[1] Incidence of the disorder is about 2.8 per 10000 liveborns.[3] There are neither racial or sexual predilection.

Diameter of the lesion of ACC of the scalp ranges from 0.5 to 10 cm. Its shape is usually circular, oval, linear or stellate.[1,4] Most of ACC lesions of the scalp occur in the vertex, just lateral to the midline.[5,6] The depth of the ulcer may involve the epidermis and the outer part of the dermis resulting in minimal alopecic scarring, but it may involve more deeper structures such as the periosteum, the skull bone and the dura.[5,6] At birth, the lesion may have already healed with scaring or remain superficially eroded, but can be deeply ulcerated. The healed lesions may appear atrophic, membranous or parchment like scar which is associated with alopecia. When multiple, lesions can be symmetrical.

ACC is a developmental inherited defect which may occur at the embryonic stage when the skin does not grow. Although etiology of ACC of the scalp is unknown, it was proposed that the defect at the center of the hair growth 'whorl' of the scalp can be due to stretching of an area of the skin as the brain and the skull underneath expand rapidly. The excessive stretch squeezes cutaneous blood vessels supplying the affected area of the scalp. When the stretch is beyond the skin elasticity, skin might rupture at the whorl causing the defect. ACC in general can also be due failure of the neural tube development during the embryonic stage which can be a consequence of DNA defect on a certain chromosome.[7-13] Many teratogens are reported to be associated with ACC such as maternal viral infections during pregnancy such as herpes simplex, varicella zoster and others.[11,14,15] Many drugs when administered during pregnancy, especially during the first trimester, can be teratogenic and may cause ACC such as metimazole, valproic acid and carbamazepine.[11,16]

Prematue rupture of the membranes which forms amniotic bands can also be the cause of some cases of ACC. Fetus papyraceous and placental infarction can also be associated with ACC.[17-21] ACC lesions are sometimes mistakenly attributed to birth trauma secondary to vacuum extraction, forceps delivery or fetal scalp monitor electrodes.

ACC is often a benign isolated defect, but it can be associated with other malformations and diseases such as limb abnormalities, cleft lip and palate, double uterus, polycystic kidneys, mental retardation, epidelmolysis bullousa and raves disease.[22-24] Frieden proposed a classification for ACC consisting of 9 groups based on the number and locations of the lesions, and the presence or absence of associated malformations (Table 1).[25] If the skin defect of the ACC of the scalp is small, a gradual recovery with epithelization and formation of a hairless atrophic scar takes place over few weeks. Gentle cleaning, bland ointment and hydrocolloidal dressing may be occasionally needed when erosion is a problem to prevent desiccation of the defect, and to promote healing of the lesion.[26] Antibiotics are only indicated if there are overt signs of infection. Small underlying skull bone defect usually closes spontaneously during the first year of life. Surgical repair of large, or multiple scalp defects can be undertaken if possib le with excision and primary closure or with the use of tissue expanders and rotation of a flap.[27,28] Truncal and limb defects despite being large in size, usually epithelize and form atrophic scars, which can be dealt with later on by plastic surgery.

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Table 1: Classification for Aplasia cutis congenita (ACC) as proposed by Frieden.[25]

Group	Description
Group 1	ACC of the scalp not associated with other anomalies. Accounts about 86% of the solitary lesions.
Group 2	ACC of the scalp associated with limb anomalies such as hypoplastic or absent distal phalanges. Adams-Oliver syndrome is a distinct subtype of this group in which there is an autosomal dominant inheritance pattern. Other associated anomalies include cutis mormorata telangiactasia congenital, hemaniomas, cranial arteriovenous malformation, skin tags and wooly hair.
Group 3	ACC of the scalp with epidermal and subcutaneous nevi, ophthalmic and neurological malformations. Findings can be typical of the epidermal nevus syndrome, including seizures, mental retardation, corneal opacities, and eye lid colobomas. It is not familial.
Group 4	ACC overlying deeper embryological malformations such as meningomyelocele, porencephaly, lepomeningeal angiomatosis, cranial stenosis, spinal dysgraphism, gastroschisis and amphalocele. The inheritance pattern varies with the associated underlying condition.
Group 5	ACC associated with fetus papyraceous (death of the twin fetus early in the pregnancy) or placental infarction. It usually appears as an extensive truncal or limb lesions which is linear or stellate. The affected neoborn is usually normal.
Group 6	ACC associated with epidermolysis bullousa (initially described as Bart's syndrome). Usually affects the lower extremities. It may be associated with pyloric stenosis, duodenal atresia, craniofacial abnormalities and nail dystrophy.
Group 7	ACC localized to the extremities (upper and lower limbs) without epidermolysis bullousa. It can be familial.
Group 8	ACC caused by exposure to teratogens such as intrauterine infection with herpes simplex and varicella zoster and administration of some drugs during pregnancy such as methimazole and valproate.
Group 9	ACC associated with malformation syndromes such as trisomy 13, 4p-syndrome, setleis ectodermal dysplasia, Johnson-Blizzard syndrome, focal dermal hypolasia (Goltz syndrome), amniotic band disruption complex, oculocerebrocutaneous syndrome and 46 XY gonadal dysgenesis.

The presented cases report includes three cases of ACC of the scalp as follows:

Case Report 1

A male Libyan baby, a product of consanguinity marriage. Delivered by an elective cesarean section due to postdate, and previous cesarean section. His birth weight was 3.1 kg. There was an absence of the skin of the scalp which appeared as a large solitary diamond shaped ulcer (diameter of) 7x8 cm and a depth of 0.4 cm), red, eroded, well demarcated and non-inflamed. The ulcer involves epidermis, dermis, subcutaneous tissue, periosteum and parts of the parietal bone. Physical examination of all systems showed no abnormal findings but X-ray of the skull showed bone defect. The baby was discharged in a good condition, and to be followed up in the outpatient clinic.

Case Report 2

A male Libyan baby, a product of nonconsanguinity marriage, delivered by caesarean section with umbilical cord around his neck. Mother aged 41 years, and was suffering during pregnancy from recurrent urinary tract infections which started from the first trimester. Was complaining also from an odorless vaginal discharge. Viral screen of the mother proved negative. The baby was apneic, has irregular breathing, but he was pink, active, not distressed and feeding well, APGAR score was 6 at 1min and 9 at 5 min after birth. His birth weight was 3.45 kg, his head circumference was 34 cm. There was an absence of the skin of the scalp which appeared as 3 oval shape abrasions of variable sizes (diameters: 1x2 cm, 3x4 cm and 4x4 cm). They were well demarcated, had irregular edges, allopecic and non inflamed (Figures 1).

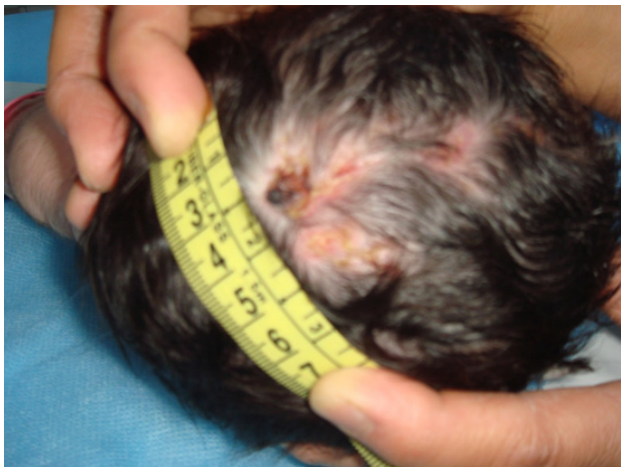

Figure 1: Size of scalp lesion

Physical examination of all systems showed no abnormal findings, but noncontrast axial CT scan of the brain revealed skull deformity; with irregular parietal bone and cavum septum pellocidum.

Case Report 3

A female Libyan baby, a product of consanguinity marriage. Birth was through normal vaginal delivery, from a 26 year old mother, who had premature rapture of the membranes. Viral screen of the mother proved negative. The baby was apneic, has irregular breathing, but she was pink, active, not distressed and feeding well, APGAR score was 9 at 1 min and 10 at 5 min after birth. Her birth weight was 3.2 kg, head circumference was 35.5 cm. There was an absence of the skin of the scalp which appeared as a large solitary ulcer that has oval shape (diameter of 9x7 cm), red, eroded, well demarcated, none inflamed and alopecic.

The baby has a tuft of hair at the sacral region of the vertebral column. Physical examination of all systems showed no abnormal findings. However, skull *X*-ray (Figure 2) showed a bone deformity, and MRI of the brain revealed dilated extra-axial cortical vein within the left cerebral hemisphere at the parietal bone convexity; with no abnormality in the intra-axial brain parenchymal intensities. The baby was discharged in a good condition, scalp lesions were dry and to be followed up in the outpatient clinic.

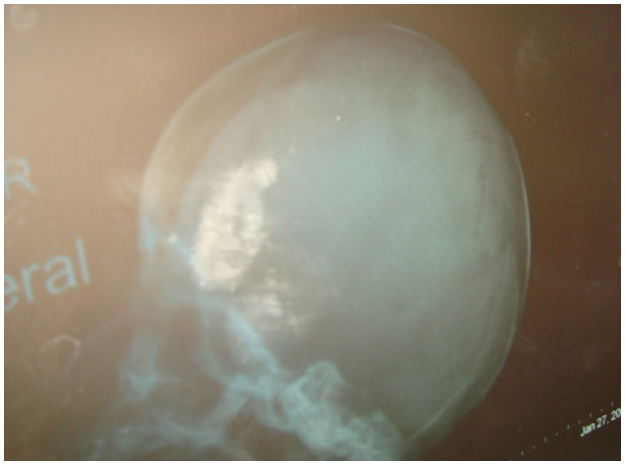

Figure 2: Skull *X*-ray of case 3

DISCUSSION

The reported three cases are ACC of the scalp in which the skull bone is also defective but no other congenital abnormalities are detected. The etiology in these three cases is unknown. ACC of case 3 could be due to prematue rupture of the membranes which forms amniotic bands. When the defect is small, it usually heals by epithelization forming a hairless atrophic scar which could be the case in the reported cases. The associated small bony defects of the skull in case of ACC of the scalp, usually closes spontaneously during the first year of life. Presence of the hair tuft in case 3 may be associated with underlying spine defects although that was not reported by the MRI.

Although in most of the cases the prognosis is good, an early intervention is required in a few to prevent complications such as hemorrhage, local infection, meningitis and sagittal sinus thrombosis. When ACC of the scalp is associated with a skull bone defect, meningitis and hemorrhage from saggital sinus can have serious consequences. Prognosis depends also on the associated malformation syndromes, diseases and the timing and choice of the intervention when required.

When inheritance can be found, genetic counseling should also be provided to parents. It prevents ACC, and the associated malformation anomalies. Screening for viral infections and advice not to take known teratogenic drugs during pregnancy, can also be an important approach to reduce the incidence of ACC.

CONCLUSION

This cases report has highlighted the presence of this condition in neonates, and in hospitals in Tripoli. It should emphasize the importance of skin inspection at birth, for early detection of the condition. To prevent secondary infection, avoid scaring disfiguration of the lesion, to apply early management of the skin defect when it is required, and to offer genetic counseling to the parents and relatives when a familial link is suspected.

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