Isolated Aplasia cutis congenita :Case report of two cases

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Abstract

This paper demonstrated two cases of Aplasia cutis congenita diagnosed after birth. No abnormalities were seen during the antenatal examination of the fetus during pregnancy. One case was treated surgically and the other conservatively, both with a good prognosis.

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Key Clinical Message

Congenital skin dysplasia, especially isolated scalp defects, are difficult to detect prenatally. The prognosis for isolated congenital scalp defects is good. Treatment options include conservative treatment and surgery. The choice of treatment depends on the patient's individual circumstances.

Abstract: This paper demonstrated two cases of Aplasia cutis congenita (ACC) diagnosed after birth. No abnormalities were seen during the antenatal examination of the fetus during pregnancy. The newborn with the larger scalp defect underwent emergency debridement and suturing combine with local flap transfer immediately after birth and recovered well one month after surgery. The newborn with a smaller scalp defect was treated conservatively after birth with local disinfection, regular dressing changes, and eventually, a scab was formed, which healed entirely after debridement one month after birth. Two children had no hair follicle growth after the healing of the scar. Aplasia cutis congenita may be associated with congenital developmental defects, excessive skin tension, intrauterine infection, or genetics. Early intervention is crucial, and the decision to treat conservatively or surgically should be made concerning the child's general condition, location, and size of the skin lesion.

Keywords : congenital; skin dysplasia; scalp defects

1 Introduction

Aplasia cutis congenita (ACC) is a rare congenital skin defect with an incidence of approximately 3/10,000, characterized by localized or extensive loss of epidermis, dermis, and occasionally subcutaneous tissue[1]. The exact etiology of ACC is unknown. The disease presents as isolated or multiple lesions that can appear anywhere on the body, but 70% to 90% of lesions are localized to the scalp's vertex[1-3]. Most ACC tends to heal spontaneously, and its fatal complications include sagittal sinus hemorrhage, meningitis, and brain herniation. The treatment of ACC is controversial. Surgical treatment includes debridement and suturing, local flap transfer, free flap transfer, and skin grafting[2]. The risks include anesthesia-related complications, intraoperative bleeding, postoperative infection, and flap necrosis[4]. Conservative treatment mostly consists of regular dressing changes to promote spontaneous epithelialization, and its risks include bleeding, prolonged healing time, and wound infection[5].

2 Case report

Case 1

The newborn, male, was admitted with "scalp defect found after birth", mother, 37 years old, father, 40 years old, G3P2, 39 weeks + 1 day of gestation, born by cesarean section due to "scarred uterus", birth weight 4210g, Apgar score of 9-9-9, clear amniotic fluid, 600 ml of amniotic fluid, one turn of the umbilical cord around the neck, and no abnormality of the placenta. After birth, a 3*2.5 cm scalp skin defect was found on the top of the head, and the skull was visible without bony defects, and the diagnosis was Aplasia cutis congenital. Ultrasound in early pregnancy and systemic ultrasound in mid-pregnancy showed no abnormalities, and Down's syndrome screening in early and mid-pregnancy showed no abnormalities.Amniocentesis was recommended for "advanced maternal age" and the karyotype and chromosome microarray of the amniotic fluid showed no abnormalities. There was no previous history of a similar disease in the child's family, and the child's mother had no history of specific drug exposure during pregnancy. After being informed of the condition, an emergency debridement and suture combined with local flap transfer was performed under general anesthesia with tracheal intubation. The operation was successful, and postoperative ceftizoxime sodium was used for anti-infective treatment, and wound dressing was changed. The wound was discharged on the 7th postoperative day, and the stitches were removed two weeks after surgery, and the wound healed well on the postoperative review one month later. On review three months after surgery, the child had no hair follicle growth at the original skin defect on the top of the head and was growing well. (Figure 1)

Case 2

The neonate, male, mother 23 years old, father 28 years old, was born in our hospital by cesarean section due to " macrodome ", with birth weight 4230g, Apgar score: 9-9-9, clear amniotic fluid, volume 500 ml, normal umbilical cord placenta. After birth, a skin defect of 1.5*1.5 cm was found on the head's top, with a depressed scalp and no local exudation, which was diagnosed as a congenital scalp skin defect. The mother of the child had no abnormal maternal examination during pregnancy. Mother has normal Down's syndrome screening results in early to mid pregnancy.There was no history of similar diseases in the child's family, and the child's mother had no history of exposure to specific drugs and teratogenic substances during pregnancy. After consultation with dermatology and pediatrics, the child was treated conservatively with topical antibiotic ointment and other medications applied to the skin defect. The scalp defect scars were well healed, and there was no hair follicle growth in the healed area. The child was growing well at three months postnatally. (Figure 2)

3 Literature Review

Literature search methods:

The literature search was conducted through the China Knowledge Network Data Service and Pubmed. The keywords for the Chinese literature search were "先天性皮肤发育不全" and "孤立性头皮缺损", while the keywords for the English literature search were "aplasia cutis congenita "and "scalp defect". Exclusion criteria: (1) Patients with congenital dyschondroplasia syndrome. (2) Cases with unknown diagnosis and treatment history and incomplete basic information. (3) Repeated cases reported by the same author at the

same institution.

RESULTS: A total of 12 neonates with isolated scalp defects meeting the inclusion criteria were reported nationally and internationally from June 2015 to June 2020 (see Table 1). Among the twelve children, five were male. The largest scalp defect was 9.0*10.0 cm and the smallest was 0.8*1.0 cm. Two cases were treated surgically, one case was readmitted for surgery after conservative treatment due to intracranial haemorrhage in the child, and the rest were treated conservatively. All patients survived and no fatal cases were reported.

4 Discussion

4.1 Etiology

ACC was first reported by Gordon in 1767 and summarized by Campbell in 1826[6]. It is occurring mainly on the top of the head but can occur anywhere on the body[5]. The cause of ACC is unknown. Genetics and exogenous factors may play a role in the development of the lesion. Suspected exogenous causes include intrauterine trauma, localized amniotic adhesions, and exposure to teratogenic substances such as antithyroid drugs, valproic acid, marijuana, heroin, alcohol, and cocaine during pregnancy[7]. The occurrence of ACC has also been associated with skin tears due to excessive tension in the fetal skin and subcutaneous tissues, with the top of the head being the most tensioned area[8]. The only thing that has been established is that the occurrence of congenital dermal dysplasia is genetically related. Most of these lesions are disseminated, but approximately 25% are autosomal dominant or recessive. Other genetic abnormalities associated with congenital dermal dysplasia include autosomal aberrations, deletions, trisomies and mutations.No identified genetic target for ACC had been identified, but a recent study suggests a possible role for the BMS1 gene. In a 2013 study that examined five generations of autosomal dominant ACC, mutations in the BMS1 gene were found to play an essential role in skin morphogenesis[9]. ACC can also be associated with various genetic syndromes such as ADAMS-Oliver syndrome, Bart syndrome, and Setleis syndrome. There was no family history of genetic predisposition or exposure to drugs and teratogenic substances during pregnancy in both cases. The same point is that both are macrodome, and it is considered that the fetus grew rapidly during pregnancy, causing congenital skin dysplasia due to hypertonicity of the skin tissue on the top of the head.

4.2 Pathology

The diagnosis of ACC is usually made only by clinical examination. Because many patients refuse to undergo local biopsy and cannot be diagnosed pathologically, the diagnosis can only be made based on the patient's age, demographics, and typical scalp involvement. The two children in this article refused the pathological examination. This disease's pathological presentation in the literature is as follows: histopathological manifestations of non-healing lesions include epidermal and dermal loss and vascular proliferation. Scar tissue formed at the top of the patient is healed head defect shows a thin or flattened layer of the epidermis but no attachment structures.

4.3 Treatment

Currently, there are two classification systems commonly used for ACC, namely the Sybert classification system and the Frieden classification system[10]. The Sybert classification system classifies ACC into six subtypes based on defect location and concomitant symptoms. However, it does not mention the complementary treatment and prognosis. Frieden further analyzes the associated abnormal manifestations of ACC, provides insight into the genetic pattern, and emphasizes the associated pathogenesis. The treatment of patients with ACC depends on the lesion's size and the presence of underlying defects. For small lesions (less than 2 cm) with no additional findings, daily cleaning of the lesion with topical antimicrobial ointment is recommended until healing is complete. Lesions will usually heal within a few weeks to a few months, leaving an atrophic, hairless scar. More extensive lesions (greater than 4 cm) are more commonly combined with underlying defects and have an increased risk of complications, including bleeding, venous thrombosis, and infection[11]. Early surgical repair with either a skin graft or a flap repair technique is recommended to avoid these complications[11]. For lesions between 2-4 cm, conservative treatment is an option and surgical treatment. Conservative treatment includes applying various dressings, continuous rehydration, topical

antimicrobial agents, and systemic antibiotics to promote spontaneous epithelialization. Conservative treatment should first be performed in a neonatal intensive care unit with the right monitoring conditions, and then the child can be cared for at home once the condition is stabilized[11]. The advantages of conservative treatment are that it avoids the risks associated with surgery and complications in the donor area. The disadvantages are that it may cause hemorrhage, meningitis, and loss of body fluids from the wound leading to hyponatremia causing brain herniation and seizures. Surgical treatment of congenital scalp defects includes simple debridement and suturing, flap transfer, autologous or allogeneic skin grafting, early or delayed cranioplasty, and tissue expansion. Although the literature reports that cranial defects can be rapidly regenerated, most scholars recommend repairing the skin along with the cranial defect for optimal results[3-5, 7, 8]. The surgical treatment complications are mainly due to general anesthesia for tracheal intubation of neonates and the inherent high risk of complex surgical operations[9]. With medical technology advancements, surgical complications are relatively low, and most of them can be effectively controlled. Compared with conservative treatment, children who underwent surgery had a significantly shorter healing time and fewer postoperative complications. In this report, two children, one treated surgically and one treated conservatively, had good healing of the scalp defect without local or systemic complications.

5 Conclusion

Most isolated ACCs without other underlying defects in combination can have a relatively benign outcome[1]. However, when complications occur, the risk of mortality increases dramatically. The most common life-threatening complication of ACC is sagittal sinus hemorrhage, seen as a lesion near the scalp[3]. Another potential complication of ACC includes secondary infection of the lesion. Patients are at increased risk of developing skin infections due to the absence or impairment of the skin's barrier to environmental microorganisms. If not treated properly, severe infections can develop into meningitis. When scalp defects are large, timely surgery can help prevent these complications. ACC patients' skin is replaced by smooth, gray, parchment-like tissue after healing, with no hair growth. Those with joint deformities are prone to a disability, and those with skin defects that cross the joints may form scar contractures that interfere with function.

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