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Review

Aplasia Cutis Congenita

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Abstract

Observations: Aplasia cutis congenita (ACC) is a rare disease which can be seen on any part of the body, but most common on the scalp. Its pathogenesis is not fully understand. Many factors have been accused such as chromosomal abnormalities, amniotic defects, intrauterine problems, thrombotic events, or teratogens. The treatment of the aplasia cutis congenita possesses a special importance because of high complication risk.

Introduction

Aplasia cutis congenita (ACC) is an uncommon malformation characterised by localized absence of skin [1]. It was first described by *Cordon* in 1767 [2], but the first scalp lesion was described by *Campbell* in 1826 [3].

Epidemiology

Until today, approximately 500 cases have been reported since 1767 [4]. The incidence of this disease is 0.5/10,000 to 1/10,000 newborns [5]. The disease is more common in females according to males, ratio of about 7:5 [6].

Etiology

The causative mechanism is exactly unknown and many different etiologies have been postulated [2]. These include chromosomal abnormalities, amniotic defects, intrauterine problems, thrombotic events, or teratogens [7]. Different teratogens used in pregnancy like Misoprostol, Cocain, Methotrexate, Angiotensin- converting enzyme inhibitors, methimasol, and benzodiazepines have been accused [8]. There is also a case report concerning low molecular weight heparin usage during pregnancy and consequent aplasia cutis on the scalp region [9]. Some viruses such as herpes simplex virus or chickenpox that cause congenital infections have also been accused [2]. Aplasia cutis congenita can also be associated with some genetic syndromes like trisomy 13, the 4p(-) syndrome [6]. Furthermore, it can be associated with numerous structural abnormalities such as cleft lip palate, limb reduction defects, epidermolysis bullosa, duodenal atresia, patent ductus arteriosus, omphalocele, polycystic kidney, gastroschisis and neurological malformations [10].

Clinical Findings

Aplasia cutis congenita is usually (86%) present as a solitary lesion over the cranial vertex just lateral to the midline, but about 15% of all cases, can also appear on other parts of the body areas, such as face, trunk, or limbs [**Figure 1**] [1, 5].

Bigliardi et al. reported a case involving the lower limb unilaterally [11] and *Verhelle* et al. reported another case located bilaterally on the abdominal skin [1]. Two cases of systemic

Table 1. ACC Classification According to *Frieden* [8].

Type	Characteristics	Inheritance
Type 1	Scalp ACC without multiple anomalies	AD or Sporadic
Type 2	Scalp ACC associated with limb abnormalities	AD
Type 3	Scalp ACC associated with epidermal or organoid nevi	Sporadic
Type 4	ACC overlying embryological malformation	Depends on underlying condition
Type 5	ACC associated with fetus papyraceus or placental infarcts	Sporadic
Type 6	ACC associated with epidermolysis bullosa (EB)	Depends on EB type: AD or AR
Type 7	ACC localized to extremities without blister	AD or AR
Type 8	ACC caused by specific teratogens	Not inherited
Type 9	ACC associated with malformation syndromes	Varies, depending on specific syndrome

AD: Autosomal Dominant; AR: Autosomal Recessive

aplasia cutis has also been reported in the literature [12, 13].

The scalp is the most common site of location for this condition, and the lesions are generally small, superficial [14], well-circumscribed, not inflamed, and vary in size 0,5 to 10 cm or larger [15]. The lesions vary in shape and circular, oval, linear, or stellate configurations can be observed [16]. Two main clinical variants of aplasia cutis of the scalp are accepted: (i) membranous type which shows a small, membrane-like surface, round or oval in shape, with or without a collar of hair, (ii) nonmembranous or irregular type which is larger, irregular in shape, and showing stellate defects [17, 18]. In some cases, the lesions may be associated with aplasia of the underlying skull. This situation can be seen when more larger defects are present [19]. A significant higher risk for involvement of the skull has been reported if the defect is larger than 1 cm^2 in diameter [6]. The most important complications of large scalp defects are infections and hemorrhage which present mortality rates of 20-30% [14]. Small lesions without involvement of bone tissue heal slowly with gradual epithelialization. If underlying bone tissue is affected, risk of complications and the mortality increases. This complications include sagittal sinus hemorrhage or thrombosis, focal infection of the lesion site or meningitis. Deaths are often due to sagittal sinus hemorrhage [20].

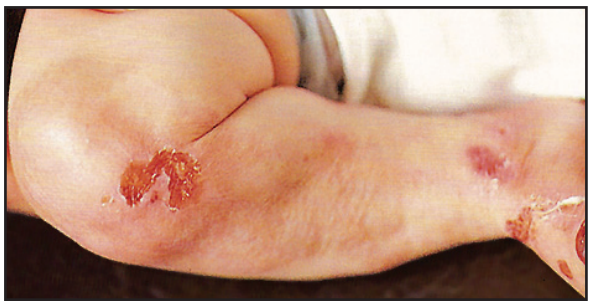

Figure 1. Aplasia cutis congenita can be seen rarely on the lower extiremity

The causative mechanism is exactly unknown and many different etiologies have been postulated [2]. These include chromosomal abnormalities, amniotic defects, intrauterine problems, thrombotic events, or teratogens [7]. The diagnosis is usually easy and clinic and histopathological features are distinct [10]. There is no epidermis or it is very thin, the underlying dermis is weak and consisted of "loosely arranged" collagen bundles and the subcutis is also thin in most of the cases [11]. Elastic fibers and dermal papillae are absent and blood vessels maldeveloped [14].

Most of the cases are sporadic but familial cases with an autosomal dominant and recessive inheritance have also been reported [8, 21]. *Frieden* proposed a classification scheme for ACC in 1986 [22]. According to this classification there are nine subgroups differing in clinical presentation, prognosis, and the associated conditions (**Table 1**) [8].

Diagnosis can be established during pregnancy [5]. Aplasia cutis congenita should be considered if the α-fetoprotein level in amniotic fluid and maternal blood has increased, or a positive acetylcholinesterase test in the presence of normal ultrasonography [5, 10].

Treatment

The treatment of ACC is still controversial. It can be treated surgically or a conservative approach can be preffered or both the choices can be taken into account [**1, 20**] according to the location, size, and depth of the tissue defect [**5**]. Small lesions can heal spontenously and may present hipertrophic or atrophic scarring, skin contracture, or hypo-hyperpigmentation after recovery [**4, 10**]. Only local wound care can be performed with povidone iodine, bacitracin, or silver sulfodiazine in the conservative therapy of ACC, but serious complications must be considered [**22**]. Complications of conservative treatment include life-threatening hemorrhage, meningitis, secondary infections, and electrolyte disturbances [**23**]. Larger lesions can be treated by surgical methods, these methods include skin grafts, local flaps, free flaps, or tissue expansion [**3, 20**]. Some probable complications after surgical options such as sagittal sinus hemorrhage upon scar debridement, flap necrosis, and graft loss have been reported [**20**].

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