

A Case of Aplasia Cutis Congenita, Type V

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Aplasia cutis congenita is a congenital localized or widespread absence of the skin. We report a case of aplasia cutis congenita, type V, in a 6-day-old male infant born with the stellate and linear skin lesions covered by granulation tissue and soft capsule with slightly elevated erythematous edges on the trunk and lower extremities without any associated family history. The patient had amniotic bands and were diagnosed as aplasia cutis, type V. The patient received conservative treatment such as antiseptic dressing and prophylactic systemic antibiotics with healing of the ulcer. (Ann Dermatol 9:(1)73~76, 1997)

Key Words : Aplasia cutis congenita, Type V

Aplasia cutis congenita is a congenital skin disease characterized by localized or widespread absence of the skin, present at birth. Since it was first described by Cordon¹ in 1767, more than 500 cases have been reported. In Korea, Park et al² first reported a case of aplasia cutis congenita on the posterior fontanelle in 1984. In 1985, Choi et al³ reported 2 cases occurring on the posterior fontanelle and both feet with no associated anomalies or family history. Aplasia cutis congenita usually takes the form of a focal ulcer over the vertex of the skull but in 20-30% of cases, the defect involves the calvaria and the dura mater. Several clinical groups are characterized by the location and pattern of skin defects, associated malformations, and the mode of inheritance. Healing usually occurs with the formation of scars⁴.

We herein describe a 6-day-old infant born with amniotic bands who had aplasia cutis congenita on the trunk and both lower extremities with no associated family history.

REPORT OF A CASE

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In April, 1996, a 6-day-old male infant presented with skin defects and scar formation on the trunk and lower extremities. There was no significant antenatal or family history. The mother had no specific disease or drug history prior to or during the pregnancy. The male infant was born by normal vaginal delivery at 40 weeks' gestation. He weighed 3.55kg and there was no evidence of perinatal fetal distress. Amniotic bands were present. On physical examination, the infant looked rather well except the stellate and linear skin lesions on both sides of trunk and both lower extremities respectively (Fig. 1). The skin defects were hairless and covered by granulation tissue and soft capsule with slightly elevated erythematous edges. Laboratory findings revealed an increased white blood count of 17,000 but the erythrocyte sedimentation rate, urinalysis, and liver function tests were within normal ranges. Also, blood culture, TORCH test, CRP, and syphilis tests were all negative. No remarkable findings could be found on chest roentgenogram, infantogram, echocardiography, neurosonography, and abdominal sonography. Chromosomal studies were normal.

A skin biopsy taken from a healing lesion showed no specific change in the epidermis, and the dermis was generally eosinophilic and homogeneous with mild infiltration of chronic inflammatory cells and granulation tissue formation.

Fig. 1. Stellate and linear skin lesions on the right trunk and lower extremities.

Skin appendages were not observed (Fig. 2).

The patient was treated with systemic antibiotics and conservative management such as antiseptic dressing to prevent secondary infection. The wounds healed without requiring any surgical operations such as skin graft. At the time of discharge, there was no skin defect left.

DISCUSSION

Aplasia cutis congenita is a heterogeneous group of disorders with localized or widespread absence of the skin. More than 80% of the patients observed have defects of the scalp, usually located on the vertex at the midline. Less often, affected areas include the forearms, knees, trunk, legs, and face. The lesions not involving the scalp are often multiple and symmetrically distributed and have a tendency to more complete, rapid healing, leaving only minimal scarring. In contrast, the aplasias of the scalp usually result in scarring alopecia that may be extensive^{5,6}. Our case healed leaving an atrophic scar.

The diagnosis of aplasia cutis congenita is primarily clinical, and the histologic appearance varies. At birth, ulcerated lesions may show complete absence of all layers of skin, occasionally extending

Fig. 2. The dermis was eosinophilic and homogenized with slight infiltration of inflammatory cells. Skin appendages could not be observed. (H&E stain, $\times 100$).

to the bone or dura. Healed lesions often demonstrate flattened epidermis, proliferation of fibroblasts in a loose connective tissue stroma, newly formed capillaries, and complete absence of adnexal structures^{7,8}.

The cause of aplasia cutis congenita remains obscure and various etiologies have been suggested. The principal theories are chromosomal abnormalities (trisomy 13,15 and 4p-syndrome), amniotic bands, fetus papyraceus or placental infarcts, traumas (intrauterine pressure, birth trauma), teratogens such as antithyroid medication, intrauterine infection with *T. pallidum* or herpes simplex virus type II, thrombosed arteriovenous malformation or spontaneous healing of hemangioma, and defective closure of the neural tube^{4,9}. Stephan et al¹⁰ proposed an explanation for the origin of the scalp vertex aplasia cutis congenita. They hypothesized that the reason the scalp is a predilection site is that it is identified as the area of the greatest biochemical stretch due to rapid underlying brain growth, thereby causing tension-induced disruption within the skin. In addition, certain genetic conditions are thought to act as the predisposing factors to developmental abnormalities of the skin. In our case, we could not find any genetic predisposition and therefore the amniotic bands may be

the plausible cause.

Malformations associated with this disorder include syndactyly, polydactyly, cleft lip and palate, focal dermal hypoplasia syndrome¹², XY gonadal dysgenesis, amniotic band disruption complex, Adams-Oliver syndrome associated with limb defects and nail dystrophy^{13,14}, holoprosencephaly, myelomeningocele, plagiocephaly, leptomeningeal angiomatosis, laryngomalacia, tracheo-esophageal fistula, congenital heart disease, cutis marmorata telangiectasia, epidermolysis bullosa^{15,16}, Bart syndrome, and polycystic kidney.

In 1986, Frieden et al¹⁷ described several distinct clinical subtypes, characterized by the location and the pattern of skin absence, the presence of associated malformations, and the mode of inheritance. Group 1 is scalp aplasia cutis congenita occurring on the vertex without multiple anomalies associated with either autosomal dominant or sporadic inheritance. Group 2 is scalp aplasia cutis congenita involving the midline scalp associated with limb abnormalities and autosomal dominant inheritance. Group 3 involves the scalp asymmetrically and is associated with epidermal and organoid nevi and occurs sporadically. Group 4 is aplasia cutis congenita overlying embryologic malformations affecting the abdomen, lumbar skin, scalp, and practically any other site. The inheritance depends on the underlying condition. Group 5 is aplasia cutis congenita with fetus papyraceus or placental infarction involving multiple symmetric areas on the scalp, chest, flanks, and extremities. It occurs sporadically and is associated with single umbilical artery, developmental delay, nail dystrophy, clubbed hands and feet, and amniotic bands^{17,18}. Group 6 is aplasia cutis congenita associated with epidermolysis bullosa which is subdivided into two types. The first type, which shows localized blistering without multiple congenital anomalies, affects the extremities and the mode of inheritance is dependent on the epidermolysis bullosa type. The second type shows widespread skin fragility with congenital anomalies and is inherited in autosomal recessive manner. Group 7 is aplasia cutis congenita localized to extremities without blistering and has either autosomal dominant or recessive inheritance. Group 8 is aplasia cutis congenita caused by specific teratogens, and group 9 is aplasia cutis congenita associated with malformation syndromes such as trisomy 13. Our case demonstrated

multiple symmetric lesions on the trunk and extremities accompanied by amniotic bands. However, there was no genetic predisposition, and our case can be classified as group 5.

When aplasia cutis congenita occurs as a small, focal scalp ulcer, it heals spontaneously. When the lesion is large with absence of underlying tissues, surgery is performed to prevent meningitis or hemorrhage. Two methods of treatment have been proposed: a conservative approach consisting of daily antiseptic dressings to allow scalp epithelialization and bone ingrowth; a surgical approach by covering the scalp defect with a rotational skin flap at birth or by a full thickness skin homograft¹⁹. Our patient was treated conservatively with prophylactic systemic antibiotics and was discharged with complete healing of the ulcer.

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