CASE REPORT

An Unusual Association of Aplasia Cutis Congenita with Twin Pregnancy and Maternal Varicella Case Report

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ABSTRACT

We report an unusual case of aplasia cutis congenita associated with twin pregnancy and maternal varicella in first trimester, occurring over the extremity of one of the twin while other twin was perfectly normal.

Keywords: Aplasia cutis congenita, fetus papyraceous, maternal varicella

Aplasia cutis congenita (ACC) is part of a heterogeneous group of disorders first reported by Cordon in 1767 and is characterized by the absence of a portion of skin at birth.[1] Majority of the cases present as a solitary lesion on the scalp. It has also been reported with twin pregnancy where it occurs in surviving twin and other twin being fetus papyraceous.[1-3] However, ACC has not been reported in twin pregnancy with survival of both the twins.[4,5] Here, we report a rare case of ACC occurring over the extremity of one of the twin while other twin was clinically normal.

CASE REPORT

Index case was male baby, product of monozygotic twin (monoamniotic, monochorionic placenta, and identical) pregnancy, born to a primigravida mother delivered by normal delivery at 36 weeks of gestation weighing 2.62 kg at birth. Mother had suffered from chickenpox at 8 weeks of gestation lasting for 10 days. Antenatal period was normal otherwise and there was no history of teratogenic drug intake, consanguinity, or similar lesions in family.

On examination, baby had absence of skin (6 cm × 3 cm) over the right thigh extending from groin to knee (Fig. 1). The margins of the lesion were well defined with some fibrosis and epithelization and the base was clean, glistening red, with visible capillaries covered with a thin transparent membrane. There were no other abnormalities on physical examination. Second twin was clinically normal weighing 2.53 kg and was handed over to the mother soon after birth.

Lesion was managed with application of emollients and silver sulfadiazine ointment and it was left



Figure 1. Baby showing absence of skin (6 cm × 3 cm) over the right thigh extending from groin to knee.

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open without any dressing. It started healing with epithelization starting at the edges by Day 5 of birth. It healed completely within 2 weeks and at follow-up after 4 months, the overlying skin was normal without any contractures. The baby was found to be developmentally normal at 1 year of age when we followed him last.

DISCUSSION

ACC most commonly manifests as a solitary defect on the scalp vertex just lateral to the midline, but sometimes it may occur as multiple lesions and symmetrically. The exact incidence of the ACC is not known due to underreporting of the condition. The lesions are noninflammatory and well demarcated, and they range in size from 0.5 to 10 cm. They may be superficial involving only epidermis and the upper dermis, resulting in minimal alopecic scarring, or the defect may extend to the deep dermis, the subcutaneous tissue, or rarely the periosteum, the skull, and the dura. Prognosis is usually excellent; they heal with epithelization and leave an atrophic scar. Complications are usually due to the associated abnormalities.[1-5] Frieden has classified ACC into 9 groups based on the etiology and associated anomalies.

Group 1: ACC of scalp without multiple anomalies.

Group 2: ACC of scalp with limb anomalies.

Group 3: ACC of scalp with epidermal and sebaceous nevi.

Group 4: ACC overlying deeper embryonic malformations, e.g., spinal dysraphism, porencephaly, and others.

Group 5: ACC associated with twin pregnancy and fetus papyraceous.

Group 6: ACC associated with epidermolysis bullosa.

Group 7: ACC of the extremities without epidermolysis bullosa.

Group 8: ACC associated with teratogens and intrauterine infections, e.g., methimazole, herpes simplex virus, and varicella-zoster virus. The lesions in this group are usually at the scalp.

Group 9: ACC associated with malformation syndromes, e.g., trisomy 13, Johanson-Blizzard syndrome, and others.

ACC in Group 5 results in death of one of the twin during the first or second trimester. The surviving twin usually remains normal except ACC of scalp. Affected patients show linear areas of absence of skin that have bilateral pattern of distribution along the flanks and the lateral aspect of the limbs.[6-8] The cause of the symmetrical ACC is vascular disruption inducing abnormal dermoepidermal development and cutaneous defect through ischemic and thrombotic events. The intrauterine death of one of the fetuses should cause the release of the thrombosis promoting material from the dead fetus. These substances can cause placental infarction, disseminated intravascular coagulation, and cutaneous lesions.[6-8] Twin pregnancy in our case was not associated with fetus papyraceous; other twin was absolutely normal and there was no evidence of placental infarcts on gross examination. The defect was unilateral on right thigh rather than being bilateral or over the scalp.

ACC in Group 8 has been reported with primary varicella zoster infection in the mother during the first trimester of pregnancy and associated features are mental retardation, chorioretinitis, and limb hypoplasia. In our case, there was history of maternal varicella occurring at 8 weeks of gestation; but there were no other features suggesting varicella as underlying cause of ACC. Thus, index case is unique having association with maternal varicella infection as well as twin pregnancy but this association is probably coincidental rather than being causative.

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