Aplasia cutis congenita

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Dear Editor

The patient was a female neonate born prematurely at 36 weeks of gestation by a Cesarean section. During pregnancy, the mother had no history of fever, drug use, or X-ray exposure. The mother was gravida 2 and had no history of abortion. Her first child was healthy. There was no record of birth trauma. The perinatal history was negative for intrauterine trauma, the use of antithyroid medication, or misoprostol.

Sharply marginated lesions were present on the trunk, foot, and arm, as multiple ulcers, which measured up to several centimeters in diameter without bullae. Examination of the infant's trunk demonstrated a left-sided full-thickness skin lesion and a similar right-sided lesion (Figure 1). There was complete absence of the epidermis and dermis at these sites, but the lesions were covered by a transparent dry membrane. The rest of the examinations were unremarkable. The patient received intravenous antibiotics upon admission, and wet-to-dry normal saline dressing with mupirocin were used on the affected areas. Echocardiography and abdominal ultrasound were normal. A detailed genetic evaluation revealed no evidence of a hereditary form of the disorder. The lesions were managed non-operatively with dressing twice daily, irrigation with normal saline, and mupirocin. After 3 weeks, the wounds were noted to be contracted, and granulation tissue covered the defects. There was no functional impairment, growth alteration, or limitation of movement.

Aplasia cutis congenita is a rare group of disorders with various etiologies characterized by the focal absence of skin at birth [1]. Several theories have been proposed to explain the pathogenesis of this disorder include incomplete closure of the neural tube, localized vascular insufficiency, amniotic membrane adhesions, teratogenic agents, and intrauterine infections, but a single unifying mechanism seems unlikely [2-4]. The diagnosis is usually based on clinical findings. Increased levels of acetylcholinesterase and α-fetoprotein have been

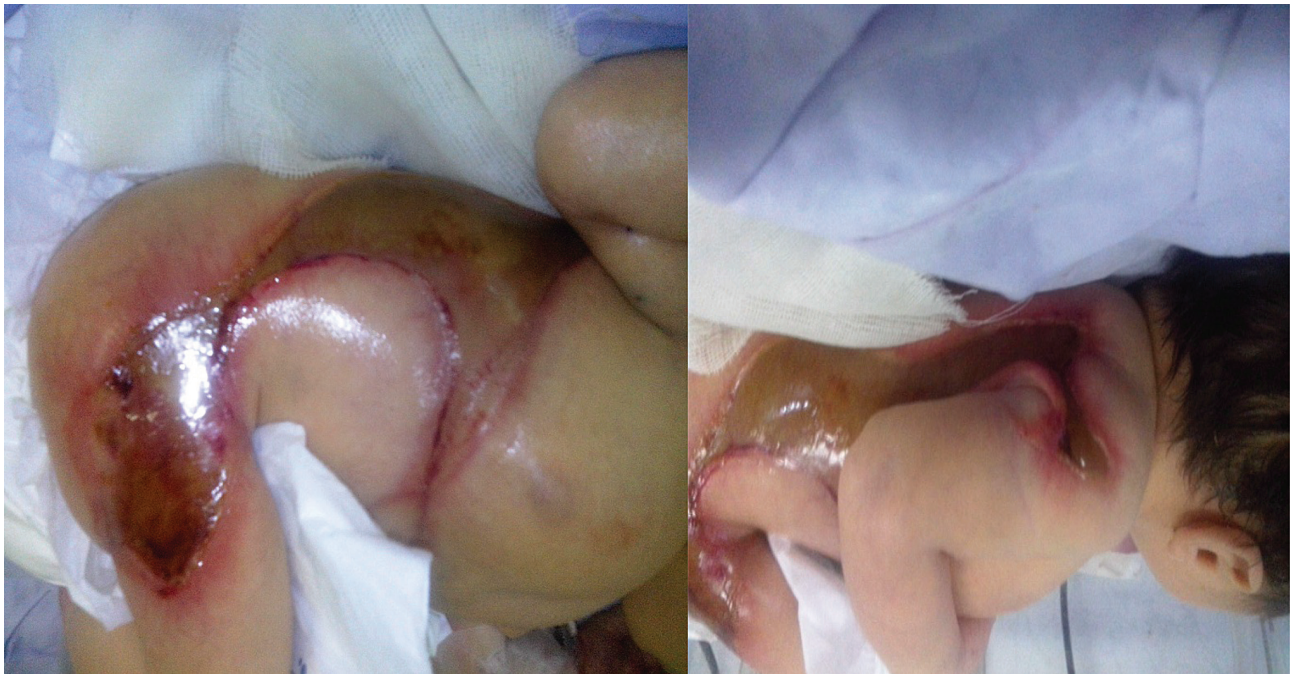

Figure 1. Sharply marginated lesions on the trunk, foot, and arm

reported in the amniotic fluid of mothers whose children have aplasia cutis [5]. Management strategies include simple observation, prevention of infection in the case of ulceration, and surgical excision or skin grafting in the case of large defects [4].

Most cases of aplasia cutis congenita occur sporadically but autosomal dominant and autosomal recessive modes of transmission have also been well documented. Aplasia cutis may overlie embryologic malformations such as meningomyelocele and spinal dysraphia, omphalocele, and gastroschisis. Scalp defects are associated with specific teratogens (methimazole, intrauterine varicella and herpes simplex) and malformation syndromes (trisomy 13, Johanson-Blizzard syndrome, amniotic band disruption complex, and the ectodermal dysplasias) [4].

Lesions are sharply marginated and may manifest as ulcers, bullae, or scars. Lesions may be solitary or multiple, measuring up to several centimeters in diameter. The congenital absence of skin is a cutaneous anomaly most often affecting the scalp, but occasionally involving the trunk and extremities, as well [6-7]. Multiple defects, particularly those on the trunk and extremities, may be strikingly symmetric in distribution [3]. Larger defects are often deeper and can extend to the dura or meninges. Histologic examination of the tissue from the defect demonstrates the absence of epidermis and a diminished number of appendageal structures and dermal elastic fibers or, in deeper lesions, the absence of all layers of the integument. There is no evidence of inflammation or pathogenic organisms [4].

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