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TOPIC HIGHLIGHT

Adams-Oliver Syndrome

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ABSTRACT

A term infant born in a resource-poor setting (Mozambique) presented with extensive disepithelization of the cranial vertex with absent underlying bony plate, disepithelization of the posterior median line, and bilateral amputation of the fifth toe. These iconic clinical features are indicative of extensive Aplasia Cutis with associated transverse limb defects, a condition known as Adams-Oliver Syndrome. Management is typically conservative; increased need for fluids, risk of electrolyte derangements and infections are possibly associated issues in the first days of life, especially in case of large lesions, and have to be carefully monitored and adequately addressed.

Key words: Adams-Oliver syndrome; Neonatology; Dermatology

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Abbreviations
ACC: aplasia cutis congenita; FP: fetus papyraceus; AOS: Adams–Oliver syndrome

CASE REPORT

We evaluated a term male newborn in a resource-poor setting (Beira, Mozambique). Pregnancy was unfollowed and no information about the gestational period was available. Maternal HIV test was negative; there was no history of drug consumption during pregnancy. Physical examination at birth revealed extensive disepithelization of the cranial vertex (Figure 1, upper left) with concomitant absence of the underlying bony plate; another extensive linear area of disepithelization was present posteriorly along the median line (figure 1, upper right). The fifth toe of both feet was amputated (Figure 1, bottom left). In order to prevent infection of the exposed areas, the patient received intravenous ampicillin and gentamycin for fifteen days, plus topical antibiotic therapy with sulfadiazine till complete re-epithelization, which occurred in one month (Figure 1, bottom right). Cerebral, spinal, cardiac and abdominal ultrasound turned out normal; histopathologic examination of the placenta was not possible due to the clinical context.

DISCUSSION

Aplasia cutis congenita (ACC) is a rare condition consisting in the

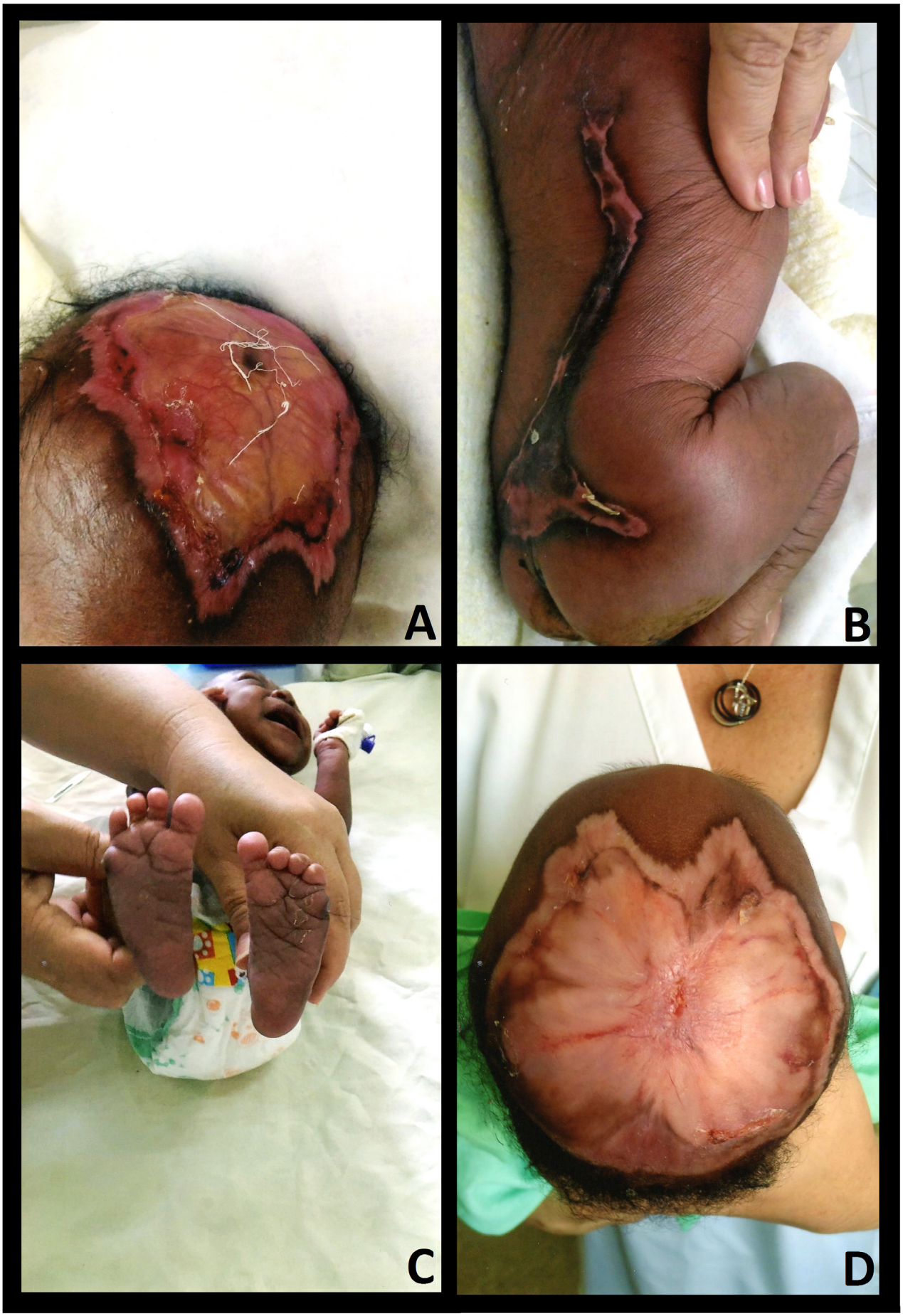

Figure 1 Upper left: extensive cranial disepithelization with absent bony plate, as seen at birth; Upper right: extensive disepithelization of the posterior median line, as seen at birth; Bottom left: bilateral fifth toe amputation, as seen at birth; Bottom right: complete cranial re-epithelization, one month after birth.

absence of all skin layers at birth. Scalp is commonly interested; trunk and limbs are also not unfrequently affected[1]. Truncal involvement in the newborn is often associated with *in utero* death of a co-twin progressively pressed flat by the growth of the remaining living fetus, and resulting at birth as a fetus papyraceus (FP). The absence of the FP at the delivery is called "vanishing twin", a condition with an incidence of 1:12,000 newborns[2]. ACC with associated terminal transverse limb defects is referred to as Adams-Oliver syndrome (AOS). In such condition, limb defects more severely affect lower than upper extremities, and can range from mild (unilateral or bilateral short distal phalanges) to severe (complete absence of whole toes –as in our patient-, fingers, feet or hands, or more proximal limb defects, often resembling an amputation). The diagnosis of this condition is basically clinical; nevertheless, congenital heart disease, brain, and abdominal abnormalities must be excluded[3,4]. Typically, conservative management is sufficient; increased need for fluids, risk of electrolyte derangements and infections are possibly associated issues in the first days of life, especially in case of large lesions, and have to be carefully monitored and adequately addressed[1,2,5].

CONCLUSIONS AND FINAL REMARKS

Differential diagnosis between ACC and AOS is not always simple because of remarkable clinical overlap between the two entities; moreover, a definitive diagnosis in the first days is not necessary since management of the two conditions is largely the same. Starting early conservative treatment (basically consisting of local wound care) is pivotal in the suspect of neonatal ACC/AOS; adequate attention also must be put in the prevention and treatment of possibly associated complications (such as infections, excessive loss of fluids, and/or electrolyte imbalance). Cerebral, abdominal, and cardiac anomalies must be excluded. Clinical evolution is usually favorable: in most cases of absent skull bony plate, spontaneous secondary closure of the cranial vault occurs within one year.

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