CLINICAL CASE

Pseudosyndactyly and amputation as the main features of the amniotic band syndrome

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ABSTRACT

Background. Amniotic band syndrome causes a wide spectrum of congenital defects such as cleft lip, hydrocephalus, and growth restriction of limbs, with or without amputation. The condition occurs in 1:1,200 to 1:15,000 deliveries. The objective of this report is to define the most important features of amniotic band syndrome.
Case Report. We report the case of a 21-year-old female with a first pregnancy. No prenatal care or ultrasound studies were carried out, but the patient reported experiencing vaginal discharge during the first trimester. The pregnancy resulted in cesarean section at 30 gestational weeks because of severe oligohydramnios. A 1200-g female was delivered with Apgar 7/8. The newborn presented congenital amputation of the right leg and pseudosyndactyly of the left hand with a ring constriction of the third phalange of the second, third and fourth finger, a left equinovarus foot and amputation of the right leg from the middle third of the tibial region. These findings were confirmed by x-ray.
Conclusions. Constriction rings, limb amputation and pseudosyndactyly are important and consistent features with the amniotic band syndrome. Functional prognosis is nonfavorable, but prognosis for life productivity is good.
Key words: amniotic band syndrome, limb amputation, pseudosyndactyly.

INTRODUCTION

Amniotic bands are fibrous strands that extend from the outer surface of the cord into the amniotic cavity. Amniotic band sequence is a spectrum of fetal malformations associated with fibrous bands that entrap fetal body parts. The frequency of occurrence is 1:1,200 to 1:15,000 births.[1] These malformations were described by Streeter, who proposed the presence of amniotic bands as a result of the theory of germplasm. However, there are other theories to explain the presence of amniotic bands. The most accepted theory is the one extrinsically developed by Torpin who postulated rupture or infection as an initial event, with a decidual reaction, formation of fibrous cords with progressive separation of the cord and recollection of the amniotic sac producing transient oligohydramnios.[2] These mesodermal flanges trap and strangle limbs, fingers or other fetal organs. The results of the deformation, amputation or disruption depend on the gestational age at which the rupture occurred.[3] It has been reported that there is an increased risk in young mothers with low educational levels, as well as in unplanned pregnancies and non-white and non-Hispanic races.[4] The malformations are generally involved with only the external organs,[5] although in some cases there are major malformations present that are incompatible with life.[6] Prognosis depends on severity and the organs involved. Varying degrees have been described depending on the level of injury: grade 1 involves the subcutaneous cellular tissue, grade 2 extends to the fascia, grade 3 extends to the fascia and requires release and grade 4 is described when amputation has occurred.[7] This report describes an infant with grade 4 amniotic band sequence.

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Case Report

We present the case of a newborn female whose 21-year-old mother was reported to be in good health. This was her first pregnancy and she experienced untreated vaginitis during the first trimester. She had irregular prenatal care

and there were no obstetric ultrasounds performed. Due to premature rupture of the membranes 5 days prior to delivery, the mother received a course of antibiotics. The infant was delivered via caesarean section at 30 weeks of gestation due to presentation of severe oligohydramnios. Birth weight of the infant was 1200 g, length was 37 cm, head circumference was 27.5 cm and Apgar score was 7/8 according to conventional time intervals. A physical examination noted hypotrophy with flat nasal bridge, widespread cutis laxa, clinodactyly of both fifth fingers, pseudosyndactyly with a constriction ring joining the apex of the phalanges including the distal phalanges of the second, third and fourth fingers of the right hand (Figure 1) and hypertelorism of the breasts. We found good pulmonary ventilation and the cardiac area was without murmurs or arrhythmias. Abdomen was normal. There were external female genitalia, left clubfoot and the right leg was amputated from the tibial region, which ends in a point with distal-type eschar lesion. Radiographically, the right leg of the newborn demonstrated the tibia with an end in a distal cone in the middle third and hypoplastic fibula (Figure 2). No cytogenetic study was performed on the patient or her parents and the patient was discharged in good condition.

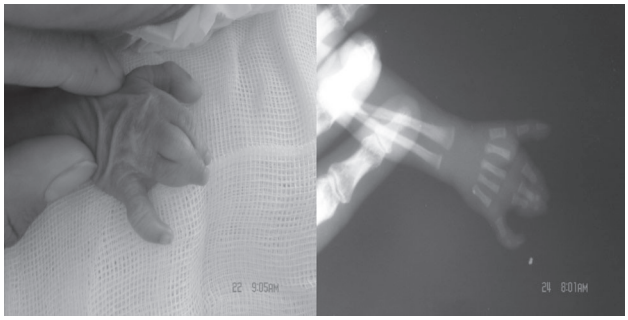

Figure 1. X-ray showing pseudosyndactyly with a constriction ring joining the apex of the phalanges including the distal phalanges of the second, third and fourth fingers of the right hand and clinodactyly of both fifth fingers.

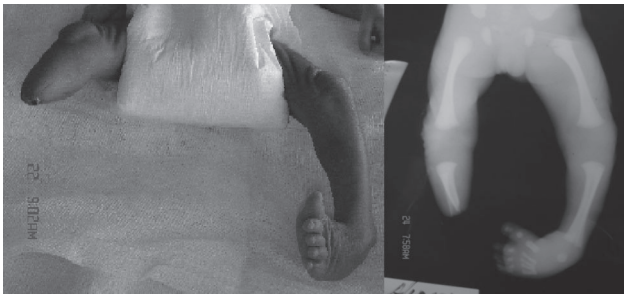

Figure 2. Amputation of the right lower limb (distal to the knee) and left equinovarus foot.

DISCUSSION

Amniotic band syndrome is not common and presents with a wide clinical spectrum of deformities, sometimes complex and lethal. The most accepted theory is that of premature rupture of membranes with amniotic band formation and disruption of fetal parts,[8] which can best be explained as a deformity in embryological growth due to compression or ischemia from an amniotic band.[9] These flanges trap and strangle any fetal part or organ with a consequent alteration of the fetal structure, whether disruption, deformation or amputation. A decidual reaction occurs and fibrous cords (amniotic bands) form, extending from the outer surface of the amniotic cavity and trapping fetal parts. As a result, a wide range of birth defects are noted with constriction rings that affect from one to all four extremities, most often the lower ones. The intrauterine constrictions are asymmetric with distal atrophy or amputations, pseudosyndactyly, lymphedema and clubfoot, in addition to severe craniofacial, visceral and trunk deformations.[10-12] In many cases these are observed in young mothers, sex-trade workers or in mothers who presented with urinary tract or cervical infections and fever syndrome during the first trimester of pregnancy.

In this case, the young mother had an untreated vaginal infection during the first trimester. Differential diagnosis must be made with other causes of amputation or congenital limb absence. There are many genetic syndromes that cause reduction of the size of the extremities:

- Holt-Oram syndrome—agenesis or hypoplasia of radius and thumb.
- VATER/VACTERL association (**V**ertebral defects, **A**nal defects, **C**ardiac, **T**racheoesophageal fistula, **E**sophageal atresia, **R**enal anomalies, **L**imb defects (extremities), where other disturbances would present that are absent in our patient.
- Unilateral hypomelia syndrome or caudal regression syndrome—often appears in children of diabetic mothers in which there is a decrease in femoral length but not a distal amputation.

This case includes alterations of two extremities: 1) the upper extremity with a grade 1 injury affecting the subcutaneous tissue with radiological bony structures of the unfused phalanges and 2) the lower limb with a grade 4 injury, which includes amputation. This has an asymmetric or multistage characteristic with no genetic or chromosomal etiology.

On the other hand, the feature of the amniotic flanges, in addition to being associated with a distal amputation at the site of the constriction zone caused by the strangulation of the flange, is the type that regularly leaves the ring pattern (constriction ring) and is related to pseudosyndactyly or distal amputation to the site of constriction, both present in this case. In our hospital, similar cases have been reported that described one with involvement of limbs, constriction rings and pseudosyndactyly with features that support the diagnosis of amniotic flanges.

The formation of amniotic bands commonly affects the extremities in the distal segments including the hand. Given the heterogeneity in the expression of this disease, treatment must be individualized, considering the functionality based on surgical programming.[13] The ultimate outcome depends on the severity of the malformation and the affected organ.[14] In this case, prognosis for the function of this patient is poor with consequences for walking, but we consider that corrective surgery and the use of a prosthesis will aid the patient in adapting to a productive life. Currently we have proposed fetal endoscopic surgical release of the amniotic bands,[6,15,16] although results have been controversial.[14] In developed countries and where legislation permits, pregnancy termination is usually recommended in fetuses with visceral or major craniofacial defects.[14] This type of patient can be offered a treatment with corrective surgery and a prosthetic in order to have a productive life.

Finally, we conclude that some important signs such as constriction rings, total or partial amputation of a limb and pseudosyndactyly present in this case support the diagnosis of amniotic band syndrome. These major defects establish a poor prognosis for function of the affected limb, although prognosis is favorable for the infant's survival.

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