

## Amniotic Band Sequence, Frequently Misdiagnosed in Cleft Patients

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**Background:** Amniotic band sequence (ABS) is a set of congenital malformations attributed to amniotic bands that entangle fetal parts during intrauterine life. Etiopathogenesis of this sequence is still unknown. Because of such a wide spectrum of possible anomalies and many combinations of their simultaneous appearance, there are no two identical cases of ABS. This diagnosis is often missed based on the secondary major defects.

**Patients and Methods :** There are cases of cleft patients treated in Plastic Surgery Division Cipto Mangunkusumo Hospital that has other congenital anomalies. The data is taken from the medical record and surgical photos (in our division from 2010-2011).

**Results :** The patients presented had either facial cleft or cleft lip and palate. These patients had other congenital anomalies such as meningoencephalocele and constriction ring syndrome or other defects on the extremities.

**Summary :** This diagnosis may not be often established because we often only see the secondary major defects such as meningoencephalocele or facial cleft. With this paper we hope that we could make this diagnosis known so we can make a multidisciplinary approach to this sequence.

**Keywords :** Amniotic Band Sequence, craniofacial cleft, ADAM sequence, limb defects

**Latar Belakang:** Terdapat beberapa kasus sumbing yang ditangani oleh divisi bedah plastik rumah sakit cipto mangunkusumo dengan kelainan kongenital lain. Data diambil dari rekam medis dan data operasi tahun 2010-2011.

**Pasien dan Metode:** Terdapat beberapa kasus sumbing yang ditangani oleh divisi bedah plastik rumah sakit cipto mangunkusumo dengan kelainan kongenital lain. Data diambil dari rekam medis dan data operasi tahun 2010-2011.

**Hasil:** Pasien yang ditemukan memiliki facial cleft atau sumbing bibir. Pasien ini juga memiliki kelainan kongenital lain seperti meningoensefalokel dan constriction ring syndrome atau defek lain pada ekstremitas.

**Ringkasan:** Diagnosis ini sering terlewat karena kita terfokus pada kelainan sekunder yang lebih berat. Dengan makalah ini diharapkan diagnosis ini tidak terlewatkan sehingga dapat dilakukan pendekatan multidisiplin.

The term amniotic band sequence refers to a set of congenital malformations attributed to amniotic bands that entangle fetal parts during intrauterine life.<sup>1</sup> It is also known as Amniotic Deformity, Adhesion, and Mutilation (ADAM) complex or Congenital Constriction Band Syndrome (CCBS).<sup>1,2</sup> The condition occurs in 1:1,200 to 1:15,000 deliveries.<sup>3</sup>

It has been postulated that several factors may have substantial influence on the ABS prevalence rate. The most frequent proposed factors are ethnicity, maternal age,

order of pregnancy, birth sex, and febrile maternal illness/drug use in the first trimester.<sup>1</sup>

The pathogenesis of ABS remains unclear. It has been widely accepted that fibrous bands happening in the first trimester of gestation as a result of amniotic rupture are the cause of infant's congenital defects.<sup>4</sup> The developing embryo is protected within two cavities. Like a balloon within a balloon, the amniotic cavity sits within the larger chorionic cavity. As the amnion expands, it eventually presses against the chorion, obliterating the early extra embryonic coelom. Usually this fusion of the two membranes is complete by

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The timing of amniotic rupture determines the nature and extent of the structural defects. Amnion rupture at the 3rd week of gestation produces anencephaly, encephalocele, meningocele, facial distortion, proboscis, eye defects, unusual facial clefting, sternal clefting, ectopia cordis or placental attachment to head and/or abdomen.<sup>6</sup> Amnion rupture in the 5th week of gestation produces unusual cleft lip, choanal atresia, limb defects, polydactyly, syndactyly, abdominal or thoracic wall defects, and scoliosis.<sup>6</sup> Amnion rupture from the 7th week onward produces craniostenosis, apparent omphalocele, amniotic bands, amputations, hypoplasia, pseudosyndactyly, ring constrictions with

distal lymphedema, foot deformities, dislocation of hip, and short umbilical cord.<sup>6</sup>

Because of such a wide spectrum of possible anomalies and many combinations of their simultaneous appearance, there are no two identical cases of ABS. This diagnose is often missed based on the secondary major defects such as meningoencephalocele, unusual facial cleft and limb deformities.

## PATIENT AND METHODS

### CASE 1

A 2,5 year old boy came to Cipto Mangunkusumo hospital with unusual facial cleft (Figure 1, upper row). We performed an operation to close the facial defect and did a lip adhesion. Six months after the first operation the patient underwent a palatoplasty to close the palatal defect. He also had multiple constriction rings on both hands and also syndactyly of the lower extremities (Figure 1, lower row).

### CASE 2

A 1,5 year old baby girl presented with occipital meningoencephalocele and constriction ring on the right fingers, and complete bilateral cleft lip palate. This patient already had a labioplasty and came to Cipto Mangunkusumo hospital to close the palatal defect. The patient has multiple deformities consisted of: cleft lip and palate, limb deformities and cranial defect (Figure 2).

The limb deformities were constriction rings on the right fingers and the cranial defect was a meningoencephalocele. We can see a faint scar post-bilateral labioplasty and the patient was never diagnosed for a specific syndrome involving this range of deformities.

## RESULTS

Deformities apparently caused by constricting bands have been described since at least the late 17th century. Now there are many anomalies that have been reported in association with congenital constriction bands, characterized by their extreme variability. These include limb reductions, congenital pseudarthrosis, clubbed feet, massive body wall



**Figure 1.** (Left to right, first row) The facial cleft defect before closure, intraoperative, and post-operative 6 months afterward. (Lower row) Upper and lower limb deformities found in this patient.



**Figure 2.** (Left) The pre-operative picture of the complete bilateral cleft lip is shown on the upper row, and the post-operative picture (middle) 8 months afterwards. The occipital meningo-encephalocele (right, upper row) and right upper limb deformities (lower row).

deficiencies, internal organ anomalies, and craniofacial deformities. Amniotic bands have been proposed as the cause of a variety of congenital anomalies that may be divided into three groups, depending on whether they affect the limbs, head, or body wall.<sup>5,7</sup>

Limb defects may include simple amputations of fingers or toes, partial syndactyly, constriction rings that encircle a limb, or even amputations that affect a significant portion of the involved arm or leg.<sup>5,7</sup> Oblique facial clefts, encephaloceles, and neural tube-like defects are the abnormalities

associated with amniotic bands affecting the head. The body wall defects may involve the chest (thoracoschizis, extra thoracic heart) or the abdomen (gastroschizis).<sup>5,7</sup>

As we can see in both cases presented in this paper, the patients have craniofacial defect and limb deformities. Neither had a body wall defect. But both have different presentation, in case 1 the patient has facial cleft and craniosynostosis whereas the patient in case 2 had a complete bilateral cleft lip and palate. The limb deformities are also different; in case 1 the patient had a multiple limb deformities including constriction ring and syndactily, while in patient 2 we can only see constriction ring on one of the upper limb.

There are two theories involving the pathogenesis of ABS. The widely accepted "extrinsic model", proposed by Torpin and Faulkner in 1966, is that the amniotic bands themselves cause the disorder (exogenous theory).<sup>8</sup> This theory relates to a premature rupture of the amnion, which leads to the passage of the fetus into the chorionic cavity. This cavity, which is poor in amniotic fluid, supports the fetus during the remainder of the gestation in close apposition to the chorion, resulting in compression and postural deformities. As a result of the rupture, a mesodermic amniotic band forms and attaches to fetal parts, which are abraded because of the close contact with the chorion; the abrasion causes amputation, constriction deformities, and facial cleft as the fetus continues to grow. The prominence of the nasal processes combined with the adjacent stomodeal orifice can lead to a free band attachment and adherence, which result in a spectrum of facial cleft defects that are similarly oriented. The intrinsic model was proposed by Streeter in 1930 and suggests that the anomalies and the fibrous bands have a common origin, caused by a perturbation of developing germinal disc of the early embryo.

Despite of the two different theories; the risk factors for ABS are ethnicity, maternal age, order of pregnancy, birth sex, and febrile maternal illness/drug use in the first trimester. Some studies found connection between ABS and mother's age (especially primiparas under

the age of 25, prematurity, abdominal trauma, unsuccessful abortion, intrauterine contraception, cerclage, chorionic villus sampling, amniocentesis, malformations of the uterus, some drugs taking like ergotamine, acetaminophen, and misoprostol<sup>8</sup>. However, no firm evidence of definite causality for any of these factors. Several studies confirm this opinion, with evidence that most of the cases of ABS have no risk factors in prenatal anamnesis. In both cases the maternal age are quite young 18-22 years old and in the second case the patient's mother only complained of falling in early pregnancy. While for case 1 the pregnancy period was uneventful.

From the wide range of deformities that both cases we included in this paper, we can conclude that this sequence has many possible anomalies and many combinations of their simultaneous appearance, which then makes us often miss this diagnosis. Over 50% of amniotic band sequence cases reported worldwide were initially misdiagnosed based on the secondary major defects. In both cases we find craniofacial defect and limb deformities. And in both cases we overlook the greater picture of a syndrome or sequence. There is a study describing the amniotic band sequence that the incidence of craniofacial anomalies is between 8.4 and 63 percent of cases.<sup>9</sup> These defects range from common cleft lip and palate to encephaloceles and anencephaly. Rare clefts are not mentioned specifically. Hall found 10 of his 39 cases (26 percent) to have both limb and craniofacial anomalies.<sup>9</sup>

## SUMMARY

ABS is not very often, but should be considered in every newborn with congenital anomalies; especially defect of extremities, head and/or body walls.<sup>8</sup> Postnatal diagnosis is made by physical examination of the newborn, with additional examinations to detect potential internal organs malformations. Because of ABS complexity, the treatment and follow-up of these children requires a team of specialist (multidisciplinary approach: plastic surgeon, orthopedic surgeon, orthodontist, ophthalmologist, neurosurgeon), according to special needs of every single patient.<sup>8</sup>

Finally, we can conclude that some important signs such as limb deformities (constriction rings, total or partial amputation of a limb and pseudosyndactyly) with craniofacial cleft presented in these two cases support the diagnosis of amniotic band sequence. These major defects establish a poor prognosis for function of the affected limb, although prognosis is favorable for the both infants' survival.

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