Amniotic band disease: About a case

ABSTRACT

Amniotic band disease (ABD) is a complex and relatively rare set of birth defects. These malformations mainly concern the limbs, the craniofacial region and the thoracoabdominal axis. The severity of these malformations is highly variable, starting from isolated skin furrows to visceral malformations that are often incompatible with life. The prognosis of ABD in the presence of craniofacial and visceral polymalformations is known and bleak. The situation is quite different in the case of superficial and isolated constriction of a limb. A medical termination of pregnancy is generally advised in the presence of severe craniofacial and visceral malformations, while isolated malformations of the limbs are accessible to surgical treatment at birth. We reported 1 case of ABD.

Key words: Amniotic band disease, polymalformations, surgical treatment.

INTRODUCTION

Amniotic band disease (ABD) is a complex and relatively rare congenital malformation [1]. These malformations mainly concern the limbs, the craniofacial region and the thoracoabdominal axis. Two main pathophysiological theories are opposed: the early rupture of the amnion (exogenous theory) would lead to the formation of fibrous bands which would themselves be responsible by strangulation for the malformations observed; the endogenous theory reports a vascular origin, the flanges then having no causal role [2, 3]. We study 1 case of ABD, whose diagnosis was made antenatally in order to recall the diversity of clinical forms and present the diagnostic and therapeutic difficulties.

OBSERVATION

A patient, aged 25, unigest and nulliparous, was referred to our department for an ABD, screened during the ultrasound scan in the 2nd trimester of pregnancy. She was group A Rh+, positive Rubella and Cytomegalovirus serology and negative Toxoplasmosis serology. In her medical and surgical history, she reported having had a gastric band in January 2014 in Russia. Her pregnancy was monitored in Russia until 15 weeks of amenorrhea (SA). The 1st trimester ultrasound was normal, fine nuchal translucency, low-risk triple test. Ultrasound performed at 20 WA showed: edema of the left hand up to half of the left forearm with a clearly visualized point of stricture (Figure 1). Right hand: 4th and 5th fingers missing (Figure 2). Medical termination of pregnancy has already been mentioned by the patient. At 20 WA 5/7, a fetoscopy was performed showing resection of the band located on the left hand as well as part of the band around the left forearm. The patient was hospitalized for 48 hours for observation, rest and cure of Dolcidium.

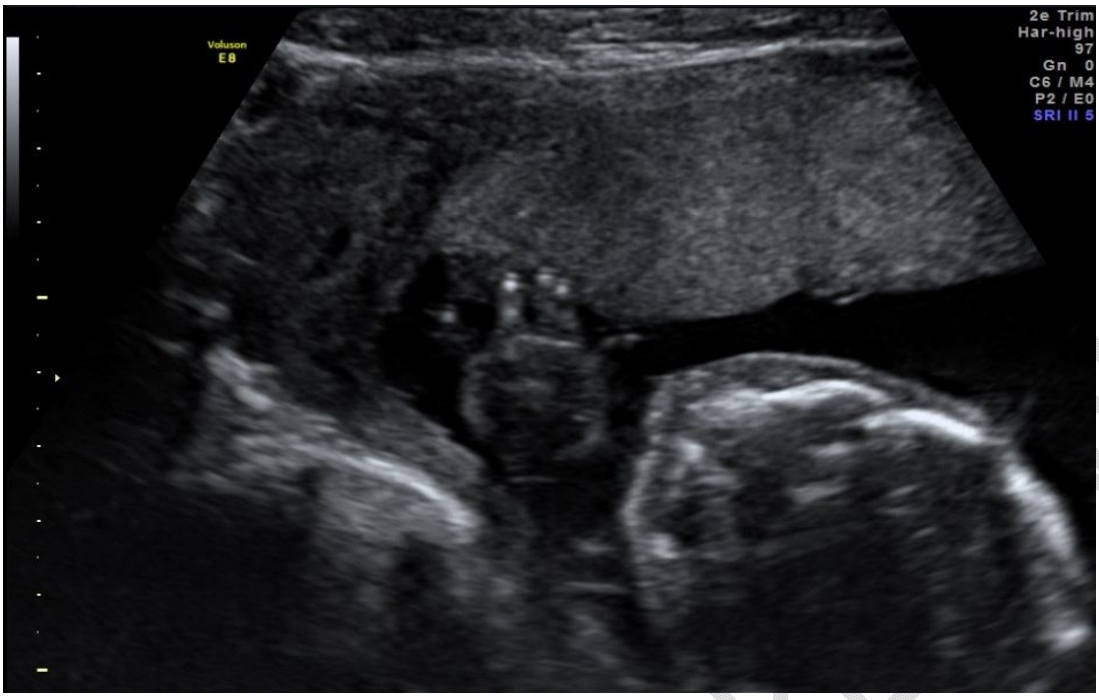
At 28 weeks 2/7: persistence of the edema in the left forearm but with good arterial vascularization in the forearm as well as in the left hand. The 5 rays of the carpus and the metacarpus seem present as well as all the phalanges. The right hand is inexaminable during this test. On the other hand there seems to be a flange at the level of the face which was not visualized before. Due the impossibility of studying the 2 limbs in detail as well as the suspicion of a bridle in front of the face, a spiral scanner was carried out in order to study the skeleton and it confirmed the absence of the 4th and 5th rays on the right, very disorganized with possible fusion (Figure 3).
At 30 weeks, a 3D ultrasound was performed at Brugmann, where the face seems quite free but again not 100% certain.
At 32 weeks: an amnio-infusion is performed, the face is well visualized and seems normal; at the level of the hands, the 10 fingers seem present, the thumbs seem present, the middle fingers seem shorter

At 33 WA 4/7, she had uterine contractions with loss of brown blood.

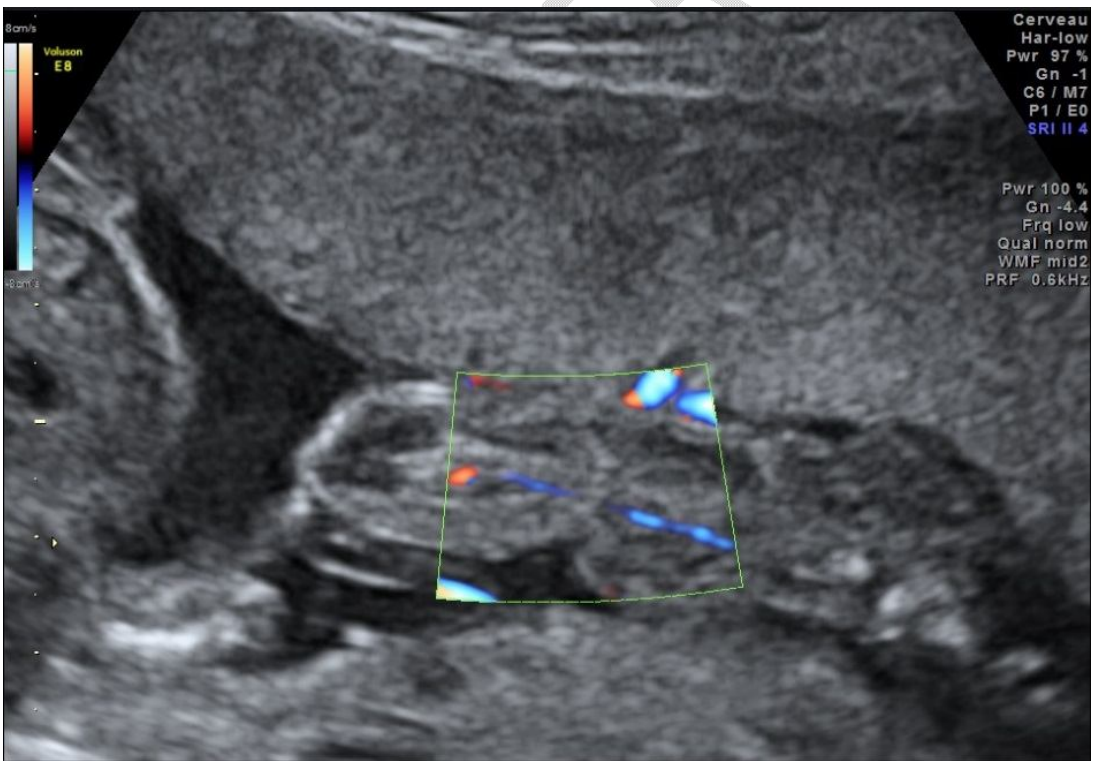
Assessment at labor room admission:
Clinical examination: abdomen tense during contractions then relaxation. Absence of uterine tenderness. On vaginal examination: cervix 2.5cm high and dilated to 1.5cm. Bloody mucous discharge.
Ultrasound of the cervix: 25 mm long. Negative urine test strip.
Biology: CRP at 25mg/dl; Hb:11.7; White blood cells: 18000; Kleihauer 3ml; urine sediment and urine culture: negative;
Pap smears: rare yeasts

Fetal assessment:
CTG: RCF well reactive, irregular uterine contractions; Echo: single fetus in cephalic presentation estimated at 2000 g; absence of visualized placental abruption. DAO IR = 0.55; DACM and Vmax <1.5 Mb normal.

Patient hospitalized and put to rest. 48h cure of Atociban as well as a pulmonary maturation by Betamethasone.
At 34 WA, spontaneous labor and vaginal birth of a boy weighing 2130 g with suspected lesions in the right hand (fusion of 4th and 5th fingers) and ring flanges around the forearm and 2 fingers of the left hand (Figure 4 and 5).
RX Left hand: short appearance of the 3rd and 4th metacarpals, dysmorphic appearance of the phalanges of the 2nd and 3rd ray.
No ossification nucleus of the 3rd phalanx of the 4th ray (Figure 6).
RX Right Hand: short aspect of the 1st phalanges of the 4th and 5th rays of which there is only the base and the proximal metaphysis. Absence of middle and distal phalanx at the 4th and 5th ray level, only a small ossification nucleus is visualized at the level of the fleshy bud (Figure 7).
Macroscopic anapathological examination of the placenta has confirmed the presence of amniotic bands (Figure 8). The newborn has benefited from a preoperative check-up and surgery with cutaneous plasty for flanges and the postoperative course was simple.



1a



1b

Figure 1. Left hand edema with well visualized stricture (1a and 1b)

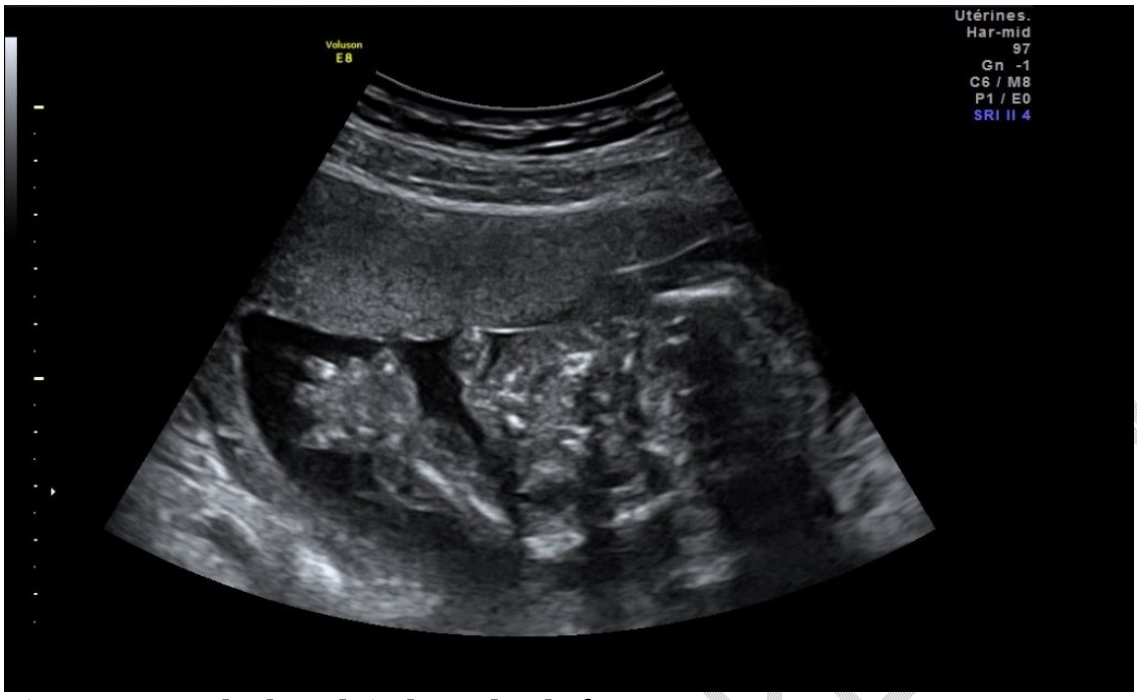


Figure 2. Right hand (4th and 5th fingers missing)

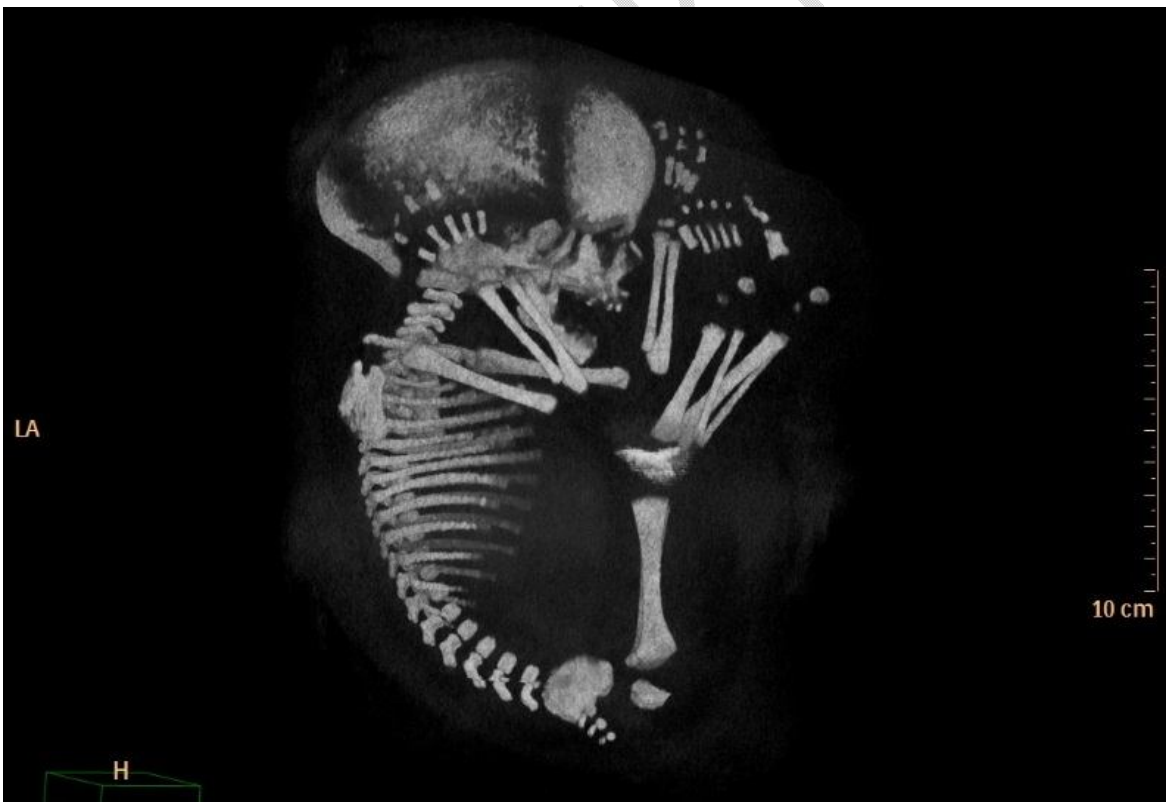


Figure 3. Absence of the 4th and 5th rays on the right, very disorganized with possible fusion

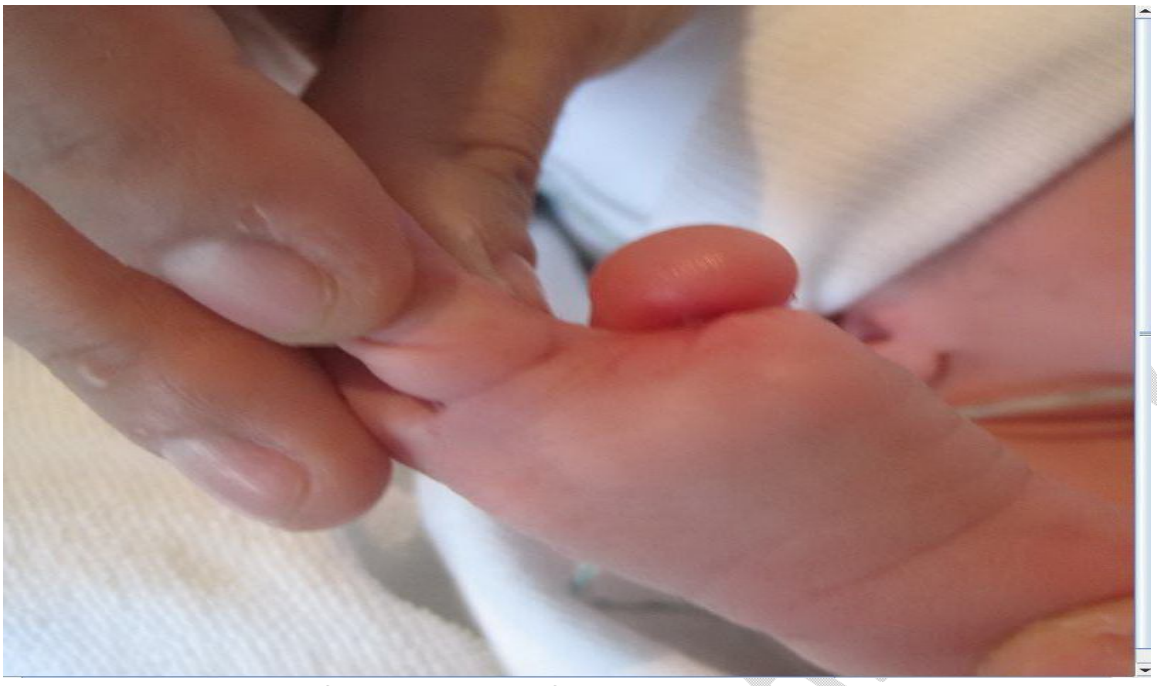
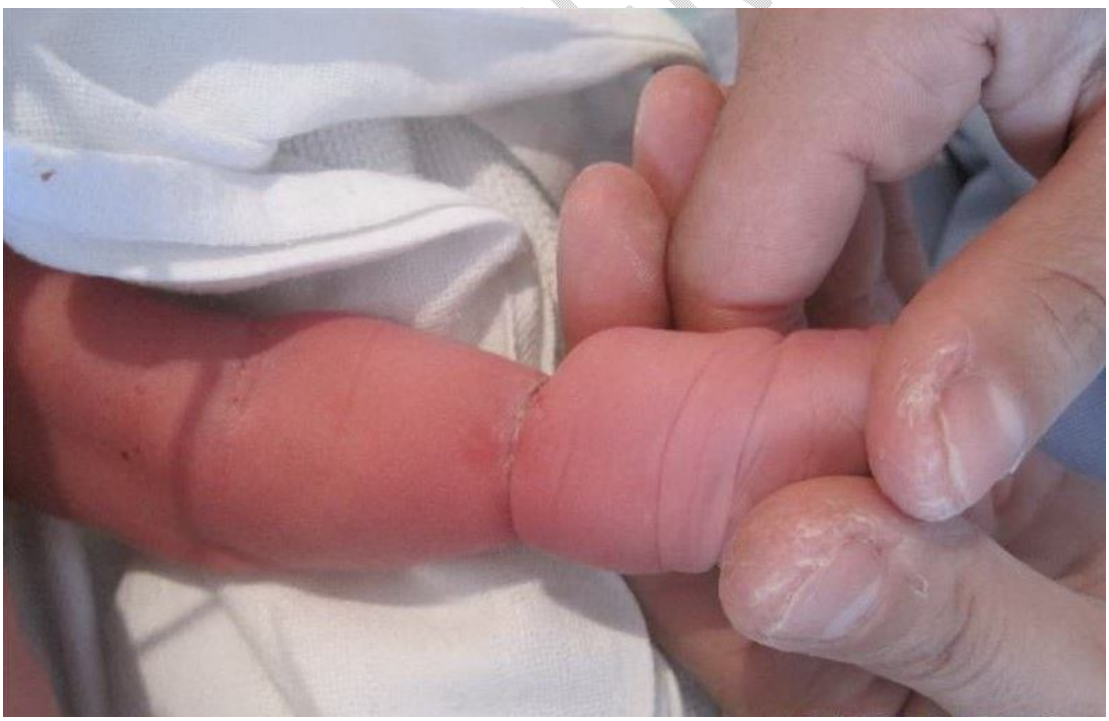
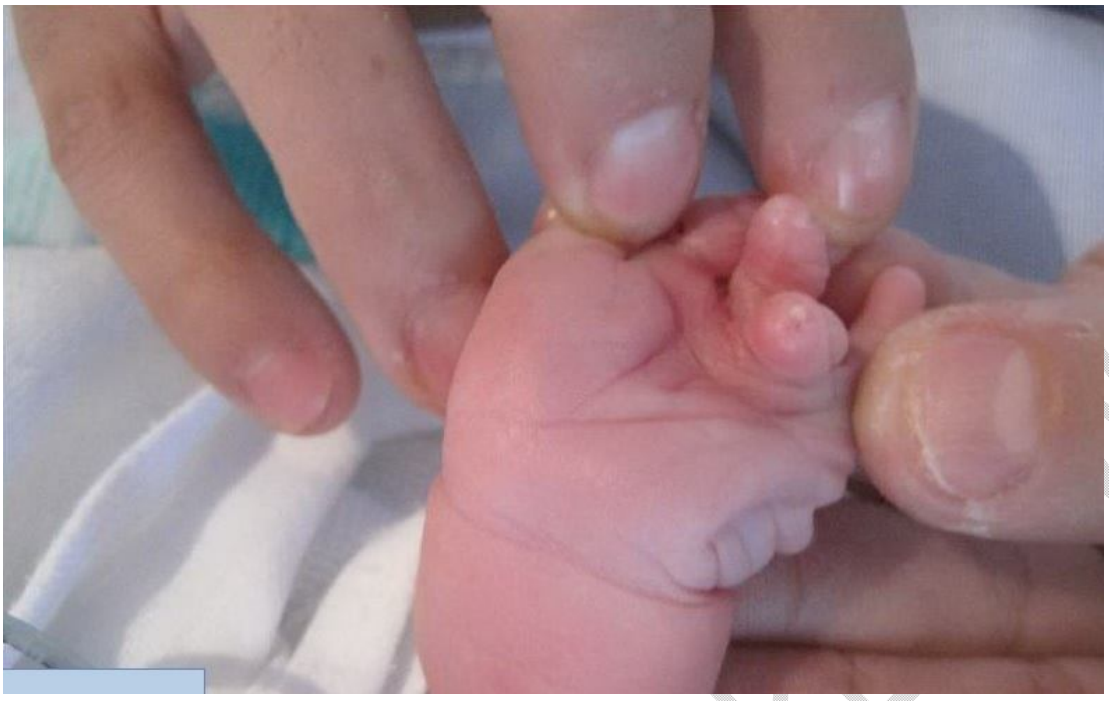


Figure 4. Fusion of 4th and 5th fingers.



5a



5b

Figure 5. Ring straps around the forearm (5a) and 2 fingers of the left hand (5b)

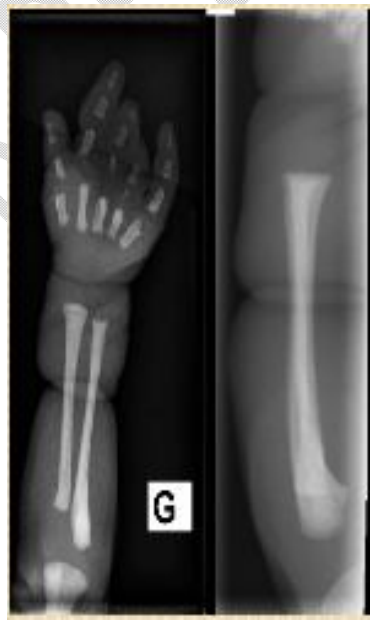


Figure 6. Short aspect of the 3rd and 4th metacarpals, dysmorphic aspect of the phalanges of the 2nd and 3rd ray. Absence of ossification nucleus of the 3rd phalanx of the 4th ray.

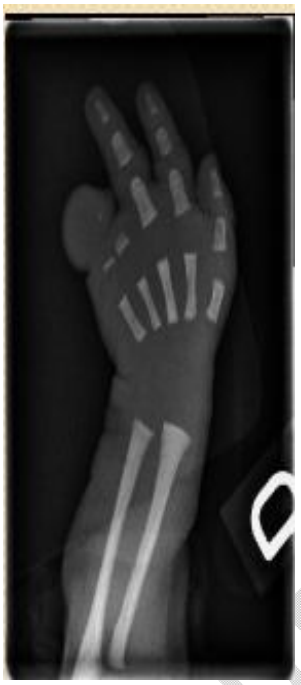


Figure 7. Short aspect of the 1st phalanges of the 4th and 5th rays of which there is only the base and the proximal metaphysis. Absence of middle and distal phalanx at the level of 4th and 5th ray, only a small nucleus of ossification is visualized at the level of the fleshy bud.

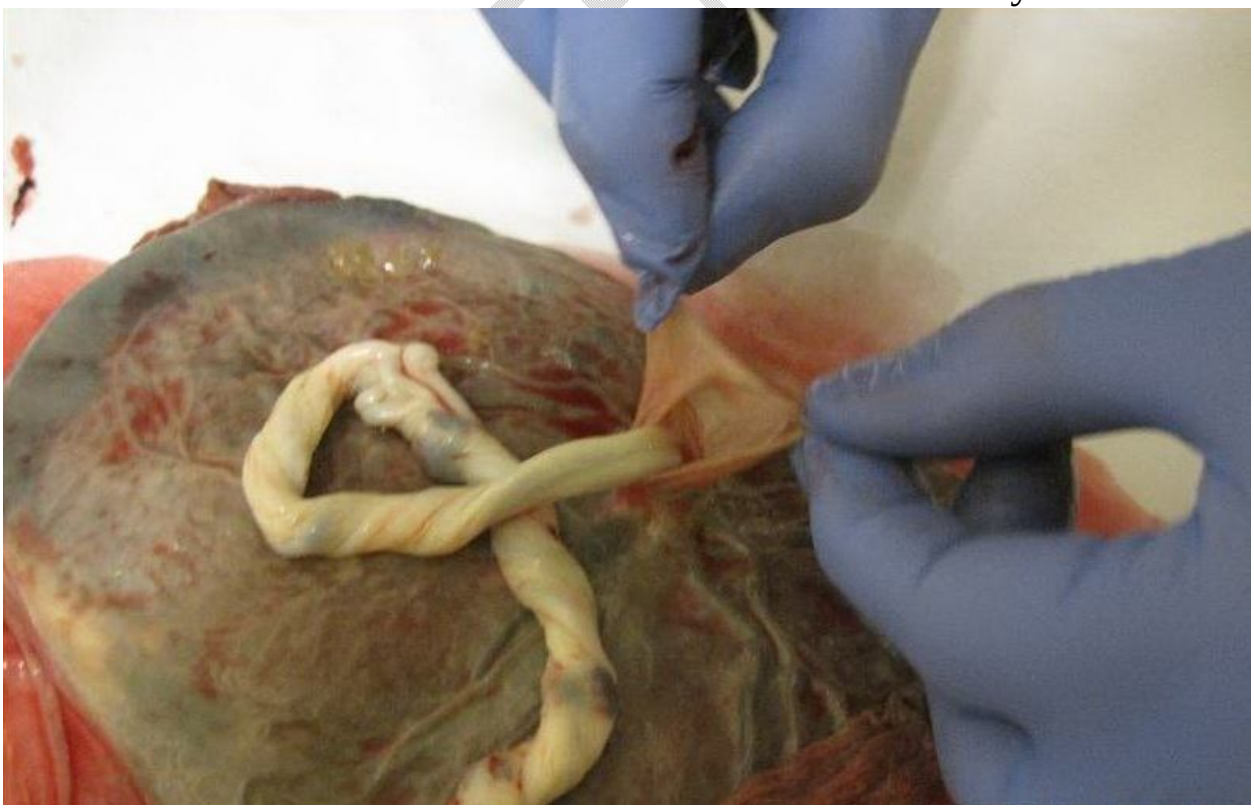


Figure 8. Amniotic bands of the placenta

DISCUSSION

ABD corresponds to a set of malformations ranging from constriction and lymphoedema of the fingers to multiple congenital anomalies mainly affecting the limbs, but also the craniofacial region and the thoracoabdominal axis [1,2]. Two theories have been discussed to explain its pathogenesis: according to the endogenous theory of Streeter in 1930, it is a malformation of the germinal disc which leads to a disturbance in the development of the amnion [4]. The exogenous theory of Torpin in 1965 opposes the previous one; it is based on the premature rupture of the amnion, which debris come to form constrictive bands on the fetal skin surface [5]. Despite numerous studies, none of the two theories has been demonstrated so far.

ABD and Limb Body Wall Complex (LBWC) syndrome are collectively referred to as amniotic band syndrome. LBWC is a malformation complex characterized by severe, multiple and fatal congenital anomalies of the fetus, with exencephaly and/or encephalocele, severe anterior parie-

tal malformations and limb malformations, with or without facial clefts [6, 7]. ABD is a relatively rare pathology, its incidence is between 1/1,200 and 1/15,000 live births. There is no racial predisposition or sex bond [1,6]. Limb malformations are found in 65% of cases, and facial malformations are present in 48% of cases [1–3].

The diagnosis of ABD is classically based on the existence of at least two of the following three signs: furrows, amputations and pseudosyndactylies. The severity of these malformations is highly variable, ranging from isolated skin furrows to visceral malformations that are often incompatible with life. The frequency of these malformations within this disease is very variable according to the authors [1,3,8,9]. Limb abnormalities are very varied and are the most common manifestations of ABD in 65% of cases. These can be cutaneous furrows from stricture, amputations, pseudosyndactylies or club feet [1, 3, 8, 9]. Finger constriction and amputation were abnormalities observed in our case.
The prognosis of ABD in the presence of craniofacial and visceral polymalformations is known and bleak. The situation is quite different in the case of superficial and isolated constriction of a limb. A medical termination of pregnancy is generally proposed in the presence of severe craniofacial and visceral malformations, while isolated malformations of the limbs are accessible to surgical treatment at birth. In case of amniotic band associated with isolated constriction of the limb, antenatal resection of the band by fetoscopy can be proposed to avoid the occurrence of amputation in utero. However, antenatal treatment remains controversial today [8-10]. Surgical treatment of constriction furrows undertaken in an emergency to save the limb is a simple technique that gives excellent results. Amniotic amputations require few corrective actions, taking into account above all the extraordinary adaptation of children to their handicap. Functional prostheses are of little use, social life prostheses help to overcome difficult times [4,5,12]. Our case progressed well after the surgery.

CONCLUSION

ABD remains rare, however its detection must be systematic in the neonatal period. It is an acquired embryofoetopathy grouping together a set of asymmetrical malformations, mainly involving the limbs and the craniofacial region. Although the pathogenesis of this disease remains controversial, this malformation syndrome requires multidisciplinary management of pregnancy, involving obstetricians, pediatricians, plastic and pediatric surgeons, radiologists. A medical termination of pregnancy may be proposed when the fetal malformations are recognized as being incompatible with life.

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