Amniotic band syndrome is a well described clinical entity presenting with deformities of the limbs, thorax, craniofacial skeleton, soft tissues and umbilical cord, but it still lacks a precise definition and a coherent hypothesis for its pathogenesis. We report on a case of first trimester diagnosis of amniotic band syndrome by sonography and fetoscopy. This revealed multiple abnormalities including facial cleft, brain and limb deformities; the appearance of the amniotic cavity was that of a cobweb containing the fetus. Post-mortem examination and histopathological studies confirmed the diagnosis of amniotic band syndrome. These results may enhance the knowledge of its natural course. In addition, based on histological and newly identified ultrastructural features, we present a hypothesis which could help to explain the aetiopathogenesis of the amniotic band syndrome.

Key words: amniotic band syndrome/facial cleft/fetoscopy/limb amputation/prenatal ultrasound

Introduction

Amniotic bands consist of fibrous strands extending from the outer surface of the chorion into the amniotic cavity. Their frequency has been estimated to range between 1 in 1234 and 1 in 15 000 live births (Sistrom and Ferguson, 1993), but mainly between 1 in 56 in pre-viable fetuses (Kalousek and Bamforth, 1988). They have been associated with preterm delivery and low birthweight (Wehbeh et al., 1993), but mainly with amniotic band syndrome (ABS). ABS is a well described clinical entity presenting with deformities of the limbs, thorax, craniofacial skeleton, soft tissues and umbilical cord (Graf et al., 1997) in a non-embryonic distribution. The malformations may present with varying degrees of severity and are found individually or collectively. However, although this entity has been recognised for more than three centuries (Portal, 1685) it still lacks a precise definition and its pathogenesis is still debated.

Two major pathogenetic theories have been proposed that disagree as to whether the amniotic bands themselves cause the fetal anomalies (exogenous theory) or are a by-product of a primary defect in the embryonic germ disc (endogenous theory).

The exogenous theory, initially proposed in the nineteenth century and then developed by Torpin (1965), holds that the syndrome arises following premature rupture of the amnion, which results in the fetus passing into the extracoelomic space. The mesodermic outer surface of the amnion would then produce fibrous strings, which entangle the extremities and operate as constrictive bands causing amputations. In a similar way, ‘slash defects’ of the face would arise as a result of the fetus swallowing the amniotic bands, and hence resulting in the formation of a non-embryonic facial cleft (Lockwood et al., 1989). The alternative endogenous theory, initially proposed by Streeter (1930), suggests that the amniotic bands and fetal anomalies seen are caused by an intrinsic, germ-line, developmental derangement. The aetiology of this process may be polygenic or teratogenic. This view is supported by reports that amniotic bands are significantly more common in monozygotic twins, a condition considered to result from a teratogenic effect (Lockwood et al., 1988). This paper reports on the early first trimester diagnosis of ABS by ultrasound, fetoscopy and pathological examination. A concept about its pathogenesis and perspectives for an early therapeutic intervention are presented.

Case report

A 26 year old woman, gravida 2, para 0, underwent a routine 10–14 weeks anomaly scan and assessment of the nuchal translucency thickness. This showed a viable fetus with 67 mm crown–rump length, 21.5 mm bi-parietal diameter (BPD) and 10 mm femur length. The nuchal translucency was 1.0 mm and a normal amount of amniotic fluid was noted. Examination of the fetal anatomy revealed flexed and grossly deformed forearms (Figure 1 left) and bilateral talipes. An amniotic band was identified, which entangled the distal end of all four limbs and extended laterally to the uterine sidewall (Figure 1 left). Cranial examination demonstrated unilateral ventriculomegaly, and suggested a median facial cleft (Figure 1 right). Fetal echocardiography revealed normal situs and cardiac connections and normal continuity of the inter-ventricular septum with the anterior wall of the aorta. As termination of the pregnancy was a strong consideration we offered the patient confirmation of the sono-graphic findings by diagnostic fetoscopy. This was performed under local anaesthesia with a 1 mm 0° diagnostic endoscope (Karl Storz, Tuttlingen, Germany) through an 18 gauge needle.
The facial anatomy appeared deformed by a central cleft-like fissure affecting the nose but not the lips. The forehead was prominent and there was micrognathia present. The limbs were seen to be entangled in multiple, cobweb-like, thin, floppy and ‘sticky’ amniotic bands (Figure 2a). These were seen to originate from the inner aspect of the amniotic membranes enclosing an apparently intact amniotic cavity (Figure 2b). Fetal movements were seen to be restricted by the constrictive amniotic bands. Bilateral talipes and flexed bilateral wrists, seen on ultrasound, were confirmed and additionally, proximal phalanx amputation defects of the second and third fingers of the right hand and of the first toe of the right foot were noted together with marked necrosis and oedema of the distal phalanges. A band was seen to arise from the fetal chin and another to attach and constrict the umbilical cord. At the end of the procedure chorionic villous sampling was performed, which revealed a normal 46, XY karyotype. A screening for maternal infections was negative. The parents were counselled with regard to the findings and prognosis and they elected to terminate the pregnancy. An uneventful surgical termination of pregnancy was performed at 12 + 6 weeks.

**Pathological examination**

Examination of the affected limbs showed some amputated digits and toes, others showed amniotic bands tightly strapped around them (Figure 3a). Histological examination of the affected fingers and toes at the level of the constriction showed atrophy of subepidermal structures. The overlying skin appeared atrophic and simplified into a one-cell thick epidermal surface. The constricting amniotic bands appeared featureless under the microscope. They were made of amorphous acellular matrix. Above the site of constriction the tip of the affected fingers and toes showed marked oedema and interstitial haemorrhage. In all instances the amniotic band never showed fusion with the adjacent surface epithelium. Scanning electron microscopy examination of amniotic bands showed thin filaments <800 nm in diameter. They appeared uniform in shape and were arranged in parallel next to each other. In addition, they showed short microappendages with a round prominent tip. These were distributed at almost regular intervals along the shaft of the fibres (Figure 3b).
Figure 3. (a) Macroscopic view of the right foot showing amniotic bands tightly strapped around the second and third toe. Amputation of the first toe has already taken place. (b) Scanning electron microscopic examination of the amniotic bands (×10 000). The filaments are less than 800 nm in diameter. Note the microappendages with a round prominent tip, which are distributed along the shaft of the fibres. Our hypothesis suggests that these thin fibres interlocked between themselves around the limbs producing a ‘velcro’-like effect. This may lead to constricting bands.

Discussion

Amniotic band syndrome is not an uncommon complication of pregnancy; however, despite its clinical importance there is no firm evidence concerning its aetiology and its natural history is still debated (Bronshtein and Zimmer, 1997; Evans, 1997). Irrespective of the different pathogenetic theories, there is little doubt that amniotic band syndrome is associated with a wide spectrum of defects in an asymmetrical and non-embryological distribution. These include severe and often lethal deformities such as acrania, microcephaly, encephalocele, cleft palate and thoracoabdominoschisis. The vast majority of cases, however, display some form of limb involvement (Moerman et al., 1992). Virtually all reports of prenatal diagnosis of ABS-related malformations made by ultrasound involved pregnancies in the second and/or third trimester of pregnancy with normal karyotype (Mahoney et al., 1985; Yamaguchi et al., 1988). Thus, intrauterine intervention, though technically feasible and successfully applied for other indications (Quintero et al., 1994; Ville et al., 1995), did not seem to be appropriate. The only available report of first trimester diagnosis, however, showed entrapment of a viable fetus by a torn amnion through serial scans, which resulted in a twisting of the umbilical cord, and this eventually was thought to be the reason for fetal death (Nishi and Nakano, 1994). Recently, serial scans documented constriction of both legs that led progressively to amputation of both lower extremities (Tadmore et al., 1997).

Our case provides further insight into the natural course of amniotic band syndrome. Sonographic and fetoscopic images clearly demonstrated that these membranes were wrapped over the limbs and other parts of the body, trapping it in a cobweb-like structure and significantly restricting the fetal movements. The presence of thin fibres causing countless hook-like microappendages could suggest that these thin fibres interlocked between themselves around the limbs producing a ‘velcro’-like effect. Histological examination showed tightly wrapped amniotic bands, but no fusion with the skin surface they were in contact with. Thus, surgical release of the constrictions could be a therapeutic option. Intrauterine intervention is now technically feasible and has been successfully applied in a number of clinical conditions such as twin—twin transfusion syndrome and other indications (Quintero et al., 1994; Ville et al., 1995). The first description of a successful attempt at releasing fetal limbs from constricting membranes by fetoscopy has been reported recently (Quintero et al., 1994). Surgical removal might prove particularly useful in cases with limited damage, e.g. constriction of the umbilical cord, a condition considered to be present in about 5% of amniotic band syndromes and associated with a high risk of intrauterine fetal death (Kanayama et al., 1995).

This case adds further information about the pathogenesis of amniotic band syndrome during the first trimester. Ultrasound and fetoscopic examination together with histological findings allow us to postulate that a progressive and potentially reversible phenomenon drives the aetiopathogenesis of the amniotic band syndrome.

References


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