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Body-stalk anomaly. A case report and review of the literature

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Abstract

Body-stalk anomaly is a severe and lethal anomaly, characterized by an anatomic defect in the ventral abdominal wall and exit of the viscera of the abdominal cavity, associating spine and limb deformities and absence or shortening of the umbilical cord. The prenatal diagnosis is suggested by ultrasound means and should be confirmed by histopathology evaluation. We present a case of a body-stalk anomaly incidentally diagnosed at 23 weeks of gestation, during routine ultrasound evaluation. Due to its fatal prognosis it is imperative to differentiate it from other abdominal wall defects.
Keywords: *body-stalk anomaly, ultrasonography*

Introduction

Body stalk anomaly (BSA) is the rarest and most severe of abdominal wall defects. It is characterized by an enlarged abdominal wall defect, severe spine deformities and a rudimentary umbilical cord. It is often complicated by anomalies of head, face and extremities[1-7].

The BSA, also known as limb body wall complex, amniotic band syndrome, cyllosomas or aplasia of the cord, has a wide phenotypic spectrum of defects[8-11]. There is no general consensus on the most appropriate name[12-17]. The differential diagnosis made with other anomalies such as gastroschisis or omphalocele, is important due to its fatal prognosis.

We report a case of BSA diagnosed in the second trimesterof pregnancy, during routine ultrasound.

Case report

A 33 year-old primiparous, poorly screened due to the lack of compliance to prenatal visits (Figure 1), was diagnosed at the emergency room with abdominal wall defect during a routine ultrasound examination at 23 weeks of gestation performed to establish the foetal sex. The patient was sent to a second evaluation in a tertiary centre for accurate diagnosis and prognosis.

The ultrasound revealed a live women foetus with large abdominal wall defect (Figure 2) with liver, stomach, intestinal loops, kidney, cardiac apex outside the body, thoracic hypoplasia due to thoracic aortic disease amendment with pulmonary hypoplasia (Figure 3), severe kyphosis and pronounced lumbosacral scoliosis (Figure 4). No membrane is seen surrounding the abdominal viscera. No intestinal loop movement is seen during examination. The placental site was on the anterior wall of the uterus, but the foetus was laying face down to the posterior wall, with the abdominal viscera being in close contact to it, showing no change in its position or any active limb movements during the examination and making harder the evaluation, despite the normal amniotic fluid volume. The umbilical cord was ill seen on Doppler examination, being occulted by the foetus and fixing it in that immobile position, spine back to the placenta, raising the suspicion of a short umbilical cord. These findings were suggestive for BSA.

The patient had not undergone a 1st trimester scan and serum biochemistry screening for Down's syndrome, therefore an amniocentecis was performed. There was no family history of cocaine intake, any malformations or consanguinity. The family was counselled regarding the fatal condition of the foetus and choose to continue.

She was admitted in our hospital several days later with vaginal bleeding and contractions, spontaneously aborting a malformed female foetus with no sign of viability, weighing approximately 400 g.

Post-mortem examination of the foetus and the foetal annexes confirmed the findings seen in ultrasonography: a 400 g women foetus with multiple malformations, some which had not been seen on ultrasound.

Skeletal deformities are represented by a severely kyphoscoliotic spine (Figure 5) changing the body axis. A large anterior abdomen wall defect is present with liver, stomach, small and large bowels, right kidney, cardiac apex and two right lung lobes outside the body (Figure 6). The abdominal viscera are free in the amniotic cavity, no membrane is found surrounding them. The peritoneal cavity is narrow containing the urinary bladder and internal genital organs. The chest examination showed a congenital diaphragmatic hernia with the ascent of the left kidney in the thoracic cavity, along with hypoplastic lungs, with three lobes on the right lung and four lobes in left one. Internal hydrocephalia is present in the brain. The discoid placenta weighed 125 g and had a short umbilical cord.

Microscopic examination of the placenta and viscera showed no recognizable abnormalities.

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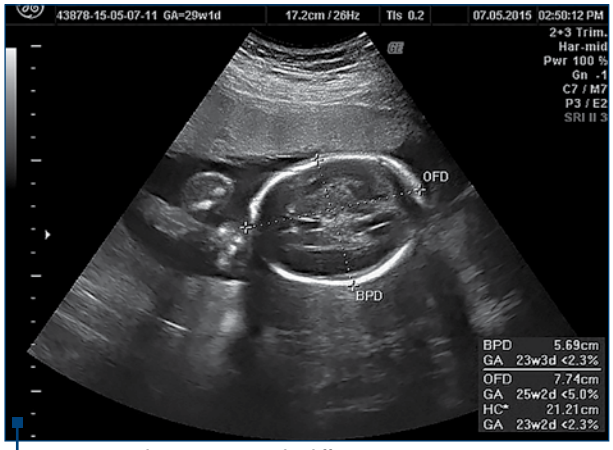

Figure 1. Foetal measurements: the difference in estimating gestational age

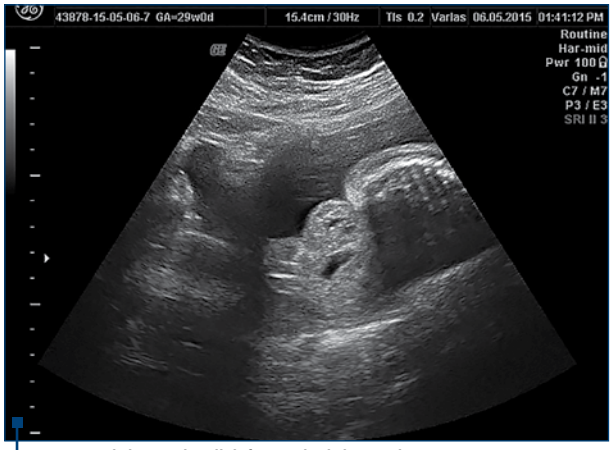

Figure 2. Abdominal wall defect with abdominal viscera of the abdominal cavity

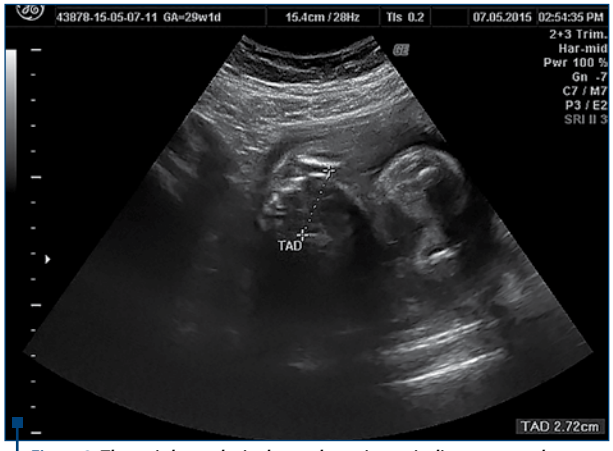

Figure 3. Thoracic hypoplasia due to thoracic aortic disease amendment

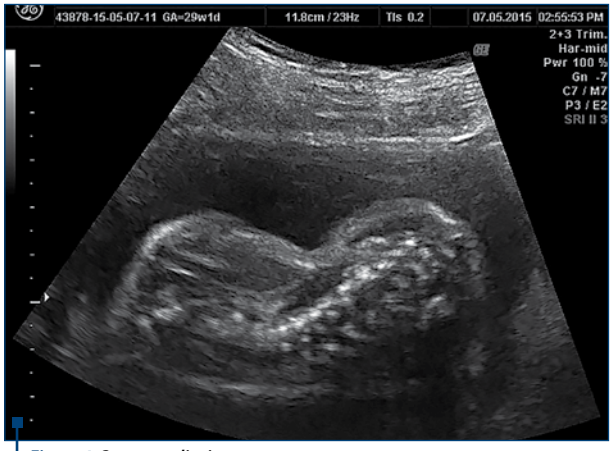

Figure 4. Severe scoliosis

Figure 5. Severely kyphoscoliotic spine changing the body axis

Figure 6. Anterior abdominal wall defect with liver, intestinal loops, stomach, cardiac apex, right kidney and right lung lobes outside the body. No membrane is seen surrounding the viscera

Three weeks after amniocentesis the result of the genetic examination of amniotic fluid cells showed normal female karyotype.

Discussion

BSA is a rare syndrome, with an incidence difficult to estimate. Different studies showed different prevalence:

one multicenter study found the prevalence around 1 in 7500 foetuses at 10-14 weeks of gestation[2], while in another the prevalence was reported at1 per 32000 births[18-24]. With more pregnancies obtained by in vitro fertilization techniques secondary to progresses made in this direction, BSA cases are currently being reported in twin pregnancies, either monozygotic or dizygotic[21,25,26] and even in triplet pregnancy[4,8]. There is evidence that supports the association of BSA with maternal abuse of cocaine[6].

The pathogenesis of BSA is uncertain. Several mechanisms try to explain the development of BSA: early amnion rupture and amniotic bands, vascular disruption of the early embryo, or an abnormality in the germinal disk that leads to the formation of an anomalous amniotic cavity[1,16].

Multiple amniotic bands interrupt embryogenesis and make the foetus to lie outside the amniotic cavity is the mechanism suggested by early amnion rupture theory in BSA occurrence[16]. This theory is challenged by recent studies, who failed finding any evidence to support the relation between amnion rupture and fibrotic bands and BSA[4]. Any event that compromise embryonic flow during the first 4-6 weeks of gestation may lead to the failure of closure of the ventral wall and persistence of the extra-embryonic coelomic cavity[12,17]. This second theory - the vascular or the endogenous theory, explain the cases of BSA induced by cocaine intake. Cocaine decreases placental blood flow through its vasospastic properties, acting as a teratogen agent[15]. But both amniotic band theory and vascular theory failed to explain all the anomalies observed in BSA[19].

Nowadays, the most commonly accepted hypothesis is Streeter's 1930 theory of abnormal embryonic folding[18], modified in 1989 by Hartwig[20]. While the trilaminar embryo is transformed into a cylindrical foetus by folding in cephalic, caudal and both lateral folds during the 5th week of gestation, aberration of the process may occur. The abnormal folding in all the axes lead to body stalk anomaly with persistence of the extra-embryonic coelomic cavity. Depending of the degree of aberrant development of each fold, various malformations are associated with body stalk anomaly[1,2,4].

Van Allen et al. set forth the diagnostic criteria for BSA in 1987. Two of the three following anomalies must be presented to establish a positive diagnosis[12,16]:

1. Exencephaly/encephalocele with facial clefts
2. Thoraco and abdominoschisis (midline defect)
3. Limb defect (i.e. club foot, polydactyly, oligodactyly, syndactyly, brachydactyly, Amelia)

Two main phenotypes have been described in the literature[10,13], each being consequence of different pathogenic mechanisms[13]:

1. The placental-cranial type: associating craniofacial defects (encephalocele/exencephaly associated with facial clefts) and amniotic bands between the cranial defects and placenta -pathogenic mechanism proposed is early vascular disruption.

2. The placental-abdominal type, no craniofacial defects are present, but associating urogenital anomalies, anal atresia, lumbosacral meningocele, short cord, persistence of extra embryonic coelom and intact amnion - it seems to be due to intrinsic embryonic abnormal development.

Our case is a placental–abdominal phenotype.

BSA is most frequently diagnosed during the second trimester of pregnancy by ultrasound[4]. Cases reported found in the third trimester are rare[17,26]. With the progress made in early ultrasound assessment of the pregnancy, nowadays the BSA is being diagnosed in the first trimester. Since 2000, when the first case of BSA was diagnosed at 9+5 weeks[22], the literature reports of cases discovered in early first trimester increased[3,5,21,23,25,27]. Using the nuchal translucency as maker for first trimester anomalies, we found a 2009 and two 2011 studies stating the importance of first trimester screening in early detection of BSA[5,28,29].

Magnetic resonance imaging technique confirmed a large anterior wall defect with herniation of the liver and bowel, limb abnormalities and scoliosis in a 14 weeks foetus previouslydiagnosed with body stalk anomalyusing ultrasonography[30-33]. Such report won't diminish the importance of early ultrasound diagnosis, mostly because it is a cheap widespread investigation for the assessment of foetal development.

A BSA should be considered when, during an ultrasound, is found a foetus with malformed and/or short umbilical cord in association with abdominoschisis/thoracoabdominoschisis with eventration of different organs, skeletal deformities, neural tube defects, persistence of the extra-embryonic celomic cavity[4]. In those cases, if laboratory testing is available, maternal serum alpha fetoprotein levels should be tested, often being elevated in the second trimester, although not as a specific marker for this anomaly[3,4,10].

If foetal karyotyping is performed, is almost always found to be normal, therefore there is no need for routine karyotyping in these cases[4]. Cases with foetal aneuploidies and BSA, diagnosed by cytogenetic analysis of chorionic villus sampling or amniotic fluid are rare and the general believe is that BSA is not a consequence of foetal aneuploidies[2,4,28].

Due to its fatal prognosis, BSA must be differentiated from other complex plurimalformative syndromssuch as: pentalogy of Cantrell, omphalocele, exostrophy, imperforateanus, spinal defects, isolated gastroschisis or omphalocele, short umbilical cord syndrome[1,16,24,31].

Whenever a pregnant woman faces the BSA diagnosis in her embryo or foetus, counselling must be offered. The patient and its family are to be assured that the disease has no risk of recurrence[17]. Only one case is mentioned in literature, the patient delivered two consecutive infants with the same condition[30]. The increased incidence of BSA in monozygotic versus dizygotic twins or singleton pregnancy is probably due to a disorder in early embryonic cleavage and represents an intermediate stage between monoamniotic twins to conjoined twins, being part of a twining process, without any risk of recurrence[1,32]. Pregnancy termination should be considered in singletons as soon as medical diagnosis is confirmed, avoiding extended

operative interventions[9], even though most foetuses are spontaneously aborted and the remaining are stillborn[10].

In twin pregnancies is hard to make a decision, the management focusing in well-being of the unaffected twin, taking also in the account the risk of preterm delivery[9,21]. Proper management should be selective feticide of the affected foetus in dichorionic twins. It should be taken into account that the foetus with the BSA is never viable, and if no other complications or outside interventions occur, the expected pregnancy outcome consists in the survival only of the healthy foetus[21].

The same no intervention attitude can be applied for monochorionic twins.

If delivered, those infants usually die soon after birth because of severe pulmonary hypoplasia[2]. Postnatal survival for a significant duration is extremely rare. We found in literature a case of a long-term survivor, the infant having several handicaps, including mental retardation[11].

Conclusions

BSA is a rare multiple malformation syndrome with fatal prognosis. Whenever ventral body wall defect is found, BSA diagnosis should be considered and careful ultrasound examination of the foetus must be performed. First trimester screening should become a "must-have" for all pregnant women. It is important to distinguish BSA from other anterior wall defects for appropriate management. ■

Conflict of interests

The authors declare that there is no conflict of interests regarding the publication of this paper.

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