CASE REPORT

AMNIOTIC BANDS, ANORECTAL AND BLADDER AGENESIS AN UNIQUE ASSOCIATION IN A TWIN PREGNANCY WITH FETUS PAPYRACEUS

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Summary

Poor cloacal development and an inadequate septation can generate a wide range of genitourinary and terminal portions of the digestive tract malformations. The bladder agenesis is an extremely rare congenital genitourinary anomaly. Successful treatment and the long-term prognosis are usually poor because of the associated abnormalities. We want to report an uncommon case related to cloacal maldevelopment, a complex association of urogenital and hindgut abnormalities in a twin pregnancy with fetus papyraceus (mummified) and amniotic bands. It is about a twin pregnancy with one fetus stopped evolving at 12 weeks of gestation and another fetus that has continued to develop up to 23 weeks. The birth occurred at 23 weeks by small Caesarean section and we extracted a mummified fetus of 45 grams and a nonviable female fetus weighed 420 g with multiple malformations. Urogenital anomalies consisted of absence of vagina and urethra, bladder agenesis with ectopic ureteric opening. In addition, this second girl fetus had anorectal agenesis, imperforate anus without fistula as a hindgut anomaly but with omphalocèle and a blind sigmoidum at this level. Clinical, anatomopathological evaluation and embryological review are made to explain the concomitant occurrence of these rare malformations.

Key words: Cloacal abnormalities, Cloacal extrophy, bladder agenesis, ectopic ureter, OEIS complex, amniotic bands, imperforate anus, twin pregnancy with fetus papyraceus

Résumé

Bandes amniotiques, agénésie ano-rectale et de la vessie, association unique dans une grossesse gémellaire avec un foetus papyracé

Le sous-développement du cloaque et le cloisonnement insuffisant peuvent produire une large gamme de malformations uro-génitales et des parties terminales du système digestif. L’agénésie de la vessie est une anomalie congénitale génito-urinaire extrêmement rare. Le traitement réussi et le pronostic à long terme sont d’habitude faibles à cause d’anomalies associées. Nous voulons rapporter un cas rare de sous-développement du cloaque, une association complexe d’anomalies urogénitales et du tube digestif dans une grossesse gémellaire avec un foetus papyracé (mommifié) et des bandes amniotiques. Il s’agit d’une grossesse gémellaire avec un foetus qui avait cessé d’évoluer à 12 semaines de gestation et un autre foetus qui a continué à grandir jusqu’à 23 semaines. La naissance a eu lieu à 23 semaines par la petite césarienne et nous avons extrait un foetus momifié de 45 gr et un foetus femme non viable pesant 420 gr aux malformations multiples. Les anomalies urogénitales résidaient en l’absence du vagin et de l’urètre, l’agénésie de la vessie à l’ouverture ectopique des urètères. En plus, ce deuxième foetus avait une agénésie ano-rectale, une imperforation anale sans fistule comme une anomalie de partie terminale du tube digestif, mais avec l’omphalocele et le côlon sigmoïde aveugle à ce niveau. Une review clinique, anatomo-pathologique et embryologique est faite afin d’expliquer l’occurrence concomitante de ces malformations rares. Mots clés: anomalies du cloaque, extrophie du cloaque, agénésie vésicale, urètre ectopique, complexe d’OEIS, bandes amniotiques, imperforation anale, grossesse gémellaire avec un foetus papyracé.

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INTRODUCTION

Complete absence or agenesis of the urinary bladder is an extremely rare developmental anomaly. This report deals with an instance of agenesis of the urinary bladder with several hindgut anomalies. The cause of agenesis of the bladder is uncertain. The exact embryological maldevelopment leading to bladder agenesis is difficult to explain but it is proposed that these anomalies occur during weeks 5-7 of embryogenesis. Bladder starts to develop by 5th week of gestation when cloaca is divided by a uro-rectal septum into urogenital sinus and anorectal canal [1]. Bladder agenesis may be the result of secondary loss of the anterior division of the cloaca, perhaps owing to a lack of distention with urine caused by failure of incorporation of the mesonephric ducts and ureters into the trigone, thus preventing urine from accumulating in the bladder [2,3]. In most cases in which the bladder is absent this is also associated with the most severe form of ureteric ectopia. However, ectopic ureter is essential for compatibility with life because it preserves the renal function. Ano-rectal malformations are found in about 1:5000 live births. They are commonly subdivided into low, intermediate and high anomalies. High anorectal malformations have a blind end of rectum or sigmoidum located above the pubo-rectal muscles. Rarely in females the lower vagina may also fail to form. The most likely explanations for anorectal ageneses include: abnormal formation of the urorectal septum, excessive obliteration of the embryonic tailgut and dorsal cloaca.

CASE REPORT

A 19 year old woman, G II, P II, diagnosed at eight weeks of gestation in a territorial hospital with diamniotic, monochorionic twin pregnancy, with one fetus that stopped evolving around 12 weeks, is sent to our hospital at 22 weeks because of a large lower anterior abdominal cyst of unknown origin, suspected omphalocele and intrauterine growth restriction to establish therapeutic conduct. In the patient’s personal history we noted that she is nonsmoker, without consanguinity in couple, without teratogenic potential disease, without diabetes, exposure to radiation or toxic substances in the first trimester. A screening for chromosomal abnormalities in the first trimester or other tests to assess fetal DNA, the double test and the nuchal translucency were not performed. Without significant data in family history, our patient gave birth by Cæsarean section in 2012, the first child was perfectly normal. On admission the patient described irregular uterine contractions; clinically we diagnosed a 22 weeks pregnancy with blood tests within normal limits.

The ultrasound at 22 weeks of gestation showing twin pregnancy with one viable fetus with fetal heart rate of 136/minute, maturity of 19 weeks and 3 days with reduced mobility by the presence of amniotic bands at the level left upper limb and left lower limb respectively (fig. 1). At the lower uterine segment we find another small amniotic sac with a non-viable, atrophied fetus with absent cardiac activity, CRL =34.5 mm, maturity of 10 weeks and 3 days (fig. 2): Single placenta anterior in upper uterine segment and normal amniotic fluid are visible.

We proceed to a systematic sonographic examination and notice a defect in anterior abdominal wall and suggestive image for omphalocele. The bladder is not visualized in the pelvis. Fixed at the anterior abdominal wall we found a round, large, thin walled transonic structure with dimensions 87/62 mm (fig. 3).

Based on the direction of the umbilical arteries (fig. 4) but also on the fact that during the examination we’ve seen a similar image of a turbulent ureteral jet (fig. 5) inside of this formation it rises the suspicion of bladder exstrophy. Throughout the entire examination (about 60 minutes) the fetus moves extremely low and the bladder was not seen in pelvis. The fetus had normal brain and spine, normal cardiac morphology and vascular connections.
The sonographic diagnosis was: Twin pregnancy with one dead fetus (papyraceus) and one living fetus with multiple abnormalities: possible extrophy of cloaca or bladder, omphalocele, amniotic bands and about three weeks intrauterine growth restriction.

Finalisation of pregnancy was initiated because of ruptured membranes with maternal bleeding and low fetal prognosis. The patient accepted and she consented to small caesarean section under spinal anesthesia which revealed a twin gestation: one a non-viable feminin fetus, weighing 420g was delivered (fig. 6). The ultrasound findings were confirmed. The extrophy of cloaca, omphalocele and imperforate anus were visualized. A single placenta was delivered and careful examination revealed there was another atrophied, mummified (papyraceus) female fetus (fig. 7) with an extremely short and thin umbilical cord apparently trapped into an amniotic band (fig. 8). Our conclusion of diamniotic monochorionic twin pregnancy with fetus papyraceus was reached.

At autopsy, the fetus weighed 420 g and on external examination a number of anomalies were noted. The genitalia were feminin with a normal uterus. The head and spine were normal. The most striking abnormality was an anterior abdominal wall defect, from which gastrointestinal organs were protruding into an omphalocele. The organs identified included mesenteron, ileum, cecum with appendix, ascending and transverse colon and a blind finished descending colon. Although these organs were encased by the peritoneum, a membrane like structure was present near the anterior abdominal wall opening (Fig. 9). The umbilical cord was short and was found in this membrane like structure (possible cloacal membrane) that was covering the omphalocele. On internal examination, there was agenesis of anal canal and rectum along with agenesis of genitourinary tract: agenesis of bladder, vagina and urethra. The uterus was normal and containing at the lower level a stratified squamous tissue (possibly incomplete developed vaginal wall). It has also been found a supernumerary left adrenal gland and ectopic ureters at the pelvic peritoneum level, apparently having communication with this cloacal
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The remaining organs appeared normal on gross examination and congested on microscopy. The karyotype was normal (46XX).

The second fetus, mummified, atrophied (papyraceus) had an inconclusive autopsy and internal organs apparently in normal range.

**DISCUSSION**

Differential diagnosis was necessary. In omphalocele the sac, which is formed from an outpouching of peritoneum, protrudes in the midline through the umbilicus. Gastrochisis is a similar birth defect, but in gastrochisis the umbilical cord is not involved and the lesion is usually to the right of midline. Parts of organs may be free in the amniotic fluid and not enclosed in a membranous (peritoneal) sac. Gastrochisis is less frequently associated with other defects than omphalocele. But in both cases there is a normal bladder. Bladder extrophy (also known as Ectopia vesicae) is a congenital anomaly that exists along the spectrum of the extrophy-epispadias complex and most notably involves protrusion of the urinary bladder through a defect in the abdominal wall. Its presentation is variable, often including abnormalities of the bony pelvis, pelvic floor, and genitalia.
persistent cloaca is a complex ano-rectal congenital disorder in which the rectum, vagina, and urinary tract meet and fuse, creating a cloaca, a single common channel. Cloacal extrophy is the most severe cloacal anomaly. It involves an anterior abdominal wall defect in which 2 hemibladders are visible and are separated by a midline intestinal plate, an omphalocele, and an imperforate anus. OEIS complex (omphalocele, extrophy of cloaca/or bladder, imperforate anus and spine defects) is a rare combination of serious defects with incidence 1 at 400,000 deliveries, described by Carey at all in 1978 [2]. Amniotic band syndrome (ABS) is a congenital disorder caused by entrapment of fetal parts (usually a limb or digits) in fibrous amniotic bands while in utero. Amniotic banding affects approximately 1 in 1200 live births. It is also believed to be the cause of 178 in 10,000 miscarriages [5]. Limb body wall complex (LBWC) is a rare fetal malformation of unknown origins. Traditionally diagnosis has been based on the Van Allen et al., criteria, i.e. the presence of two out of three of the following anomalies: Exencephaly or encephalocele with facial clefts, thoraco and/or abdominoschisis and limb defects. [6].

Pathogenesis

Exstrophy of the bladder (EB) and exstrophy of the cloaca (EC) are generally recognizable as distinct clinical entities. In patients with EB, the posterior bladder wall is exposed through a midline defect of the abdomen. The umbilicus is inferiorly displaced and located close to the superior margin of the exstrophic bladder. In contrast to classic EB, EC is commonly associated with omphalocele, spinal defects, and incompletely formed external genitalia and is always associated with imperforate anus. Some authors state that EC and EB constitute two distinct disorders, but others consider them part of a “continuum,” representing different levels of severity within the same spectrum [7].

Fetus papyraceus is a rare condition which describes a mummified fetus in a multiple gestation pregnancy in which one fetus dies and becomes flattened between the membranes of the other fetus and uterine wall. A possible mechanism is the transfixion of thoracoblastic proteins from the macerated twin to the surviving twin, leading to disseminated intravascular coagulation (DIC) which then leads to intrauterine central nervous system damage and the mother could develop preterm labor, infection, puerperal hemorrhage, consumptive coagulopathy, or obstruction of labor [8]. A pathogenetic hypothesis for the association between cloacal anomalies and twinning is that the partial or complete duplication of the organizing center within a single embryonic disc may increase the risk of mesodermal insufficiency accounting for failure of complete development of the cloacal membrane and extrophy [9]. During later stages of gastrulation, the caudal eminence functions as a developmental field that is modulated by homeobox genes and a variety of other factors. Siebert et al. [9] suggest that it is possible that the process of twin formation might disrupt the caudal eminence or its derivatives resulting in the defects seen in OEIS complex [3]. Etiology of either bladder extrophy or cloacal extrophy, it seems reasonable to accept that an event such as failure of cloacal septation or cloacal membrane breakdown during the fourth to sixth week of intrauterine life may have the potential to cause abnormalities in the adjacent lumbarosacral somites, depending upon the severity of the original abnormality in the adjacent lumbarosacral somites [10].

Conclusions

In a fetus with bladder agenesis and with hindgut abnormalities, like in our case, it is proposed that the division of cloaca is abnormal but there is either a primary developmental failure or secondary atrophy of the urogenital sinus or anal canal. Our case is unique, we have not encountered in the specialty literature so complex combination of fetal abnormalities. We are talking about a presence of amniotic bands in a twin pregnancy with a fetus papyraceus. We did not know what caused premature death of one of the twins but it may have been a mechanical cause by fixing omblical cord in an amniotic band. Analysis of our case indicates that amniotic band syndrome may not primarily be the consequence of amniotic or vascular disruption but rather the result of multifactorial processes responsible for the developmental malformations and fetal ecto-mesodermal disruption. The fetus that continued evolution up to 23 weeks with 3 weeks intrauterine growth restriction presented multiple malformations somewhat similar to the OEIS complex without neural tube defects. As we say, this case represents a very complex unique association of fetal malformations. We have not found any similar global communication. We can consider rather a case of a cloacal extrophy and maldevelopment at the same time with multiple malformations: absence of vagina and urethra, bladder agenesis with ectopic ureteric opening, anorectal agenesis, imperforate anus, omphalocele and supernumerary left adrenal gland. Medical management in such cases is generally very difficult.

Survival will depend on the extension of the cloacal extrophy and the genitourinary and intestinal defect. In less severe forms, good outcome with corrective surgery is possible. Cloacal extrophy is lethal due to obstruction of the urinary tract and association with renal and pulmonary complications. So early prenatal diagnosis is required to give parents the option to terminate the pregnancy. In cases where parents decided to continue the pregnancy, serial scans are necessary and it is also helpful to plan the appropriate perinatal management. The surgical management is typically undertaken in the postnatal period (48 to 72 hours) as a multidisciplinary approach involving neonatologists, pediatric surgeon, pediatric urologist, pediatric neurosurgeons, genetic, and pediatric endocrinologist. Treatment options in this disorder include continent and noncontinent urinary and digestive diversion, either internally or externally. Complex urinary reconstruction (creation of a continent urinary reservoir) may be undertaken later in life. The prognosis of infants with cloacal abnormalities is variable, depending on the severity of the structural defects.
REFERENCES