Pre- and Postnatal Features of Hydrometrocolpos in One Fetus of a Dizygotic Twin Pregnancy

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Abstract

A female fetus of a dizygotic pregnancy presented with a retrovesical cystic mass at 16 weeks. Severe and recurrent fetal ascites developed at 25 weeks. Pre-eclampsia, probably due to mirror syndrome, precipitated fetal extraction, which led to severe prematurity complications and neonatal death. Necropsy showed: cloacal anomaly, anal atresia, ambiguous genitalia and bicornuate uterus.

Keywords: Prenatal diagnosis, ultrasound, cloaca, hydrocolpos.

INTRODUCTION

Fetal hydrometrocolpos is one rare cause of fetal abdominal cystic mass, being the most frequent those of renal etiology, bowel dilatation or ovarian cysts in females. Topography, fetal sex, progression of the structure, and MRI (magnetic resonance imaging) can help diagnose the etiology and in the last few years the use of 3D (three-dimensional) ultrasound has become a widely employed tool in fetal malformations. The majority of cases reported were found in the third trimester or in the early postnatal period. The exact diagnosis is rarely performed before the late second trimester, thus preventing the parents decide on the fate of the pregnancy, where legally available.

The many etiologies of hydrometrocolpos constitute the main difficulty for differential diagnosis. We present a case of hydrometrocolpos associated to cloaca, anal atresia, ambiguous external genitalia and bicornuate uterus in one female fetus of a dizygotic twin pregnancy achieved by IVF (in vitro fertilization). Diagnostic approach involved 3D ultrasound, paracentesis and MRI. Prognosis of a persistent cloaca is worse than isolated hydrocolpos or urogenital sinus. Although cloacal syndrome it is not always lethal, in this case severe preeclampsia developed in the mother and the fetuses had to be delivered and died due to prematurity complications.

CASE REPORT

A 36 year-old caucasian woman G2A1 with a dizygotic twin pregnancy conceived by IVF underwent second trimester ultrasound examination at 16 weeks that revealed an abdominal malformation in one of the two female fetuses. The family history was noncontributive and parents were nonconsanguineous. An earlier scan at 12 weeks was normal as was the combined screening test. The ultrasound scan demonstrated a 20 × 17 mm tubular hypoechogenic mass behind the bladder (Figs 1 to 3).

Hydrometrocolpos syndrome was suspected. Fetal biometries were within the normal range and amniocentesis revealed normal karyotypes. Fetal echocardiography was normal. Maternal and amniotic liquid serological tests were negative. Once informed the parents decided against selective termination of pregnancy due to the risk of complications for the other twin.

The image remained unchanged until 25 weeks when, severe and nonassociated to polyhydramnios ascitis was demonstrated, that jeopardized the fetuses’ lives (Fig. 4). The malformed fetus underwent transabdominal ultrasound-guided paracentesis and 200 ml of serous liquid was obtained. Microbiological study of the fluid was negative, biochemical assay was noncontributive and cytology
Demonstrated squamous cells suggestive of genital origin. Ascitis recurred within 24 hours. Aspiration of the fetal intracystic fluid was technically unfeasible.

MRI confirmed a 26 × 13 mm cystic lesion behind the urinary bladder and showed a nondilated intestine (Fig. 5). The other fetus had no apparent malformations. The patient developed severe preeclampsia at 27 weeks (probably associated with maternal mirror syndrome) and a cesarean section was performed. A 970 gm female infant was delivered with no malformations but died within 20 days due to severe prematurity complications. The second twin weighted 1110 gm and showed a right abdominal distention caused by a cystic mass, as well as collateral venous circulation, ambiguous genitalia, and anal atresia with a single perineal opening (Fig. 6). Upon postnatal ultrasound examination both kidneys were normal, urinary bladder could not be seen and small intestine was dilated. Two days after birth the infant underwent surgery that identified one purulent fluid filled cavity and colostomy was performed. The infant died eight days later due to septic complications. Necropsy demonstrated anal atresia, cloacal structure that communicated bladder, rectum and vagina and a bicornuate uterus.

DISCUSSION

hydrocolpos is caused by obstruction to vaginal fluid drainage; among the causes described are McKusick-Kaufman syndrome, vaginal or external genitalia malformation such as labial fusion, cloacal anomaly, imperforated hymen and persistent urogenital sinus. Cases of persistent urogenital sinus or cloacal syndromes (depending on the moment of arrest of embryonic development) like the present one are sporadic while McKusick-Kaufmann syndrome is autosomal recessive and have cardiac defects and polydactyly. The fluid that accumulates is not only of genital origin, as intestinal and urinary fistulae are often associated.

A few dozen cases of hydrometrocolpos have been described before in literature. Dhombres et al (2007) reported a persistent urogenital sinus in a dizygotic pregnancy, whereas in our case the cloacal anomaly developed in one twin of a dizygotic IVF pregnancy and consisted on the above mentioned malformations (cloacal structure with bicornuate uterus), while heart defect, renal obstruction, or polydactyly were ruled out prenatally. Didelphys or bicornuate uterus are not uncommon associations to hydrocolpos demonstrating the initial müllerian ducts abnormal development, although prenatal ultrasound can fail to reveal this particularity of the uterus. If fluid fails to accumulate, isolated hydrocolpos or hydrometrocolpos is not usually seen until puberty, and the few cases described prenatally are caused by maternal hormonal influence. Most published cases were diagnosed in the third trimester and included hydronephrosis caused by urinary tract obstruction. In the present case the mass...
Fig. 3: Three-dimensional niche reconstruction of fetal hydrometrocolpos at 24 weeks pregnancy

Fig. 4: Two-dimensional ultrasound scan showing fetal ascitis at 26 weeks pregnancy

Fig. 5: Magnetic resonance imaging showing cystic lesion and non-dilated intestine at 24 weeks pregnancy
was patent since the 16 weeks of pregnancy and consisted of a cloacal structure where, the bladder, rectum and bicornuate uterus converged but did not obstruct the renal pelvis. We believe that hydronephrosis would have appeared had ascitis not developed before. The cyst was demonstrated on a routine ultrasound scan as fluid accumulated in the cloacal structure, probably due to urine accumulation. Ascitis was not due to rupture of the uterus but we assumed it was caused by tubal drainage to the peritoneum as described before.22

Cloacal anomalies in female fetuses have an incidence of 6:100,000 and are not necessarily associated to mental retardation or to other malformative stigmata although molecular diagnosis is recommended to rule out syndromic causes of hydrocolpos such as Langer-Gideion23 in cases where, extragenital malformations are present. Postnatal therapy of isolated hydrometrocolpos consists on drainage and reconstructive surgery but in the present case severe prematurity complications lead to neonatal death.

Ultrasound findings are those of an abdominal elongated fluid filled mass that in the present case showed thick walls, as would not a megacystis. Differential diagnosis with intestinal anomalies required the use of three-dimensional ultrasound where, the thick wall of the uterus is shown. Ovarian cyst was ruled out, and the use of MRI allowed identification of intestinal surrounding structures.

Necropsy established the definite diagnosis of the cloacal syndrome.

What this paper adds are the pre- and postnatal imaging features of the syndrome as well as the early report of a cloacal syndrome in a pregnancy achieved by assisted reproduction techniques and the use of three-dimensional ultrasound to assess the etiology of the cyst.

REFERENCES