Prenatal diagnosis of hydrometrocolpos in a Down syndrome fetus

Erik Dosedla, Marian Kacerovsky, Pavel Calda

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Short title: Hydrometrocolpos and Down syndrome

Abstract

Prenatal diagnosis of hydrometrocolpos in a Down syndrome fetus caused by an imperforate hymen, with spontaneous evacuation on the third day of life, is reported. In this case, sonographic evaluation in the 37th week revealed a large retrovesical sharply marginated, heterogeneous, unilocular cystic structure within the fetal abdomen extending to the left side of the umbilicus.

Introduction

Hydrometrocolpos is defined as dilatation of the vagina and uterus, either with mucous content generated by uterine and cervical glands, mediated by maternal estrogen production (secretory hydrometrocolpos or mucometrocolpos), or by accumulation of urine in the presence of a vaginal obstruction (urinary hydrometrocolpos)[1]. Hydrometrocolpos is a rare congenital abnormality, with an incidence of 1:16,000 neonates[2-3]. It is most commonly discovered in the third trimester of pregnancy[3-5] and makes up 15% of abdominal masses found in female fetuses. Generally, reproductive outcomes in chromosomally normal fetuses after drainage of the mucous collection are reported to be good[6].

Case report

In October 2009, a 39-year-old woman, gravida 5, para 3, was referred to the department at 37 weeks, 1 day of pregnancy because of a pathological cardiotocography examination and suspicion of fetal growth restriction.

The mother's family history was insignificant. She did not take any drugs from conception through the first three months of pregnancy, but she smoked more than 20 cigarettes while pregnant. Exposure to other teratogens was negative.

First-trimester aneuploidy screening was not performed, but the ultrasound examination in the 14th week was reported to be normal. Serologies for toxoplasmosis, rubella, cytomegalovirus, herpes simplex, HIV, hepatitis B surface antigen, and treponema pallidum hemagglutination (TPHA) were negative. The triple test (AFP, hCG, and Estriol) performed at 16 weeks was positive with a risk for trisomy 21 of 1:16. The mother was counseled on the risk of a chromosomal abnormality, but refused invasive testing (amniocentesis). Morphology ultrasound scan at 20 weeks of pregnancy was recommended, but was not accepted by the pregnant woman. Ultrasound biometry and morphology examination of the fetus performed in the 30th week were normal.

Upon the patient's admission at 37 weeks, the ultrasound examination revealed a living female singleton fetus with a composite sonographic age of 33 weeks, 4 days (intrauterine growth restriction of over 4 weeks), anhydramnios, and pathological blood flow in the umbilical artery (blood flow class II). A large retrovesical sharply marginated, heterogeneous, unilocular cystic structure measuring 5.3 × 4.7 × 4.3 cm within the fetal abdomen extending to the umbilical insertion was described. The anal canal and the rectum were clearly visualized. There was no anomaly of the uropoetic system. Three-dimensional datasets were also obtained, which were reviewed with tomographic ultrasound imaging

(TUI) (Figures 1, 2). No other anomalies were found. The diagnosis of fetal hydrometrocolpos was made based on sonographic findings. All scans were carried out with an ultrasound system Voluson 730 Expert (GE Medical Systems, Milwaukee, WI, USA) equipped with a convex 4-8 MHz abdominal transducer (RAB 4-8L).

Due to nonreassuring fetal status (biophysical profile 3/10), a female neonate of 2550g with Apgar score of 6/9/10 was delivered the same day by cesarean section. The neonatal examination revealed typical signs of Down syndrome and a mass in the lower and middle abdomen, and a relatively broad hymen. The diagnosis of hydrometrocolpos was confirmed. Hydrometrocolpos resolved spontaneously on the third day after delivery when the attending physician observed a spontaneous discharge of mucous fluid. The abdominopelvic fluid-filled cystic mass was not seen on a subsequent ultrasound examination. The final diagnosis was hydrometrocolpos caused by an imperforate hymen, with spontaneous evacuation on the third day of life. The newborn karyotype revealed trisomy 21 (Down syndrome).

Discussion

Every pelvic cystic mass in a female fetus can enter into differential diagnosis of hydrometrocolpos. Hydrometrocolpos is present as retrovesical unilocular cystic oval or funnel-like mass with low echogenicity, which is dependent on the intracystic content. It is associated with many malformations like Klippel-Feil anomaly, McKusick-Kaufman syndrome, Ellis van Creveld syndrome, and Bardet-Biedl syndrome. Usually, ovarian cysts are rare unilateral hypoechogenic cystic masses. Urachal cysts are unilocular cysts located between urinary bladder and umbilicus. The urinary tract obstruction appears in most cases as an anechogenic dilatated megaurether, urinoma, or megavesica. Anterior sacral meningocele is a very rare anechogenic cystic mass extending from the sacrum to the urinary bladder. Sacrococcygeal teratoma arises in the presacral areas

and is the most common tumor of the newborn period. Type IV is completely internal with no external component. Ultrasound appearance of the teratomas is very heterogeneous – from solid to cystic, with high vascularity.

In hydrometrocolpos, mucous content accumulates due to imperforate hymen or failure of Müllerian duct fusion[4,7]. Imperforate hymen is a rare genital anomaly in which a layer of epithelized connective tissue, which forms the hymen, has no opening and completely obstructs the vaginal introitus[8]. Prenatal ultrasound diagnosis of hydrometrocolpos is rare[9]. In the present study, the ultrasound appearance of the hydrometrocolpos was nearly solid because of the mucous content of the mass, and therefore it would be more correct to use term *mucometrocolpos*. When hydrometrocolpos is suspected, it is recommended that a detailed ultrasound examination be performed to rule out associated malformations. The observation revealed that the first ultrasound sign of hydrometrocolpos in a female fetus is abdominal distension. It is important to examine the pelvis in all three orthogonal planes. Therefore, TUI was used, which has advantages in studying complex fetal lesions[10-12]. A perspective of the topographic anatomy of the fetal abdomen and informative images were obtained with TUI. The four-dimensional visualization allows better understanding of the anatomic conditions and helps during the process of prenatal parental counseling.

As reported earlier, the rectum and the anal canal were visualized and the kidneys were intact, so complete cloacal dysgenesis as well as persistent urogenital sinus could be ruled out. Postnatal confirmation of the hydrometrocolpos caused by imperforated hymen is based on the presence of a lower abdominal mass in a female infant that does not disappear after urinary bladder cathethrisation[13]. Hydrometrocolpos associated with 47, XXX karyotype and 45, XO karyotype has been described in two cases[14-15]. In

the case of sonographically detected hydrometrocolpos, karyotypization of the fetus is recommended, but invasive testing was rejected by the patient.

Reference

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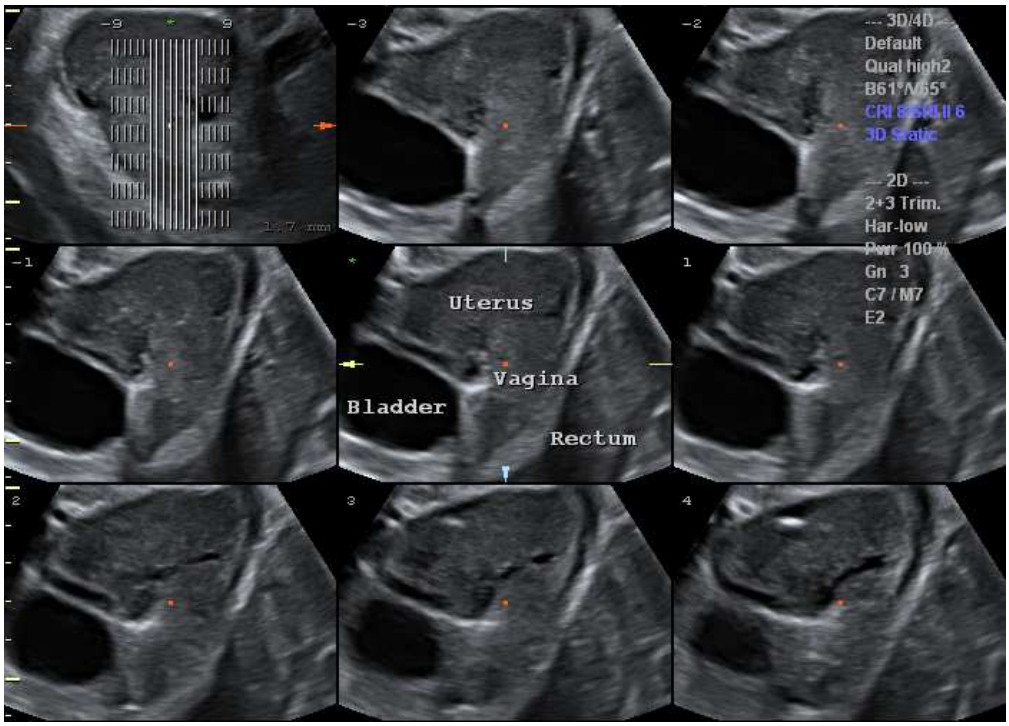

Figure 1. Tomographic ultrasound imaging of fetal pelvis shows a set of sagittal slices which demonstrate a large retrovesical hydrometrocolpos.
246x177mm (72 x 72 DPI)

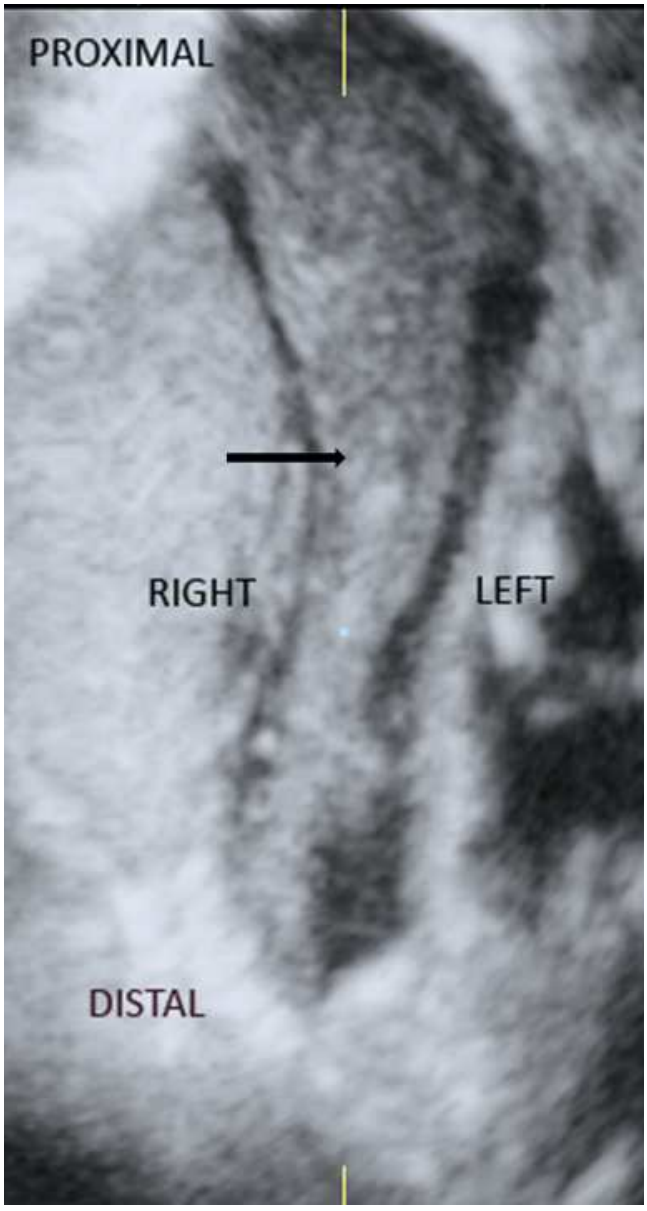

Figure 2. Coronal static 3D image combined with volume contrast imaging in C-Plane (VCI-C improves tissue contrast resolution in real-time, coronal plane imaging) shows a pelvic cystic mass (hydrometrocolpos) marked with arrow.
101x190mm (96 x 96 DPI)

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Prenatal diagnosis of hydrometrocolpos in a Down syndrome fetus caused by an imperforate hymen, with spontaneous evacuation on the third day of life, is reported. In this case, sonographic evaluation in the 37th week revealed a large retrovesical sharply marginated, heterogeneous, unilocular cystic structure within the fetal abdomen extending to the left side of the umbilicus.

Deleted: We used four-dimensional ultrasound tomographic imaging in studying the complex fetal lesion. To date, there is no evidence of association between hydrometrocolpos and Down syndrome.

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Deleted: The differential diagnostic features of hydrometrocolpos are summarized in Table 1.

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