FETAL HYDROMETROCOLPOS, UTERUS DIDELPHYS WITH LOW VAGINAL AND ANAL ATRESIA: DIFFICULTIES IN DIFFERENTIATION FROM A COMPLEX CLOACAL MALFORMATION: A CASE REPORT

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Summary: *Fetal hydrometrocolpos, uterus didelphys with low vaginal and anal atresia: difficulties in differentiation from a complex cloacal malformation: a case report:* Hydrometrocolpos, occurring in approximately 1/6000 newborn girls, can be caused by a stenotic urogenital sinus, a severe cloacal malformation, but also by other conditions such as an imperforate hymen, a midline vaginal septum and vaginal atresia. The prenatal differential diagnosis of this wide spectrum of conditions is not easy and requires a multidisciplinary approach with follow-up scans and MRI to access the severity of the condition.

A non-consanguineous couple was referred in the first pregnancy at 30 weeks. The father, 30 years of age, of Kaukasian origin, and the mother of Asian origin, 26 years of age. Ultrasound at 30 weeks revealed ambiguous genitalia (with suspicion of clitoral hypertrophy), a septated structure located behind the bladder compatible with hydrometrocolpos with a uterine malformation (uterus didelphys), a single umbilical artery, mild ascites and growth on the tenth centile. The differential diagnosis included a vaginal atresia, a urogenital sinus and a more severe cloacal malformation. After serial scans, MRI and counselling by an experienced surgeon the preferential diagnosis of a cloacal malformation was made and a late pregnancy termination was performed. Pathological examination revealed :low vaginal atresia with uterus didelphys, anal atresia with rectovaginal fistula and a normal urinary tractus.

The differential diagnosis between hydrometrocolpos due to vaginal atresia or due to a more severe cloacal malformation is not straightforward. Care should be taken in decision making and counselling patients with these complex prenatal malformations.

Key-words: Multiple congenital anomalies – Fetal malformation – Anogenital anomalies.

INTRODUCTION

This report describes a third trimester diagnosis of hydrometrocolpos due to a low vaginal atresia with anal atresia and the difficulty in differentiation from a complex cloacal dysgenesis.

CASE REPORT

The couple, without medical history, was referred at 30 weeks gestation by the local gynecologist with the ultrasound diagnosis of a single

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umbilical artery in their first pregnancy. Follow-up examinations in the first and second trimesters of pregnancy occurred abroad.
Ultrasound at 30 weeks revealed ambiguous genitalia (with suspicion of clitoral hypertrophy), a septated structure located behind the bladder compatible with a uterine malformation, a single umbilical artery, mild ascites and growth on the tenth centile (Fig. 1).
To differentiate between a vaginal atresia, a urogenital sinus malformation and a more severe cloacal malformation additionally an MRI scan was performed confirming the septated structure posteriorly from the bladder suspicious of a hydrometrocolpos, the ascites likely due to intraabdominal spil, meconium filling of the rectum but possible anorectal atresia and the other ultrasonografically described malformations (Figs 2-3).
After referral to a tertiary center the likely diagnosis of a cloacal dysgenesis was made and the couple was councelled abroad by an experienced surgeon in the field of cloacal anomalies. Due to the infaust prognosis given by the surgeon the couple had a third trimester pregnancy termination in another center.
Pathological examination revealed : a female fetus (weight 1900 g, length 43 cm, head circumference 30 cm) with a single umbilical artery, a low vaginal atresia with doubled vagina, uterus didelphys and secondary hydrometrocolpos and due to the pressure of the hydrometrocolpos a mild hydronephrosis with otherwise normal urinary tract. Additionally an anal atresia with rectovaginal fistula was present.

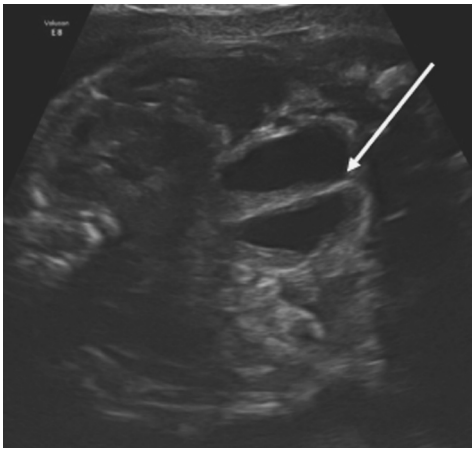

Figure 1: ultrasound at 30 weeks reveals a septated structure posteriorly from the bladder (arrow), ascites

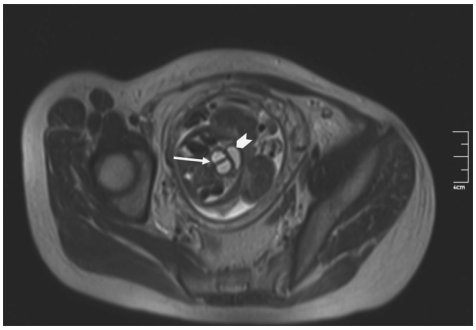

Figure 2: Axial T2 weighted image of the fetus at the level of the pelvis shows the bilocular fluid containing structure (uterus didelphus – arrow) which is located posterior from the bladder (arrowhead).

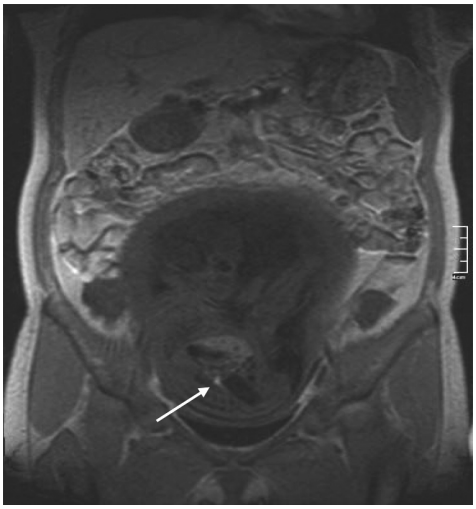

Figure 3: Sagittal T1 weighted image of the fetus reveals T1 hyperintens signql in the rectosigmoid (arrow) at the level of the dorsocranial pole of the structure without continuation towards the anus. This is highly suggestive for recto-anal hypoplasia.

DISCUSSION

A cloacal malformation is not common with an estimated incidence of 1/50000 births.

Typically in cloacal malformation there is a common opening for the urinary, genital and gastrointestinal tract. Normally in embryogenesis this common opening is present at 5 weeks gestation and at six weeks the cloaca is divided by the growth of the urorectal septum giving rise to the urogenital sinus anteriorly and the hindgut posteriorly.

The urogenital and anal membranes split by week 10 to produce a patent urogenital and rectal tract (5).

The close relationship between the sinus urogenitalis and the anorec-

tal canal explain the frequently seen association of hydrometrocolpos with imperforate anus (Fig. 2).

The prenatal ultrasound appearance of cloacal malformation can vary depending on the type of malformation and typically shows transient fetal ascites, a bilobed cystic structure arising from the fetal pelvis which may contain debris, a poorly visualized fetal bladder, bilateral hydronephrosis, oligohydramnios, growth retardation and vertebral anomalies in a female fetus (5).

A cloacal malformation can result in urine drainage into the vagina (common canal) with secondary hydrocolpos and pressure on the urinary tracts with oligohydramnios and impaired kidney function. Urine drains through the fallopian tubes giving ascites and the chronic irritation ultimately can obstruct the tubes (3, 5).

A cloacal malformation can cause hydrometrocolpos but other possible causes of dilatation of the vagina (and uterus) can be a stenotic urogenital sinus, an imperforate hymen, a midline vaginal septum or vaginal atresia (3).

In stenotic urogenital sinus and cloaca, the fluid accumulating in the vagina is urine, while in vaginal atresia the fluid is the result of cervical and vaginal secretions produced under influence of maternal estrogens.

Hydro(metro)colpos occurs in approximately 1/ 16000 newborn girls.

It's important to try to differentiate prenatally between hydrocolpos with good prognosis as seen in imperforate hymen and those with a worse prognosis as seen in cloacal malformation.

With ultrasound a simple not-septated cystic structure behind the bladder is more suggestive of hydrocolpos due to imperforate hymen, while a septated vagina (uterus) may present a uterus didelphys but also cloacal /rectal anomalies or syndromic diseases (hydrocolpos can be part of Mc Kusick Kaufman syndrome, Langer Giedon syndrome and Vacterl association) (1-4).

Digestive malformations can be suspected by ultrasound in the presence of a dilated colon, atypical anal appearance or calcifications in the intestinal lumen as the result of mixing of meconium and urine as seen in cloacal malformations (4).

MRI can be helpful to evaluate the digestive tract for meconium filling (dark on T2-weighted signals and bright on T1-weighted signals): failure to visualize meconium is suggestive of anorectal atresia or of cloacal malformation.

The present case with a hydrometrocolpos due to low vaginal atresia, with double vagina and uterus didelphys, secondary hydronephrosis and anal atresia did not match with a full cloacal malformation.

Although images (ultrasound, MRI) showed a septated structure be-

hind the bladder, ascites, no continuation of the anorectal canal and hydronephrosis in a female fetus, the fetal bladder appeared normal, meconiumfilling was present and the postmortem examination revealed a normal urinary tractus and no persistent cloaca.
Therefore, care should be taken in making decisions on pregnancy termination in late pregnancy and in counseling patients with complex malformations.

CONCLUSION

This report describes a third trimester diagnosis of hydrometrocolpos due to a low vaginal atresia with anal atresia and the difficulty in differentiation from a complex cloacal dysgenesis.
Care should be taken in decision making for these couples, implying a multidisciplinary approach, serial scans to follow-up the evolution of the malformation, no immediate decisions on pregnancy termination and in case of termination pathology to compare if the suspected prenatal diagnosis was finally correct

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