Prenatal diagnosis of McKusick–Kaufman/Bardet–Biedl syndrome

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Summary

A 24-year-old primigravid woman with no remarkable history and antenatal follow-up was referred to our clinic with the diagnosis of fetal megacystis. Ultrasound examination revealed an oval shaped hypoechoic cystic mass measuring $80 \times 55 \times 50$ mm occupying the fetal pelvis in a female fetus. The fluid inside the cyst was heterogeneous giving a two level appearance. A normal looking urinary bladder was visualised separately which was located in normal position. Further sonographic evaluation revealed postaxial polydactyly in both hands. The fetal echocardiography and amniotic fluid index was normal. An obstetrical MRI was done for the suspicion of a hydrometrocolpos and confirmed the diagnosis. The infant was delivered by vaginal route at term. Physical examination of neonate confirmed postaxial polydactyly in hands. After birth a cystoscopy performed by the paediatric surgeons revealed a urogenital sinus and vaginal atresia. A catheter was placed in the uterine cavity to drain it. Nine months later the baby went on a 'vaginal pull through' operation to reconstruct the vagina and the urethra. The baby is now 3 years old and doing well.

BACKGROUND

This is a rare anomaly. Hydrometrocolpos should be differentiated from other pelvic masses such as ovarian cysts, because while the former can be associated with obstructive lesions such as urogenital sinus, vaginal atresia and syndromes associated with mental retardation, the prognosis for latter is excellent.

CASE PRESENTATION

A 24-year-old primigravid woman was referred to our clinic for the evaluation of fetal pelvic cystic mass which was initially thought to be megacystis by the referring physician. Medical and obstetrical history of the patient was unremarkable for teratogenic exposure, congenital infection or parental consanguinity. The patient had an uneventful antenatal follow-up until then. Nuchal translucency was normal and triple test results were low risk at the local clinic. At admission, the patient was 30 weeks and 4 days pregnant according to last menstrual period. Ultrasound examination revealed a singleton female fetus concordant with the gestational age. A hypoechoic cystic mass measuring $80 \times 55 \times 50$ mm in maximal diameter occupying the fetal pelvis was detected (figure 1). The fluid inside the cyst was heterogeneous giving a two level appearance. The normal looking urinary bladder was visualised separately which was located in normal position. Bilateral kidneys were visible and were normal in size and position.

Fetal echocardiography was normal. Further sonographic evaluation revealed postaxial polydactyly in both hands (figure 2). No other concomitant fetal anomaly was detected. Amniotic fluid index was normal. At 39 weeks of gestation, a 3400 g female infant was delivered by vaginal route. Physical examination of neonate revealed

postaxial polydactyly in both hands as well as vaginal atresia.

INVESTIGATIONS

Because the fetus was female, there was a suspicion of an ovarian cyst. But the pelvic mass looked more elliptical than round and fluid inside it gave a two level appearance. An obstetrical MRI was planned for the suspicion of a hydrometrocolpos. MRI confirmed the diagnosis of hydrometrocolpos (figure 3).

DIFFERENTIAL DIAGNOSIS

Hydrometrocolpos and postaxial polydactyly are common to both McKusick–Kaufman and Bardet–Biedl syndromes. However a differentiation of these two syndromes is not possible in the neonatal period. Retinitis pigmentosa, obesity and mental retardation in Bardet–Biedl syndrome does not develop until children are 5 years old. So children initially diagnosed with McKusick–Kaufman syndrome should be followed up. Both syndromes are transmitted by an autosomal recessive pattern, and parents should be counselled accordingly.¹

TREATMENT

After birth, a cystoscopy was performed by the pediatric surgeons. The diagnosis was a urogenital sinus and vaginal atresia. A catheter was placed in the uterine cavity to drain it. Nine months later the baby went on a 'vaginal pull through' operation to reconstruct the vagina and the urethra.

OUTCOME AND FOLLOW-UP

The baby is now 3 years old and doing well.

Figure 1 Fetal hydrometrocolpos – ultrasound image.

Figure 2 Postaxial polydactyly.

DISCUSSION

In 1964 McKusick *et al*² reported that postaxial hexadactyly, hydrometrocolpos and congenital heart disease are a part of a syndrome transmitted autosomal recessively. Later Dungy *et al*⁶ found hydrometrocolpos secondary to vaginal atresia and bilateral postaxial hexadactyly in

an offspring of first-cousin parents. Kaufman *et al*⁴ suggested that postaxial polydactyly and/or congenital heart disease may sometimes accompany hydrometrocolpos. Hydrometrocolpos is the congenital cystic dilatation of the vagina and uterus due to congenital distal vaginal obstruction. A variety of urogenital malformations may

Figure 3 Fetal hydrometrocolpos – MRI image.

lead to vaginal obstruction. These malformations include imperforate hymen, vaginal agenesis, vaginal hypoplasia or transverse septum. The pathophysiologic mechanism of hydrometrocolpos is the accumulation of secretions from fetal cervical and endometrial glands under maternal oestrogen stimulation.⁵ Rarely, a vesicovaginal fistula and accumulation of the urine can be cause of hydrometrocolpos.⁶ Congenital hydrometrocolpos accounts for 15% of intra-abdominal cystic masses in female infants. It has also been associated with urinary or intestinal tract abnormalities.⁷

Clinical spectrums of affected fetuses vary depending on the mass effect of hydrometrocolpos and presence of associated abnormalities. In some cases, hydrometrocolpos has been reported to cause postrenal obstruction of

urinary outflow. Moreover fetal ascites and intra-abdominal calcifications have been reported in fetuses with hydrometrocolpos.⁸

Prenatal detection of hydrometrocolpos necessitates detailed fetal sonographic examination to identify the presence and extent of associated abnormalities. Ultrasonographic evaluation of affected fetuses may reveal stigmata suggesting presence of underlying genetic disorders. Ultrasonography is the gold standard for examination of the fetus and detecting morphologic/structural fetal anomalies. MRI may also be helpful in diagnosis.

In conclusion fetal hydrometrocolpos is a significant finding necessitating detailed sonographic examination due to variety of underlying genetic diseases.

Learning points

- ▶ In female fetuses with a pelvic cyst, determining the origin of the cyst is important since hydrometrocolpos may be associated with congenital syndromes.
- ▶ Whenever a pelvic mass is detected, it is important to examine the other systems carefully in order not to miss any other anomaly which may suggest a congenital syndrome.
- ▶ It is important not to diagnose the baby immediately because Bardet–Biedl syndrome may be differentiated from McKusick–Kaufman syndrome only after 5 years.

Competing interests None.

Patient consent Obtained.

REFERENCES

1. **Slavotinek AM**, Biesecker LG. Phenotypic overlap of McKusick–Kaufman syndrome with bardet–biedl syndrome: a literature review. *Am J Med Genet* 2000;**95**:208–15.
2. **McKusick VA**, Bauer RL, Koop CE, *et al*. Hydrometrocolpos as a simply inherited malformation. *JAMA* 1964;**189**:813–16.
3. **Dungy CI**, Aptekar RG, Cann HM. Hereditary hydrometrocolpos with polydactyly in infancy. *Pediatrics* 1971;**47**:138–41.
4. **Kaufman RL**, Hartmann AF, McAlister WH. Family studies in congenital heart disease, II: A syndrome of hydrometrocolpos, postaxial polydactyly and congenital heart disease. *Birth Defects Orig Art Ser* 1972;**VIII**:85–7.
5. **Reed MH**, Griscom NT. Hydrometrocolpos in infancy. *Am J Roentgenol Radium Ther Nucl Med* 1973;**118**:1–13.
6. **Mahoney PJ**, Chamberlain JW. Hydrometrocolpos in infancy: congenital atresia of the vagina with abnormally abundant cervical secretions. *J Pediatr* 1940;**17**:772.
7. **Ekenze SO**, Ezegwui HU. Hydrometrocolpos from a low vaginal atresia: an uncommon cause of neonatal intestinal and urinary obstruction. *Afr J Paediatr Surg* 2008;**5**:43–5.
8. **Gul A**, Yildirim G, Gedikbasi A, *et al*. Prenatal ultrasonographic features of persistent urogenital sinus with hydrometrocolpos and ascites. *Arch Gynecol Obstet* 2008;**278**:493–6.

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