CASE REPORT

Fetal ascites and hydrometrocolpos due to persistent urogenital sinus and cloaca: a rare congenital anomaly and review of literature

Aruna Nigam,¹ Manisha Kumar,² Shilpa Gulati²

¹Department of Obstetrics and Gynaecology, Hamdard Institute of Medical Sciences and Research, New Delhi, Delhi, India

²Department of Obstetrics and Gynaecology, Lady Hardinge Medical College, New Delhi, Delhi, India

Correspondence to

Dr Aruna Nigam,
prakasharuna@hotmail.com

Accepted 24 January 2014

SUMMARY

Fetal ascites can occur due to many heterogeneous disorders. Its association with hydrometrocolpos because of persistent urogenital sinus and cloaca is extremely rare. A 29-year-old primigravida presented at 32 weeks of gestation with ultrasonographic evidence of fetal ascites, a cystic pelvic mass, hydronephrosis and oligohydramnios. Fetal ascites in this case was due to fetal urine draining through fallopian tubes into the abdomen as a result of vesicovaginal fistula and distal vaginal atresia. The antenatal ultrasound results along with autopsy findings are discussed. Though rare, a persistent urogenital sinus is to be suspected in isolated fetal ascites cases where the viral tests are negative and there is no evidence of cardiac anomalies as this is a treatable anomaly if diagnosed at early gestational age.

BACKGROUND

Fetal ascites is generally caused by immune and non-immune hydrops fetalis, congenital infections, cardiac malformations, gastrointestinal and genitourinary anomalies.¹ Here, we present a case of fetal ascites with hydrometrocolpos secondary to persistent urogenital sinus and cloaca. The early prenatal diagnosis gives a good prognosis for this condition. Serial ultrasound, with appropriate viral screen and exclusion of chromosomal abnormalities, is essential to make a diagnosis.

CASE PRESENTATION

A 29-year-old primigravida referred to our institute at 32 weeks of gestation with evidence of isolated fetal ascites on ultrasound. Her general physical and systemic examination was unremarkable. An

abdominal examination revealed 28 weeks size uterus, with fetus in longitudinal lie, cephalic presentation and regular fetal heart sound. Amniotic fluid was reduced.

INVESTIGATIONS

A detailed ultrasound examination revealed gross fetal ascites and a bilobed cystic structure in the lower abdomen in the presacral region ([figure 1](#)). There was severe oligohydramnios; one kidney showed hydronephrosis ([figure 2](#)) and other could not be visualised. Fetal thoracic volume was reduced and mostly occupied by heart, suggestive of pulmonary hypoplasia. The spine was apparently normal and no malformations were found in other organs, including the heart. Her blood group was B positive; indirect Coomb's test and TORCH serology were negative.

DIAGNOSIS

A diagnosis of persistent urogenital sinus with hydrometrocolpos and fetal ascites with pulmonary hypoplasia and severe oligohydramnios was made.

OUTCOME AND FOLLOW-UP

During the course of investigations, fetal death occurred. The patient went into spontaneous labour on the following day and delivered a 1.8 kg fetus.

Fetal autopsy

External examination of the fetus ([figure 3](#)) revealed enlarged abdomen, low set ears, short limbs and congenital talipes varus. External genitalia were ambiguous ([figure 4](#)). No external opening in the

Figure 1 Antenatal ultrasound of fetus showing bilobed cystic structure in the lower abdomen with ascites.

Figure 2 Antenatal ultrasound of fetus showing bilobed cystic structure in the lower abdomen with dilated renal calyces.

To cite: Nigam A, Kumar M, Gulati S. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2013-202231

Figure 3 External appearance of fetus showing enlarged abdomen, short limbs and congenital talipes varus.

perineum was visualised. Fetal autopsy revealed ascites with enlarged bladder, dilated uterus and vagina, fallopian tube filled with fluid and enlarged large bowel and rectum (figure 5). One of the kidneys was enlarged and the other was dysplastic and small. A communication was appreciated on passing the infant feeding tube from the bladder which came out from the vagina (figure 6), suggestive of vesicovaginal fistula.

DISCUSSION

Cloacal anomalies are rare malformations with a variable antenatal sonographic appearance which are very difficult to diagnose antenatally. Persistent urogenital sinus with persistent cloaca is an extremely rare and variable urogenital malformation. It is secondary to failure of urethrovaginal division at 6 weeks of gestation, resulting in confluence of vagina and urethra proximal to the introitus.²⁻⁴ The outflow obstruction leads to flow of the urine into the vagina which further results into hydrometrocolpos (cystic dilation of the vagina and the uterus due to fluid accumulation), and is seen as anechoic or hypoechoic retrovesical mass in ultrasound or bilobed cystic structure in the lower abdomen, as in this case. Very rarely, because of the outflow obstruction, the efflux of urine through the fallopian tubes in the abdominal cavity leads to fetal ascites, as is in this case. Hydrocolpos and hydrometrocolpos with persistent urogenital sinus can be associated with a wide spectrum of other urogenital and ano-rectal malformations, such as imperforate hymen, vaginal atresia, transverse vaginal septum and cloacal anomaly. Vaginal atresia and cloacal anomaly were present in our case. In utero urinary hydrometrocolpos can be

Figure 4 Enlarged clitoris and labia with no perineal opening.

Figure 5 Fetal autopsy showing enlarged uterus and tubes with ascites.

responsible for dilation of upper urinary tract, renal dysplasia and oligohydramnios, which is a bad prognostic sign.⁵ Large fetal ascites can also lead to diaphragmatic compression, pulmonary hypoplasia and fetal death, as in our case.

Most of the case reports with successful outcome had two or three perineal openings in contrast to our case which had no external opening in the perineum, which point towards the persistent cloaca associated with confluence of vagina, urethra and rectum, as reported by Adam *et al.*⁶

Differential diagnosis of cystic structure in fetal abdomen includes abdominal cyst masses, such as ovarian cyst, mesenteric cyst, urachal cyst, duplicated urinary bladder and other conditions such as mega cystitis-microcolon intestinal hypoperistalsis syndrome, obstructive uropathy and anterior sacral teratoma type IV.^{4,6} Hydrometrocolpos is also a feature of some genetic syndromes, that is, Bardet-Biedl, McKusick-Kaufman and Pallister-Hall syndromes.⁷

Thus, a systematic protocol should be followed to find the underlying cause, as an aetiology can be identified in 92% of the cases.^{8,9} As fetal ascites is an important associate of fetal hydrops, it is important to determine whether it is isolated or whether other signs of hydrops are present and investigations like Coomb's test and TORCH profile should be conducted. The case reported here had hydronephrosis and ascites. Fetal echocardiography is necessary to exclude cardiac disorders that could be the cause of ascites.

The existence of clitoral hypertrophy in this case indicates the possibility of associated congenital adrenal hyperplasia which cannot be established in our case as the fetus died during the

Figure 6 Demonstration of infant feeding tube going into the cut dome of the bladder and coming out through vaginal incision.

course of investigations although autopsy did not reveal any adrenal gland enlargement.

Associated pulmonary hypoplasia, along with unilateral renal hypoplasia, is an additional finding which has not been reported in other similar cases on review of literature.

Persistent urogenital sinus with hydrometrocolpos is a treatable malformation but the fetal survival depends on the other associated anomalies and the time of prenatal diagnosis. An accurate prenatal diagnosis of urogenital and cloacal malformations is difficult to make due to their rarity and variability of presentation. This rare anomaly can be seen in utero at 20–24 weeks of gestation.⁶ Prenatal evaluation by MRI can be used to know the type of malformations. An early evaluation and vaginal and urinary system decompression is recommended to avoid later complications. Chen *et al* suggested prenatal drain-

age of the hydrometrocolpos by ultrasound-guided aspiration to alleviate the abdominal distension and severe urinary tract obstruction.¹⁰ Postnatally intermittent vaginal catheterisation, antibiotic prophylaxis and surgical treatment after 1 or 6 months are recommended by different authors.

The successful outcome of this anomaly depends on correct and early prenatal diagnosis, absence of renal dysplasia or neonatal cardiorespiratory distress and teamwork between different specialists, including obstetricians, neonatologists and paediatric surgeons. Appropriate parental counselling is also an integral part as the prognosis is not well known.

Contributors All the authors contributed significantly in the manuscript.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

Learning points

- Fetal ascites is generally caused by immune and non-immune hydrops fetalis congenital infections, cardiac malformations and gastrointestinal and genitourinary anomalies.
- Persistent urogenital sinus with hydrometrocolpos is a malformation that can be treated but the fetal survival depends on the other associated anomalies and the time of prenatal diagnosis. This rare anomaly can be seen in utero at 20–24 weeks of gestation.
- An early evaluation of vaginal and urinary system decompression is recommended to avoid later complications.
- The successful outcome of this anomaly depends on correct and early prenatal diagnosis, absence of renal dysplasia or neonatal cardiorespiratory distress and teamwork between different specialists, including obstetricians, neonatologists and paediatric surgeons.
- Fetal autopsy is indicated in all the cases of fetal demise and abnormal ultrasound findings so as to find out the cause and make the complete diagnosis.

REFERENCES

- 1 Leung WC, Lam YH, Tang MH. Isolated foetal ascitis. *Hong Kong Med J* 2001;7:432–4.
- 2 Arena F, Romero C, Cruccetti A, *et al*. The neonatal management and surgical correction of urinary hydrometrocolpos caused by a persistent urogenital sinus. *BJU Int* 1999;84:1063–8.
- 3 Gul A, Yildirim G, Gedikbasi A, *et al*. Prenatal ultrasonographic features of persistent urogenital sinus with hydrometrocolpos and ascitis. *Arch Gynecol Obstet* 2008;278:493–6.
- 4 Camanni D, Zaccara A, Capitanucci ML, *et al*. Isolated fetal ascitis secondary to persistent urogenital sinus. *Obstet Gynecol Int* 2009;2009:219010.
- 5 Blask AR, Sanders RC, Gearhart JP. Obstructed uterovaginal anomalies: demonstration with sonography. Part I. Neonates and infants. *Radiology* 1991;179:79–83.
- 6 Adam MC, Ludlow J, Brock JW III, *et al*. Prenatal urinary ascites and persistent cloaca: risk factors for poor drainage of urine or meconium. *J Urol* 1998;160:2179–81.
- 7 Pautela J, Melo MA, Borges G, *et al*. Prenatal diagnosis of persistent urogenital sinus with duplicated hydrometrocolpos and ascites—a case report. *Fetal Diagn Ther* 2010;28:229–32.
- 8 El Bishry G. The outcome of fetal ascites. *Eur J Obstet Gynecol Reprod Biol* 2008;13:43–6.
- 9 Schmider A, Henrich W, Reles A, *et al*. Etiology and prognosis of fetal ascitis. *Fetal Diagn Ther* 2003;18:230–6.
- 10 Chen CP, Liu FF, Jan SW, *et al*. Ultrasound-guided fluid aspiration and prenatal diagnosis of duplicated hydrometrocolpos with uterus didelphys and septate vagina. *Prenat Diagn* 1996;16:572–6.

Copyright 2014 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- Submit as many cases as you like
- Enjoy fast sympathetic peer review and rapid publication of accepted articles
- Access all the published articles
- Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow