

SHORT COMMUNICATION

EVOLUTION OF SONOGRAPHIC FINDINGS IN A
FETUS WITH AGENESIS OF THE URETHRA,
VAGINA, AND RECTUM

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SUMMARY

In a 12-week gestation fetus, routine ultrasound examination suggested dilated loops of bowel. Repeat scans at 13 and 15 weeks showed normal growth but persistence of bowel dilatation. At 20 weeks there was megacystis, hyperechogenic bowel, ascites, and oligohydramnios. The diagnosis of cloacal abnormality was suspected and the parents chose to have pregnancy termination. Post-mortem examination demonstrated female pseudohermaphroditism with agenesis of the urethra, vagina, and rectum.

KEY WORDS: ultrasound; cloacal abnormality; female pseudohermaphroditism

CASE REPORT

A healthy 17-year-old primigravida presented for a routine ultrasound examination at 12 weeks of gestation. The scan demonstrated a singleton pregnancy and the crown-rump length (52 mm) was compatible with the gestation calculated from the last menstrual period. The fetal abdomen was distended by the presence of multiple cysts compatible with loops of dilated bowel (Fig. 1), but there was no other obvious abnormality. The parents were counselled that this was a very unusual finding because bowel obstruction usually presents after 24 weeks. The possibility was raised that the findings were due to an underlying cloacal abnormality with retrograde filling of the bowel with urine. Repeat scans at 13 and 15 weeks showed normal growth but persistence of bowel

dilatation. At 20 weeks, an ultrasound scan demonstrated megacystis, bilateral mild hydro-nephrosis and hyperechogenic bowel, ascites and oligohydramnios (Fig. 2). The patient was counselled regarding the risks of cloacal abnormality and pulmonary hypoplasia and opted for termination of pregnancy.

At post-mortem examination there was a phenotypically normal male fetus (Fig. 3) consistent with 21 weeks of gestation and weighing 442 g. The chin was receding, the ears were large and flat, and there were only two vessels in the umbilical cord. The abdomen was distended and the abdominal wall was thin. Internal examination confirmed the presence of ascites with clear straw-coloured fluid. There was urethral atresia with associated dilatation of the bladder and ureters but the kidneys were normal. The internal genitalia were female with normal ovaries, hydrometrocolpos, and vaginal atresia; the uterus was bicornuate. The ovaries appeared normal. There was anal and rectal agenesis and the entire colon was distended

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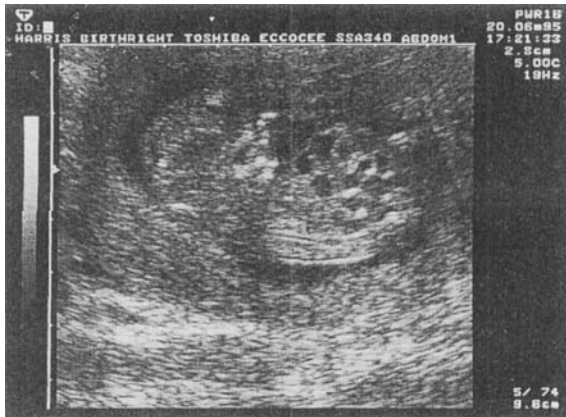


Fig. 1—Prenatal ultrasonographic longitudinal view of the fetus at 12 weeks of gestation showing multiple cysts compatible with loops of dilated bowel



Fig. 2—Prenatal ultrasonographic transverse view of the fetus at 20 weeks of gestation showing liver, hyperechogenic bowel, ascites, and oligohydramnios

with meconium. There was also agenesis of the middle segment of the oesophagus and moderate pulmonary hypoplasia. X-ray examination demonstrated abnormalities of the second, third, and fourth sacral vertebrae and absence of the coccyx. Chromosome analysis was performed on fetal skin and ovary and the karyotype was 46,XX and therefore the fetus had female pseudohermaphroditism.

DISCUSSION

This report illustrates the evolution of sonographic findings between 12 and 20 weeks of gestation in a case of female pseudohermaphroditism (FPH) with urethral, vaginal, and anal

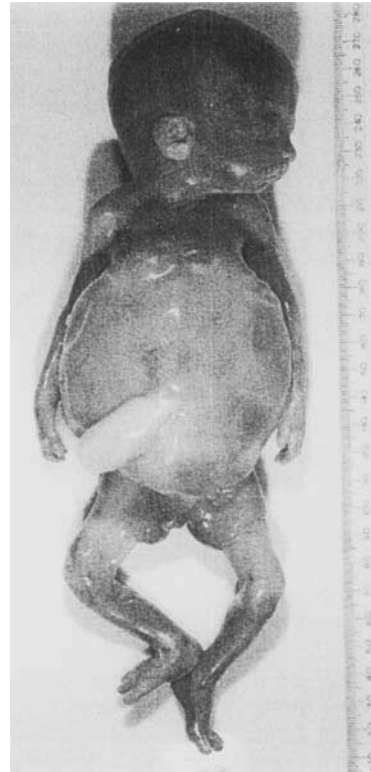


Fig. 3—Post-mortem photograph of the fetus at 21 weeks of gestation showing normal male external genitalia, a receding chin, large flat ears, and a grossly distended abdomen

atresia. At 12 weeks the sonographic finding was dilated bowel, but by 20 weeks the bowel was not distended and there was megacystis, ascites, and oligohydramnios. Small bowel obstruction is usually diagnosed by ultrasonography as distended sonolucent loops of bowel after 24 weeks of gestation because presumably before this gestation the volume absorbed by the stomach and upper bowel is greater than the amount swallowed. Anal and rectal agenesis cannot usually be diagnosed by ultrasound examination even during the third trimester because all swallowed amniotic fluid is absorbed in the small bowel. A possible explanation for our finding that in rectal agenesis the bowel is dilated at 12 weeks but subsequently resolves is that normally in the early second trimester most of the swallowed fluid is not absorbed but is passed through the rectum into the amniotic cavity. In rectal agenesis, the fluid is retained in the distended bowel and with advancing gestation and increasing absorption in relation to the degree of swallowing, there is resolution of the dilated

bowel. If this hypothesis were true, then prenatal diagnosis of rectal agenesis may become feasible by ultrasound scanning at 12 weeks.

An alternative explanation for the findings is that the underlying condition was a cloacal abnormality, where the upper vagina, upper urethra, and sigmoid colon open into a common cavity. The dilated loops of bowel at the 12-week scan could have been the consequence of preferential retrograde filling of the colon with sonolucent urine; despite urethral atresia, the amniotic fluid volume was normal because the main source of fluid at this gestation is not fetal urine. Subsequently, increased pressure within the common cavity resulted in megacystis and hydrometrocolpos with retrograde flow of urine through the Fallopian tubes into the peritoneal cavity. Obliteration of a possible fistulous connection between the common cavity and the blind-ending sigmoid colon could then explain the lack of bowel dilatation at the 20-week scan.

Previous reports on the prenatal diagnosis of cloacal abnormalities are confined to the second trimester of pregnancy. The typical sonographic appearance is a cystic pelvic mass which may be associated with ascites, hydronephrosis, spina bifida, exomphalos, and oligohydramnios (Petrikovsky *et al.*, 1988). The prognosis of cloacal abnormalities is related to the presence of associated anomalies and the amniotic fluid volume. In a review of eight publications reporting on 11 cases with multiple defects, the seven with oligohydramnios resulted in stillbirth or early neonatal death, whereas in three of the four cases with normal amniotic fluid volume the babies survived after postnatal surgery (Petrikovsky *et al.*, 1988).

FPH refers to prenatal masculinization of the external genitalia in a genetically female fetus. This may be the consequence of intrauterine exposure to masculinizing hormones, due to such conditions as fetal congenital adrenal hyperplasia, virilizing maternal adrenal or ovarian tumours, polycystic ovarian disease, maternal congenital adrenal hyperplasia, and exogenous hormone administration. FPH can also occur as a multisystem developmental abnormality without a hormonal trigger (Perloff *et al.*, 1953). The condition is sporadic and the primary pathogenetic mechanism

is not clearly understood but may be due to abnormal expression of genes which would normally be regulated by testosterone (Seaver *et al.*, 1994). In a series of 36 cases of FPH with associated anomalies, uterovaginal, renal, gastrointestinal and other abnormalities including vertebral or cardiac defects were found in 75, 72, 72, and 58 per cent of the cases, respectively (Lubinsky, 1980). The specific defects associated with the condition include duplex uteri, blind-ending vagina, recto-vaginal fistula, renal agenesis, megacystis, tracheo-oesophageal fistula, imperforate anus, sacral agenesis, and myelomeningocele (Lubinsky, 1980; Wenstrup and Pagon, 1985; Seaver *et al.*, 1994). As illustrated in our case, the condition may also be associated with oesophageal abnormalities and in this respect there is an overlap with the VACTERL syndrome (vertebral, anal, cardiovascular, tracheo-oesophageal, renal, and limb malformations) (Temtamy and Miller, 1974). Anal atresia and genitourinary anomalies are also associated with the caudal regression syndrome (renal agenesis or dysplasia, sacral agenesis, and lower limb hypoplasia) (Loewy *et al.*, 1987).

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