CHAPTER 1
Antenatal Diagnosis: Surgical Aspects

CASE 1

At 18 weeks' gestation, right fetal hydronephrosis is diagnosed on ultrasonography.

Q 1.1 Discuss the further management during pregnancy.

Q 1.2 Does antenatal diagnosis improve the postnatal outlook for this condition?

CASE 2

An exomphalos is diagnosed on the 18-week ultrasound scan.

Q 2.1 What further evaluation is required at this stage?

Q 2.2 Does this anomaly influence the timing and mode of delivery?

Antenatal diagnosis is one of the most rapidly developing fields in medical practice. While the genetic and biochemical evaluation of the developing fetus provides the key to many medical diagnoses, the development of accurate ultrasound has provided the impetus to the diagnosis of surgical fetal anomalies. At first, it was expected that antenatal diagnosis of fetal problems would lead to better treatment and an improved outcome. In some cases, this is true. Antenatally diagnosed fetuses with gastroschisis are now routinely delivered in a tertiary-level obstetric hospital with neonatal intensive care in order to prevent hypothermia and delays in surgical treatment, and the results of treatment have improved. In other cases, such as congenital diaphragmatic hernia, these expectations have not been fulfilled because antenatal diagnosis has revealed a number of complex and lethal anomalies which in the past never survived the pregnancy and were recorded in the statistics of fetal death in utero and stillbirth.

Indications and timing for antenatal ultrasound

Most pregnancies are now assessed with a mid-trimester morphology ultrasound scan, which is usually performed at 18–20 weeks' gestation [Fig.1.1]. The main purpose of this examination is to assess the obstetric parameters of the pregnancy, but the increasingly important secondary role of this study is to screen the fetus for anomalies. Most fetal anomalies can be diagnosed at 18 weeks, but some only become apparent later in the pregnancy. Renal anomalies are best seen on a 30-week ultrasound scan, as urine flow is low before 24 weeks. Earlier transvaginal scanning may be performed in special circumstances, such as a previous pregnancy with neural tube defect, and increasingly to detect early signs of aneuploidy. Fetal magnetic resonance imaging is increasingly being used to assess the developing fetus in cases of suspected or confirmed fetal anomalies without exposing the fetus or mother to ionising radiation.

Natural history of fetal anomalies

Before the advent of ultrasonography (as earlier), paediatric surgeons saw only a selected group of infants with congenital anomalies. These babies had survived the pregnancy and lived long enough after birth to reach surgical attention. Thus, the babies coming to surgical treatment were already a selected group, mostly with a good prognosis.

Jones' Clinical Paediatric Surgery, Seventh Edition. Edited by John M. Hutson, Michael O'Brien, Spencer W. Beasley, Warwick J. Teague and Sebastian K. King.

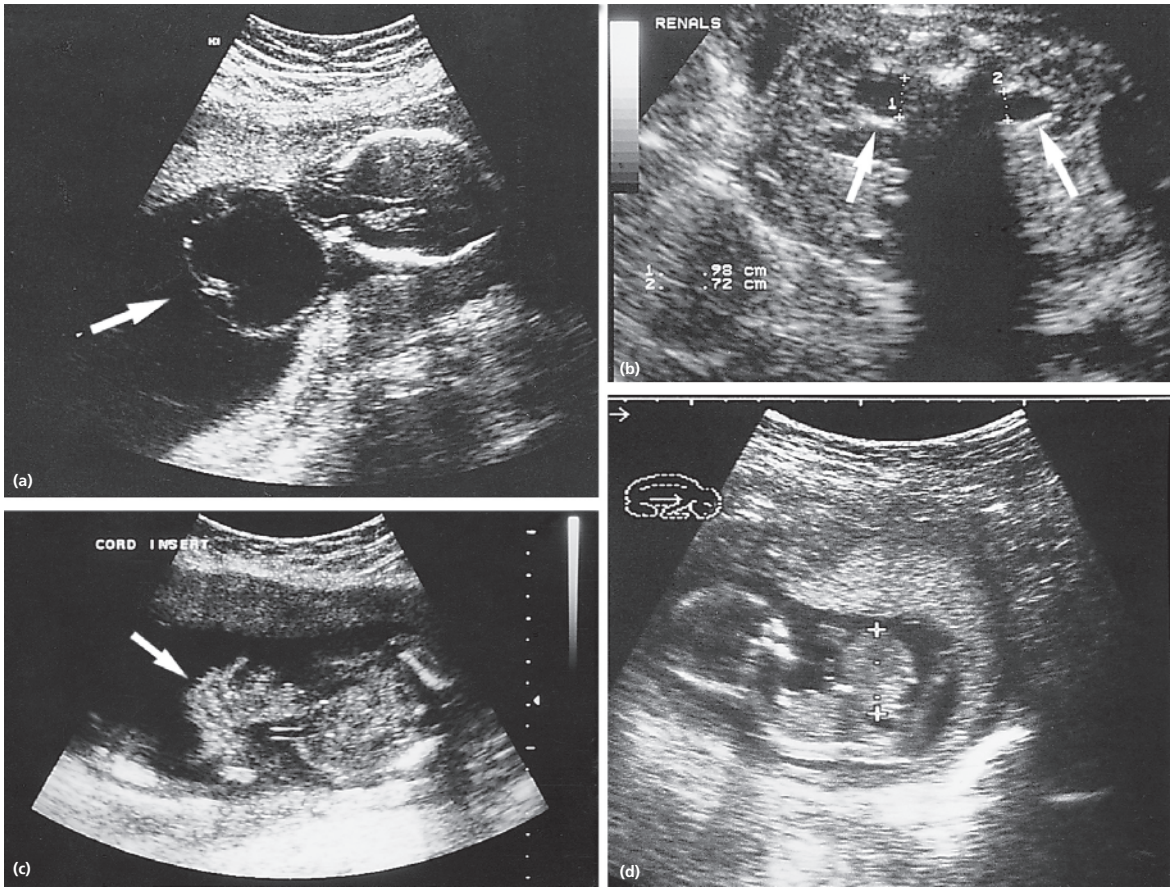

Figure 1.1 (**a**) Encephalocele shown in a cross section of the fetal head. The sac protruding through the posterior skull defect is arrowed. (**b**) Bilateral hydronephrosis shown in an upper abdominal section. The dilated renal pelvis containing clear fluid is marked. (**c**) The irregular outline of the free-floating bowel in the amniotic cavity of a term baby with gastroschisis. (**d**) A longitudinal section through a 14-week fetus showing a large exomphalos. The head is seen to the left of the picture. The large sac (marked) is seen between blurred (moving) images of the arms and legs.

Antenatal diagnosis has exposed surgeons to a new group of conditions with a poor prognosis, and at last, the full spectrum of pathology is coming to surgical attention. For example, posterior urethral valve causing obstruction of the urinary tract was thought to be rare, with an incidence of 1:5000 male births; most cases did well with postnatal valve resection. It is now known that the true incidence of urethral valve is 1:2500 male births, and these additional cases did not come to surgical attention as they developed intrauterine renal failure, with either fetal death or early neonatal death from respiratory failure because of Potter syndrome. It was thought that antenatal diagnosis would improve the outcome of such congenital anomalies, but the overall results have appeared to become worse with the inclusion of these severe *new* cases.

In the same way, antenatal diagnosis has exposed the significant *hidden mortality* of congenital diaphragmatic hernia [Fig. 1.2]. Previously, congenital diaphragmatic hernia diagnosed after birth was not commonly associated with multiple congenital anomalies, but now, antenatal diagnosis has uncovered a more severe subgroup with associated chromosomal anomalies and multiple developmental defects. It is now apparent that the earlier the congenital diaphragmatic hernia is diagnosed in utero, the worse the outcome.

Despite these problems, there are many advantages in antenatal diagnosis. The outcomes of many congenital

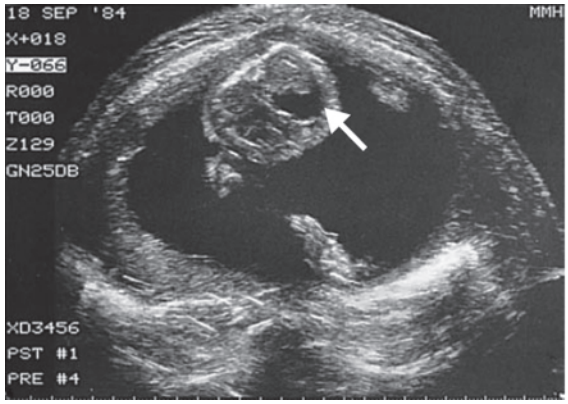

Figure 1.2 Cross section of a uterus with marked polyhydramnios. The fetal chest is seen in cross section within the uterus. The fluid-filled cavity within the left side of the chest is the stomach protruding through a congenital diaphragmatic hernia (arrow).

anomalies are improved by prior knowledge of them before birth.

Management following antenatal diagnosis

Fetal management

Cases diagnosed antenatally may be classified into three groups:

Good prognosis

In some cases, such as a unilateral hydronephrosis, there is no role for active antenatal management, and the main task is to document the progress of the condition through pregnancy with serial ultrasound scans. The detailed diagnosis is made with the more sophisticated range of tests available after birth, and the incidence of urinary tract infections (UTIs) may be reduced with prophylactic antibiotics commenced at birth. Thus, a child with severe vesicoureteric reflux may go through the first year of life without any UTIs. If the parents receive counselling by an experienced surgeon, they have time to understand the condition, its treatment and prognosis. With such preparation, the family may cope better with the birth of a baby with a congenital anomaly.

The paediatric surgeon also has an important role to play in advising the obstetrician on the prognosis of a particular condition. Some cases of exomphalos are easy to repair, whereas in others, the defect may be so large that primary repair will be difficult. In addition, there may be major chromosomal and cardiac anomalies, which may alter the outcome. In other conditions, the outlook for a congenital defect may change as treatment improves. Gastroschisis was a lethal condition before 1970, but now, management has changed and there is a 95% survival rate. In those cases with a good prognosis, fetal intervention is not indicated, and the pregnancy should be allowed to continue to close to term. The mode of delivery will usually be determined on obstetric grounds. Babies with exomphalos may be delivered by vaginal delivery if the birth process is easy. Primary caesarean section may be indicated for major exomphalos to prevent rupture of the exomphalos and damage to the organs such as the liver, as well as for obstetric indications. There is evidence that in fetuses with large neural tube defects, further nerve damage may occur at vaginal delivery, and caesarean section may be preferred in this circumstance. If urgent neonatal surgery is required, for example, in gastroschisis, the baby should be delivered at a tertiary obstetric unit with appropriate neonatal intensive care. In other cases (e.g. cleft lip and palate), where urgent surgery is not required but good family and nursing support is important, delivery closer to the family's home may be more appropriate. Antenatal planning and family counselling give us the opportunity to make the appropriate arrangements for the birth. A baby born with gastroschisis in the middle of winter in a bush nursing hospital in the mountains, many hours away from surgical care, may have a very different prognosis from a baby with the same condition born at a major neonatal centre.

Poor prognosis

Anencephaly, congenital diaphragmatic hernia with major chromosomal anomalies or urethral valve with early intrauterine renal failure are examples of conditions with a poor prognosis. These are lethal conditions, and the outcome is predetermined before the diagnosis is made.

Late deterioration

In most cases, initial assessment of the fetal anomaly will indicate a good prognosis with no reason for interference. However, later in gestation, the fetus may deteriorate, and some action must be undertaken to prevent

a lethal outcome. An example would be posterior urethral valve causing lower urinary tract obstruction. Early in the pregnancy, renal function may be acceptable with good amniotic fluid volumes, but on follow-up ultrasound assessment, there may be loss of amniotic fluid with oligohydramnios as a sign of renal failure. There are several approaches to this problem. If the gestation is at a viable stage, for example, 36 weeks, labour may be induced, and the urethral valve treated at birth. If the risks of premature delivery are higher, for example, at 28 weeks' gestation, temporary relief may be obtained by using percutaneous transuterine techniques to place a shunt catheter from the fetal bladder into the amniotic cavity. These catheters tend to become dislodged by fetal activity. A more definitive approach to drain the urinary tract is intrauterine surgery to perform a vesicostomy and allow the pregnancy to continue. This procedure has been performed with success in a few cases of posterior urethral valve. These patients are highly selected, and only a few special centres are able to perform intrauterine surgery. At present, this surgery is regarded as experimental and reserved for rare situations, but this may not always be the case.

Surgical counselling

When a child is born with unanticipated birth defects, there is inevitably shock and confusion until the diagnosis is clarified, and the family begins to assimilate and accept the information given to them. Important treatment decisions may have to be made urgently while the new parents are still too stunned to play any sensible part in the ongoing care of their baby. Antenatal diagnosis has changed this situation. New parents may now have many weeks to understand and come to terms with their baby's condition. With suitable preparation, they may play an active role in the postnatal treatment choices for their newborn baby.

The paediatric surgeon who treats the particular problem uncovered by antenatal diagnosis is in the best position to advise the parents on the prognosis and further treatment of the baby. Detailed information on the management after birth, with photographs before and after corrective surgery, allows the parents to understand the operative procedures. The opportunity to meet other families with a child treated for the same condition may give time for the pregnant woman and her partner to understand the problem prior to birth. Handling and nurturing the baby immediately after birth is an important part of bonding. Parents and nursing staff suddenly confronted with a newborn baby with an unexpected anomaly, such as sacrococcygeal teratoma, may be afraid to handle the baby prior to the baby being taken away to another hospital for complex surgery. Parents in this situation may take many months to bond with the new baby and to understand fully the nature of the problem. Prepared by antenatal diagnosis, parents realise they may handle and nurture the baby, understand the nature of the surgery and form a bond with the baby. Thus, instead of being stunned by the birth of a baby with a significant malformation, the new parents may play an active part in the postnatal surgical management and provide better informed consent for surgery.

KEY POINTS

- Antenatal diagnosis with ultrasound scanning has revealed the natural history of some anomalies and made the prognosis seem worse (e.g. congenital diaphragmatic hernia, posterior urethra valve).
- Antenatal diagnosis has allowed surgical planning (and occasional fetal intervention), as well as providing time for parents to be informed prior to the birth.

Further reading

Fleeke AW (2012) Molecular clinical genetics and gene therapy. In: Coran AG, Adzick NS, Krummel TM, Laberge J-M, Shamberger RC, Caldamone AA (eds) *Pediatric Surgery*, 7thEdn. Elsevier Saunders, Philadelphia, pp. 19–26.

Lee H, Hirose S, Harrison MR (2012)Prenatal diagnosis and fetal therapy. In: Coran AG, Adzick NS, Krummel TM, Laberge J-M, Shamberger RC, Caldamone AA (eds) *Pediatric Surgery*, 7th Edn. Elsevier Saunders, Philadelphia, pp. 77–88.