Introduction

1 Antenatal Diagnosis – Surgical Aspects

Case 1

At 18-weeks' gestation, right fetal hydronephrosis is diagnosed on ultrasound.
Q 1.1 Discuss the further management during pregnancy.
Q 1.2 Does the antenatal diagnosis improve the postnatal outlook for this condition?

Case 2

An exomphalos is diagnosed at the 18-week ultrasound.
Q 2.1 What further evaluation is required at that 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. Whilst 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 for the diagnosis of surgical fetal anomalies. At first, it was expected that the 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 will be delivered in a tertiary-level obstetric hospital with neonatal intensive care in order to prevent hypothermia, and the results of treatment have improved. In other cases, such as in diaphragmatic hernia, these expectations have not been fulfilled because antenatal diagnosis has opened a Pandora's box of complex and lethal anomalies, which in the past never survived the pregnancy, and were recorded in the statistics as fetal death *in utero* and stillbirth.

Indications and timing for antenatal ultrasound

All pregnancies are now assessed with a mid-trimester ultrasound, which is usually performed at 17–18-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 anomalies are picked up at 18 weeks, but some only become apparent later in the pregnancy. Renal anomalies are best seen on a 30-week ultrasound as urine flow is low before 24 weeks. Earlier ultrasound examinations may be performed with transvaginal scanning in special circumstances, such as a previous pregnancy with neural tube defect and increasingly to detect early signs of aneuploidy. Magnetic resonance imaging of the developing fetus may be another means of fetal assessment in future.

Natural history of fetal anomalies

Before the advent of ultrasound, 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.

Antenatal diagnosis has brought surgeons into contact with 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 valves causing obstruction of the urinary tract were thought to

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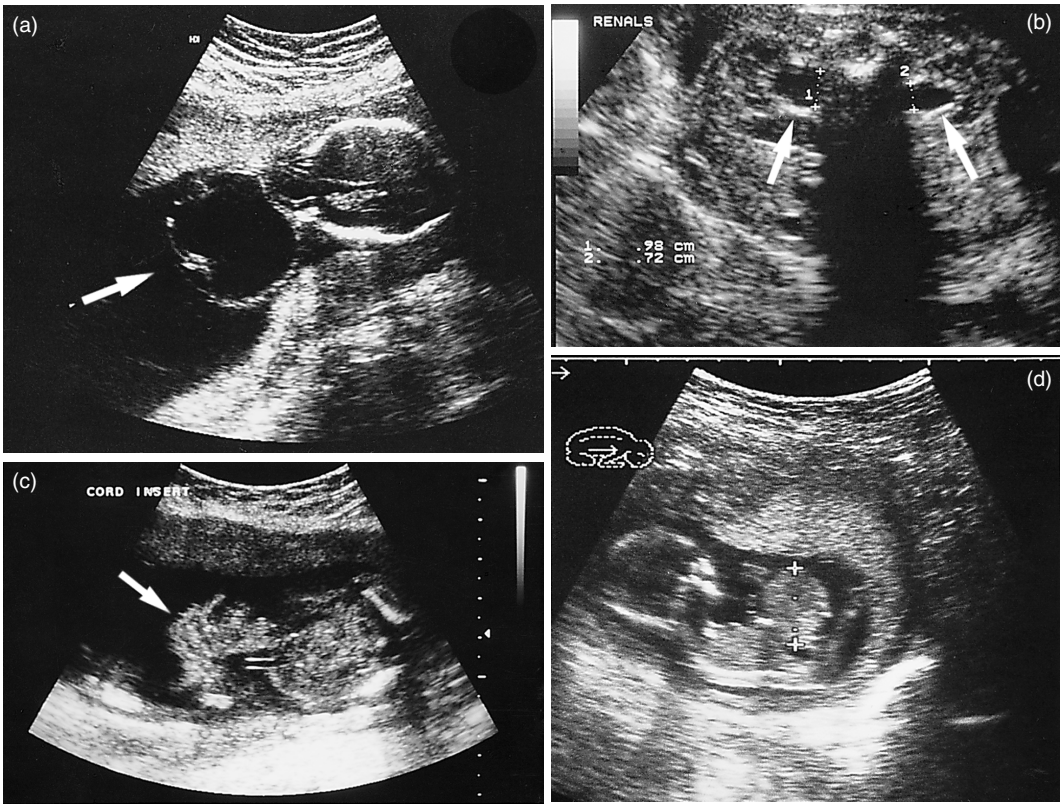

Figure 1.1 (a) Encephalocele shown in a cross section of the fetal head. The sac protruding through the posterior skull defect is arrowed. (b) Bilateral hydronephrosis shown in an upper abdominal section. The dilated renal pelvis containing clear fluid is marked. (c) The irregular outline of the free-floating 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.

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 valves 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 *in utero* or early neonatal death from respiratory problems such as Potter's 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 these severe 'new' cases being included.

There are similar problems with the antenatal diagnosis of diaphragmatic hernia [Fig. 1.2]. Congenital diaphragmatic hernia was not associated with multiple congenital anomalies when cases presented after birth. Now antenatal diagnosis of diaphragmatic hernia has uncovered a more severe subgroup with associated chromosomal anomalies and multiple developmental defects. It would seem that the earlier the diaphragmatic hernia is diagnosed, the worse is the outcome.

Despite these problems, there are many advantages in antenatal diagnosis. The outcome of many conditions is improved by the prior knowledge of 'congenital' anomalies.

Management following antenatal diagnosis

Fetal management

Cases diagnosed antenatally may be classified into three groups.

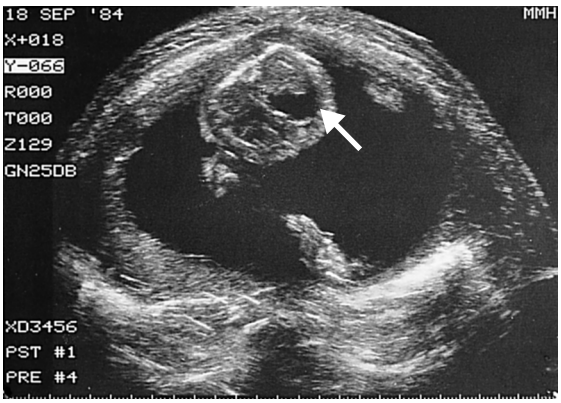

Figure 1.2. Cross section of a uterus with marked polyhydramnios. The fetal chest is seen in cross section within the uterus. The fluid-filled cavity within the left side of the chest is the stomach protruding through a diaphragmatic hernia (arrow).

Good prognosis

In some cases, such as a unilateral hydronephrosis, there is no place for active management, and the main task is to track the progress of the problem through pregnancy with serial ultrasound. A detailed diagnosis is made with the more sophisticated range of tests available after birth, and UTIs are prevented with prophylactic antibiotics commenced at birth. Thus, a child with severe vesico-ureteric reflux may go through the first year of life without any UTIs. If the parents receive counselling by a surgeon experienced in the care of the particular problem, they have time to understand the condition. In the case of cleft lip seen on fetal ultrasound, the parents will have time to understand the nature of the problem by seeing photographs of the condition before and after surgery, and will have the chance to meet other families with this condition. With such preparation, the family can cope better with the birth of a baby with congenital anomalies.

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. Moreover, in some fetuses there may be major chromosomal and cardiac anomalies associated with the exomphalos which may alter the outcome. In exomphalos, therefore, the prognosis varies from good to poor. In other conditions, the outlook for a congenital defect may change as treatment improves. Gastroschisis was a lethal condition before 1970, but now management of the disease 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 run its course. 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. In other obstetric circumstances, caesarian section may be indicated to prevent rupture of the exomphalos. There is evidence that spina bifida cases may undergo further nerve damage at vaginal delivery, and caesarian section may be preferred in this circumstance. If urgent neonatal surgery is required, for example, for gastroschisis, the baby should be delivered at a tertiary obstetric unit with a neonatal intensive care unit and neonatal surgical service. In other cases, for example, cleft lip and palate, where urgent surgery is not required but good family and nursing support is important, delivery close 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, will have a very different outlook from a baby with the same condition born at a major neonatal centre.

Poor prognosis

Anencephaly, diaphragmatic hernia with major chromosomal anomalies or urethral valves 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

Initial assessment of the fetal anomaly indicates a good prognosis with no reason for interference, but, later in gestation, the fetus deteriorates and some action must be undertaken to prevent a lethal outcome. An example would be the lower urinary tract obstruction seen in posterior urethral valves. Early in the pregnancy, renal function may be acceptable with good amniotic fluid volumes. However, on follow-up ultrasound assessment, there may be loss of liquor with oligohydramnios as a sign of intrauterine renal failure. There are several ways to treat this problem. If the gestation is at a viable stage, for example, 36 weeks, labour could be induced and the urethral valves treated at birth. If the risks of premature delivery are higher, for example, for 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 valves. These patients are highly selected and only a few special centres perform intrauterine surgery. At present this surgery is regarded as experimental, and reserved for rare situations, but this may not always be the case.

Antenatal ultrasound has become the most important means of diagnosing fetal anomalies and has given us a valuable means of understanding the natural history of developmental abnormalities.

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 begin to assimilate and accept the information given to them, and make plans for the future. Important treatment decisions have to be made urgently while the new parents are still too stunned to play any sensible part in 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 problem. With suitable preparation, they can play an active role in the postnatal diagnosis and treatment choices for their newborn baby.

The paediatric surgical specialist 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 and come to terms with the surgical procedures. The opportunity to meet other families with a child treated for the same condition gives time for the pregnant woman and her partner to understand the problem before the 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 the unexpected finding of a gross anomaly, such as sacrococcygeal teratoma, may be afraid to handle the baby who is then taken away to another hospital for complex surgery. Parents in this situation may take many months to relate to the new baby and understand the nature of the problem. Prepared by antenatal diagnosis, the nursing staff and parents realise they can handle and nurture the baby. They understand the nature of the surgery and maintain their bond with the baby, Thus instead of being stunned by the birth of a malformed baby, the new parents can play an active part in the postnatal surgical management and provide better informed consent for surgery.

Key Points

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

Further reading

Flake AW (2006) Molecular clinical genetics and gene therapy. In: Grosfeld JL, O'Neill JA, Fonkalsrud EW, Coran AG (eds) *Pediatric Surgery*, 6th Edn. Mosby, Elsevier, Philadelphia, pp. 11–20.

Harrison MR (2006) The fetus as a patient. In: Grosfeld JL, O'Neill JA, Fonkalsrud EW, Coran AG (eds) *Pediatric Surgery*, 6th Edn. Mosby, Elsevier, Philadelphia, pp. 77–88.