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## CHAPTER 43

# Obstetrical Diseases and Foetal Anomalies

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### INTRODUCTION

Obstetrical ultrasonography (US) plays an important role in the management of pregnancies. The practice of performing two or three scans routinely for each pregnancy is fast becoming a norm. Most centres offer a screening scan for foetal anomalies in the second trimester. Routine scans in the first trimester for dating of the pregnancy and growth scans in the third trimester are also becoming a norm in some centres. Advances in the last two decades have made US the modality of choice in the diagnosis of many foetal anomalies and other complications found in pregnancy. With current equip-

ment, a number of foetal anatomical disorders can now be confidently diagnosed. The availability of higher resolution scans has resulted in more accurate assessment of difficult anatomical areas such as the foetal heart.

The diversity of organ systems involved in foetal imaging necessitates the involvement of multidisciplinary teams to determine the course of management for different anomalies. Such teams include obstetricians, neonatologists, paediatricians of various subspecialties, paediatric surgeons, geneticists, pathologists, and other professionals from paraclinical disciplines. Together, they provide the assistance necessary in the counselling of affected parents. Despite the numerous controversies and ethical issues that surround obstetrical US,

it is still a rewarding experience and in most cases, will undoubtedly continue to provide reassurance to many pregnant mothers.

### INDICATIONS FOR OBSTETRICAL US

1. Complications during the first trimester.
2. Detection of foetal life.
3. Dating of pregnancy (first and early second trimesters).
4. Detection of multiple pregnancies.
5. Screening for foetal anomalies.
6. Assessment of foetal growth.
7. Assessment of foetal well being.
8. Placental disorders.
9. Guide interventional procedures (e.g. amniocentesis, chorionic villus sampling).
10. Detection of uterine and adnexal abnormalities.

#### First trimester scan

The indications for performing a scan during the first trimester include:

- Estimation of menstrual age (dating scan)
- Vaginal bleeding
- Hyperemesis
- Other signs or symptoms referable to a pelvic pathology



Figure 43.1: US scan shows a right ectopic pregnancy (E) from in-vitro fertilisation, located next to the right ovary (ov).

#### Estimation of menstrual age

Dating scans should preferably be performed during the early first trimester, where ethnic variation of foetal size is minimal. Parameters used in the dating of pregnancies include gestational sac size and foetal crown-rump length (CRL). CRL is the more commonly used and reliable parameter of the two. By using this parameter, gestational age may be estimated to within a week [1].

#### Vaginal bleeding

The purpose of the scan is to confirm the presence of an intrauterine gestation, assess for signs of foetal life, and rule out ectopic pregnancies. In the early gestational stage, correlation of the scan findings and the serum human chorionic gonadotrophin (hCG) level is necessary to rule out ectopic pregnancies. Transvaginal US (TVUS) improves the sensitivity in detection of early intrauterine pregnancies. US in patients with symptoms of threatened abortion or abnormal vaginal bleeding during the first trimester may reveal one of the following features listed below.

**Absence of gestational sac** may be due to wrong dates, complete/incomplete abortion, or ectopic pregnancy. The pregnancy may be too early for visualisation of the intrauterine sac. Alternatively, the patient may have had a complete or incomplete abortion. Lastly, ectopic pregnancy (Fig. 43.1) must be considered. With TVUS scans, most intrauterine gestational sacs should be seen when the serum hCG is at least 1,500 IU/L (Third International Reference Preparation) or higher. Due to variations in skills and ultrasound equipment among different centres, each centre will need to establish its own threshold value of serum hCG for the detection of early intrauterine pregnancies. In cases of ectopic pregnancies, free fluid and an adnexal mass, which is separate from the ovary, may be identified.

If the **gestation sac is present**, an assessment



Figure 43.2: Molar pregnancy. Longitudinal US scan shows an enlarged uterus containing a large mass with numerous small cystic spaces.

may be made to determine if the pregnancy is likely to continue or not. Indications of a failed pregnancy in the presence of a gestational sac include absence of yolk sac when the mean sac diameter is 10 mm or larger, and absence of an embryo in a sac with a mean diameter of more than 15 mm. These dimensions are applicable to TVUS scans. The mean sac diameter is obtained by measuring the inner wall of the gestational sac in three orthogonal planes, and averaging out the dimensions. The lack of appropriate growth of the gestational sac over time is often used to confirm non-viability in such cases. Other indicators of threatened abortion include gestational sac irregularity, haemorrhage around the sac, and foetal bradycardia.

### **Hydatidiform moles**

The classical clinical presentation, as well as the US presentation of hydatidiform mole consisting of a mass with numerous small cystic areas within the uterus (Fig. 43.2), is becoming less common [2], due to the widespread use of US during the first trimester and the early evacuation of failed pregnancies. Some molar pregnancies are not distin-

guishable from anembryonic gestational sacs. With the use of TVUS scans and improved resolution of today's machines, the minute cysts associated with hydropic villi can be seen in smaller lesions.

### **Detection of foetal life**

US is often used to assess for foetal life. Signs of foetal life include foetal heart activity and movements. These signs are used in the first trimester assessment when the embryo is seen, as well as during later stages of pregnancy. While absence of cardiac activity is the sign to look out for in cases of suspected intrauterine death, there are other signs such as overlapping of the skull sutures (Fig. 43.3), abnormal foetal posture, and echogenic foci, that represent gas in the foetal circulation.

### **Screening for foetal anomalies**

Besides foetal biometry, screening for foetal anomalies is a routine aspect of the mid-trimester US scan that is usually performed from 18 to 20 weeks. Depending on the skill of the operator and the type of equipment used, screening scans may be performed as early as 16 weeks. Some centres offer



Figure 43.3: US scan shows overlapping skull sutures (arrow) indicative of foetal death.

first trimester screening with transvaginal US scanning. With earlier screening scans, there may be a need to attend more than one session, as some structures, such as the foetal heart, may not be well visualised.

A systematic review of the foetal anatomy can reveal a wide range of abnormalities. They range from actual physical anomalies to “soft” markers of chromosomal abnormality (Table 43.1). Physical anomalies may be due to the arrested development, persistence of primitive forms, malformations, or acquired conditions, such as tumours and infections. “Soft” markers refer to features that often do not constitute a physical abnormality, and may even be found in normal foetuses. Various studies have

shown some association between these signs and abnormal karyotypes. For a better understanding of the development of malformations in the foetus, the reader is encouraged to read texts on embryology [3]. Besides abnormal foetal anatomy, other ultrasonographical indicators of possible foetal abnormality include abnormal liquor volume and early growth retardation.

### CENTRAL NERVOUS SYSTEM ANOMALIES

The following are the more commonly diagnosed anomalies of the central nervous system:

- Neural tube defects: cranial, spinal, and combined
- Cranial: anencephaly, cephalocele
- Spinal: meningocele, myelomeningocele
- Combined: Arnold-Chiari malformation

<b>TABLE 43.1</b> <b>Ultrasonographical Markers of Chromosomal Abnormality</b>
<b>Physical anomalies</b>
Holoprosencephaly
Agenesis of corpus callosum
Dandy-Walker malformation
Mild ventriculomegaly
Cardiac structural anomalies
Duodenal atresia
Diaphragmatic hernia
Omphalocele
Foetal hydrops
Hypoplastic/ absent radius
Early intrauterine growth retardation
<b>“Soft” markers</b>
Nuchal translucency
Choroid plexus cyst
Echogenic bowel
Intracardiac echogenic foci
Abnormal amniotic fluid volume



Figure 43.4: Anencephalic foetus. US scan show an absence of cranium. Brain is replaced by disorganised soft tissues (large white arrows). Base of skull (black arrows) is present.

- Disorders of ventral induction: holoprosencephaly, dysgenesis of the corpus callosum
- Dandy-Walker malformation
- Others: tumours, infections, benign cysts

### Anencephaly

This is the most severe form of neural tube defect. It is due to failure of closure of the cephalic end of the neural tube. The supratentorial structures, which include the cerebral hemispheres and cranium, are not developed. The cranial structures may be replaced by a mass of disorganised angiomatous tissues (Fig. 43.4). The brain stem is formed, and is responsible for sustaining life in-utero. Infants born with anencephaly die at birth or soon afterwards. Female foetuses outnumber males in this anomaly. On US, the anencephalic foetus has a “frog-like” facies due to the position of the higher location of the orbits. The absence of the head is readily evident when one tries to obtain the biparietal diameter or head circumference.

### Spina bifida

Spina bifida results from non-closure of the caudal neural tube that forms the spinal cord. The degree of severity of this lesion varies. It can range from isolated non-development of the neural arches of the vertebra that is covered (spina bifida occulta),



Figure 43.5: Longitudinal US scan shows a spina bifida with a meningocele (arrows). Separation of the posterior ossification centres of the lower lumbar spine (arrowheads) is present.

to a defect associated with a sac (spina bifida cystica), to open defects in which the spinal cord is exposed to the exterior (myeloschisis). It can occur anywhere along the spine, but is more common in its lower half. Antenatal US is able to detect the open defect. Features include splaying of the bony ossification centres of the vertebral column, presence of a sac (Fig. 43.5) which may be due to a meningocele or myelomeningocele, or an open defect without the sac. The anomaly may sometimes be associated with malformations of the brain stem and cerebellum (Arnold-Chiari malformation).

### Cephaloceles

Cephaloceles result from defects in the development of the cranium. The term refers to the protrusion of intracranial structures through a cranial defect (Fig. 43.6). This may be due to herniation of



Figure 43.6: Transverse US scan shows an occipital encephalocele (black arrows) and a defect in the skull vault (white arrows). There is also absence of amniotic fluid due to bilateral renal agenesis.

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Figure 43.7: US scans show a large encephalocele (arrows) with microcephaly (arrowheads). Left image shows large portions of the cerebral hemispheres lying outside the cranium.

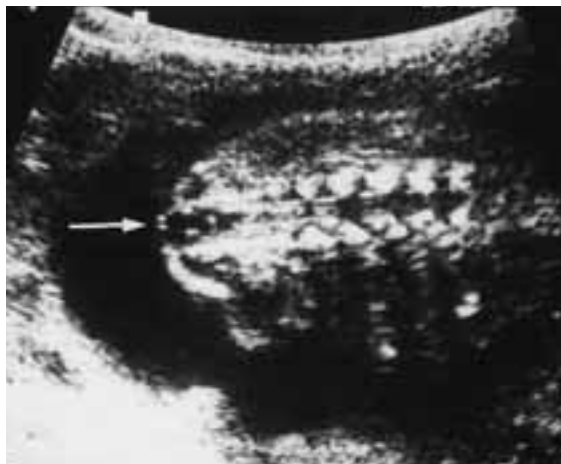


Figure 43.8: Longitudinal US scan shows an open spina bifida defect (arrow) at the lumbosacral region in a foetus with Arnold-Chiari malformation.

meninges (meningocoele) or brain (encephalocoele). The most common site of occurrence is in the midline of the occipital region, followed by the frontal midline, and least commonly, in the parietal region. Amniotic bands, which are sometimes the cause of these defects, result in lesions that are away from the midline. The encephalocele may contain a substantial amount of brain tissue, resulting in microcephaly (Fig. 43.7). It may be a feature of a syndrome, e.g. Mecke-Gruber syndrome.

### Arnold-Chiari malformation

A combination of spina bifida, ventriculomegaly, brain stem and cerebellar tonsillar hernia-



Figure 43.9: Arnold-Chiari malformation. (A) Transverse US scan shows effacement of the cisterna magna and deformity of the cerebellar hemispheres (arrows). (B) Transverse US scan shows bifrontal depressions in the skull (arrows) resulting in a "lemon" head.



43.10A



43.10B

Figure 43.10: Holoprosencephaly. (A) US scan shows a single large ventricle and fused thalami (T). (B) The foetus also has a single nostril and hypotelorism (closely-set orbits).

tion is seen in this condition. The spina bifida may be in the form of an open defect (Fig. 43.8), or is associated with a myelomeningocele. The cranial findings of this condition may sometimes be more readily diagnosed than the spina bifida. The cisterna magna is effaced (Fig. 43.9A) due to the downward displacement of the cerebellar tonsils and the medulla oblongata. In some cases, the cerebellum becomes severely compressed and is difficult to image. In a number of cases, the cross-section of the cranium resembles the shape of a lemon (Fig. 43.9B), due to the bifrontal depressions.

### Holoprosencephaly

The condition consists of a spectrum of anomalies with varying degrees of severity due to non-cleavage of the forebrain. Facial development is also affected, and the degree of severity reflects that of the brain. Facial anomalies include cyclopia, hypotelorism, midline facial cleft, and a single nostril (Fig. 43.10). Depending on severity, holoprosencephaly may be classified, from the most severely affected to the least, as alobar holoprosencephaly, semilobar holoprosencephaly and lobar holoprosencephaly.



Figure 43.11: Transverse US scan shows agenesis of corpus callosum with high riding third ventricle (3V). Widely-separated bodies of lateral ventricles (arrowheads) with dilatation at the occipital (O) horn (better seen in the half of the cranium away from the transducer).

### Dysgenesis of the corpus callosum

This anomaly may occur on its own, or in association with the Dandy-Walker malformation or holoprosencephaly. It may be recognised on an antenatal US scan by the appearance of the lateral ventricles that are slightly dilated at the occipital horns (colpocephaly). The bodies of the lateral ventricles are also parallel and the frontal horns are further apart. The high-riding third ventricle may sometimes be mistaken for the septum cavum pellucidum that is absent in this condition (Fig. 43.11). It is difficult to obtain a sagittal view of the foetal



Figure 43.12: Dandy-Walker malformation. Transverse US scan shows the cerebellar hemispheres (arrowheads) are widely separated by a dilated fourth ventricle (4V).



Figure 43.13: Hydrocephalus. Transverse US scan shows dilated frontal (F) and temporal (T) horns. (C: cerebellum).



Figure 43.14: Transverse US scan shows subtle finding of hydrocephalus. The choroid plexuses (c) in the dilated lateral ventricles are leaning towards the dependant ventricular walls.

head in order to visualise the corpus callosum. Due to the variety of outcomes in individuals with this condition, it is usually difficult to counsel the mothers of affected foetuses. Karyotyping is advised in such cases.

### Dandy-Walker malformation

In this condition, the cerebellar vermis fails to develop. The cerebellar hemispheres are widely separated. The fourth ventricle is dilated and communicates with the dilated cisterna magna (Fig. 43.12). The resulting appearance is similar to that of a posterior fossa cyst [4]. Associated anomalies include hydrocephalus, atresia of the exit foramina of the fourth ventricle, agenesis of the corpus callosum, encephalocele, and meningocele. Extracranial anomalies can occur in the heart, and the genitourinary, gastrointestinal, and skeletal systems [5]. Karyotyping is advised.

### Ventriculomegaly

Dilatation of the ventricles (Fig. 43.13) may be due to obstructive hydrocephalus, atrophy of the brain, or abnormal brain development (e.g.



Figure 43.15: Transverse US scan shows marked hydrocephalus. The cerebral cortex is thinned out. The choroid plexuses (arrows) are "dangling" towards the dependent part of the head. (T: thalamus).



Figure 43.16: Transverse US scan shows a choroid plexus cyst (between cursors).



Figure 43.17: US scan shows a non-specific echogenic lung (m). Differential diagnoses include sequestered lung, cystic adenomatoid malformation and bronchial obstruction.

colpocephaly). Although measurements for normal ventricular atrial diameter have been used to determine the presence of ventricular dilatation, morphological assessment of the choroid plexus is just as important. The upper limit of normal for the ventricular atrial diameter is 10 mm [6]. Assessment of the size of the choroid plexuses in relation to ventricular dimensions is also essential in assessing foetuses with ventricles that are at the upper limits of normal range [7] (Fig. 43.14). Lateral ventricles that are dilated show displacement of the choroid plexuses towards the dependent ventricular walls (Fig. 43.15).

### Choroid plexus cysts

These cysts (Fig. 43.16) are usually present in



Figure 43.18: Congenital diaphragmatic hernia. Transverse US scan shows loops of bowel (arrows) in the left side of the foetal thorax, displacing the foetal heart (FH) to the right. (S: spine).



Figure 43.19: Cystic adenomatoid malformation of the lung. Longitudinal US scan shows a large echogenic mass with cysts (arrowheads) in the foetal thorax.

isolation, but may be associated with trisomy 18. The discovery of such cysts warrants detailed scanning to look for features of trisomy 18. These cysts generally resolve after 24 weeks of pregnancy, and are not by themselves of clinical significance.

## NON-CARDIAC THORACIC ANOMALIES

These abnormalities may be detected on the basis of abnormal lung echogenicity (Fig. 43.17),



Figure 43.20: Longitudinal US scan shows a sequestered lung as an echogenic mass (m) under the diaphragm. (A: aorta).



Figure 43.21: Hydrothorax. Longitudinal US scan shows fluid outlining the foetal lung (L). (H: heart).



Figure 43.22: Abnormal four-chamber view due to atrio-ventricular septal defect or septum primum defect (asterisk).

presence of cystic structures, and displacement of the foetal heart, or its axis. The differential diagnoses of a foetal thoracic mass include:

- Congenital diaphragmatic hernia (Fig. 43.18)
- Congenital cystic adenomatoid malformation (Fig. 43.19)
- Sequestered lung (Fig. 43.20), congenital lobar emphysema, bronchogenic cyst
- Tracheal atresia (bilateral echogenic lungs)

Differentiation of the various types of thoracic masses may sometimes be difficult, given the overlap of ultrasonographical appearances. The development of hydrops and polyhydramnios are signs of poor foetal outcome. Other predictors of possible poor outcome include the degree of lung compression by the mass [8], the stage of pregnancy at which the anomaly is detected, and the presence of other anomalies. The improvement of chest findings and “disappearance” of echogenic lung masses on follow-up antenatal US have also been described. Post-natal follow-up of such cases, which should include CT scans, may reveal lung anomalies [9].

### Hydrothorax

Pleural effusion (Fig. 43.21) is an uncommon complication that may be seen in isolation, or together with other features of foetal hydrops. The cause for the unexplained isolated cases is probably due to blockage of lymphatic drainage. Aspiration of the pleural effusions may be performed antenatally. Some cases may progress rapidly to hydrops, and therefore, such cases should be monitored closely. The insertion of thoraco-amniotic shunts had been shown to alleviate the problem in some cases [10, 11].

### CARDIAC ANOMALIES

Anomalies in the foetal heart may be detected on the basis of an abnormal four-chamber view of the heart, abnormal cardiac axis, and abnormal out-flow tract features. Colour Doppler US is used to



Figure 43.23: Abnormal four-chamber view of a hypoplastic left heart. The normal left cardiac ventricle (asterisk) is not seen.

evaluate changes in blood flow resulting from some of these anomalies. Most anomalies can be identified with grey-scale US imaging.

#### Cardiac anomalies with abnormal four-chamber view

- Septal defects (if large enough)
- Atrio-ventricular septal defect (endocardial cushion defect) (Fig 43.22)
- Hypoplastic right/left heart (Fig. 43.23)
- Epstein anomaly
- Critical pulmonary/aortic stenosis
- Moderate/severe aortic coarctation
- Cardiac tumours

#### Anomalies that can present with a normal four-chamber view

- Tetralogy of Fallot
- Transposition of great vessels
- Double outlet right ventricle
- Small ventricular/atrial septal defects
- Mild aortic/pulmonary valvular stenosis
- Mild aortic coarctation

The discovery of a foetal cardiac anomaly usually warrants karyotyping [12]. The risk of chromosomal abnormality is further increased when other extracardiac anomalies are also present.



Figure 43.24: Foetal hydrops. US scan shows marked ascites (asterisk) outlining the liver (L) and bowels (G).



Figure 43.25: Foetal hydrops. Transverse US scan shows a fluid collection (asterisk) within the oedematous scalp.

#### Foetal hydrops

Foetal hydrops is characterised by the accumulation of excessive fluid in the body cavities (Fig. 43.24), and in the more severely affected cases, generalised subcutaneous oedema (Fig. 43.25). Hydrothorax, pericardial effusion and ascites occur together. In the early phase, isolated fluid accumu-



Figure 43.26: Duodenal atresia in Down syndrome. (A) Antenatal transverse US scan shows a “double-bubble” sign due to duodenal atresia. There was associated polyhydramnios. Post-natal (B) radiograph of the foetal chest and abdomen confirms the presence of the “double-bubble” due to duodenal obstruction.

lation may be seen. Close monitoring of these cases is essential. It is associated with a high risk of foetal death. Non-immune hydrops is caused by a wide spectrum of conditions. The causes include cardiac anomalies, infections, lymphatic obstruction, aneuploidy (Turner syndrome), haematological causes, vascular malformations resulting in high output cardiac failure, and placental anomalies. It necessitates detailed examination of the foetal heart, and checking for other malformations, as well as a host of laboratory investigations.

### NON-RENAL ABDOMINAL ANOMALIES

Abnormalities affecting the bowel may not present during the time of the mid-trimester screening US scan. These include bowel obstruction, and even cases of atresia. Such anomalies are often associated with polyhydramnios. The following anomalies may be detected on examining the foetal abdomen and the umbilical cord insertion site:

- Absent gastric bubble
- Duodenal atresia and other small bowel obstructions
- Abdominal wall defects: omphalocele, gastroschisis
- Cystic masses: choledochal cyst, ovarian cysts, hepatic lesions

#### Absent gastric bubble

When the foetal stomach is not visualised for 45 minutes during a screening US scan, a repeat scan should be performed at a different sitting. Non-visualisation of the stomach, or the finding of a smaller than average-sized one, may be a transient finding in normal cases. In cases where this finding persists, the likelihood of an abnormal outcome increases [13]. Abnormalities that are associated with non-visualisation of the stomach include oesophageal atresia, oligohydramnios, neuromotor syndrome, hydrops, or ectopic location of the

stomach (e.g. in congenital diaphragmatic hernia). A careful search for other anomalies should be made.

### Duodenal and small bowel obstruction

There are many causes of duodenal obstruction. They may be intrinsic (e.g. atresia, stenosis) or extrinsic (e.g. annular pancreas, Ladd bands). Diagnosis is based on the finding of a “double-bubble”, which represents the distended stomach and duodenum (Fig. 43.26). The two bubbles should interconnect. Peristalsis may be observed in the stomach. About 30% of foetuses with duodenal atresia have Down syndrome. Unfortunately, the double-bubble sign may not be present during the second trimester screening scan for earlier diagnosis of these cases.

Obstruction can also occur in other parts of the foetal bowel. Small bowel obstruction is more commonly detected than large bowel obstruction during the antenatal period, as it is more easily recognised. The foetal colon has a wide variation in size compared to small bowel, and this makes detection harder. Small bowel obstruction is suspected when long loops of distended bowel are seen. The upper limits for small bowel measurements are 7 mm for its lumen, and 15 mm for its length [14]. Polyhydramnios is more commonly seen in cases of obstruction in the proximal small bowel. The commonest cause of obstruction is intestinal atresia resulting from an ischaemic event. Uncommon causes include meconium ileus due to cystic fibrosis, and acute events such as volvulus.

Small bowel obstruction should be differentiated from hydronephrosis and multicystic dysplastic kidney which may have similar appearances. Hydronephrosis may be traced to their origins from the kidneys. Dilated bowel loops are distinguished from multicystic dysplastic kidneys by their tubular configuration. Bowel perforation in-utero is a known complication of small bowel obstruction. It may give rise to peritoneal calcifications and meconium pseudocysts.



Figure 43.27: Large omphalocele. US scan shows a membrane covering the herniated abdominal contents (arrows). (ST: stomach, ABD: abdomen, S: spine).

### Abdominal wall defects

The two most commonly detected abdominal wall defects are omphalocele and gastroschisis. The omphalocele is differentiated from gastroschisis by the presence of a membrane covering the herniated structures and the insertion of cord vessels into the sac. The herniated structures include bowel, liver and occasionally, other structures (Fig. 43.27). In gastroschisis, only the small bowel is herniated, and occur adjacent to the cord insertion. Omphaloceles are associated with increased risk of chromosomal anomalies and other congenital malformations, unlike gastroschisis where the complications are usually confined to the herniated bowel. The diagnosis should be only made after the 12th week of pregnancy to avoid misdiagnosis of the physiological midgut herniation that occurs between the 8th and 12th weeks.

### Cystic abdominal masses

The differential diagnoses of cystic masses in the foetal abdomen include choledochal, ovarian, mesenteric, duplication, urachal, hepatic and renal cysts. Conditions such as pelvi-ureteric junction obstruction, and the “double-bubble” of duodenal obstruction may also produce cyst-like structures.



Figure 43.28: US scan shows bilateral hydronephroses (K) due to bilateral pelvi-ureteric junction obstructions.

The masses may be distinguished by their location in the abdomen. Choledochal cysts are usually seen in the region of the porta hepatis. Ovarian cysts usually present in the lower abdomen of female foetuses. Renal cysts are located posteriorly, close to the spine.

## GENITOURINARY ANOMALIES

Antenatally-diagnosed renal anomalies include:

- Hydronephrosis
- Multicystic dysplastic kidney
- Renal agenesis
- Renal ectopia

### Hydronephrosis

Dilatation of the urinary tract may result from obstruction, vesico-ureteric reflux or an intrinsically-dilated system (primary megaureter). Obstruction can occur at the pelvi-ureteric junction (Fig. 43.28), the vesicoureteric junction, or bladder outlet (e.g. posterior urethral valves in male foetuses). In the case of bladder outlet obstruction, there are usually bilaterally-dilated urinary tracts. Urinary ascites may complicate severe obstructions. Oligohydramnios is an ominous sign. Apart from the risk of pulmonary hypoplasia, it is also an indication of possible renal impairment, or a very severe obstruction of the bladder outlet. It is not possible

to distinguish between distal ureteric obstruction and vesicoureteric reflux. The foetal renal pelvis is visible on most mid-trimester screening US scans. Numerous studies have been performed to determine the significance of various renal pelvic dimensions. In order to detect all cases of abnormal renal pelvic dilatation, some workers have recommended further follow-up or investigation of foetuses with anteroposterior renal pelvic diameters of 4 mm or more [15,16] found during the mid-trimester screening examination.

### Multicystic dysplastic kidney

Multicystic dysplastic kidneys are due to severe early urinary tract obstruction, and are commonly the result of pelvi-ureteric atresia. Contralateral renal anomalies may occur. They include contralateral hydronephrosis, renal agenesis, and dysplasia. Bilateral non-functioning kidneys may be suggested by poor amniotic fluid volume, non-visualisation of the urinary bladder, and its lack of distension in the foetus over time. Bilateral renal disease and absence of amniotic fluid when seen together usual-



Figure 43.29: Transverse US scan shows bilateral multicystic dysplastic kidneys (arrowheads). The renal parenchyma is replaced by tissue containing numerous cysts that do not connect. Note the absence of amniotic fluid. (P: placenta, S: spine).



Figure 43.30: US scan shows an ectopic right kidney (between cursors) sited next to the urinary bladder (B).

ly indicate a lethal condition, i.e. the foetus will probably not survive outside the uterus. The typical appearance of a multicystic dysplastic kidney consists of a multiloculated cystic structure in which the cysts do not interconnect (Fig. 43.29). The sizes of the cysts vary, and may change with gestation. The changes may be due to enlarging or diminishing sized cysts. The cysts may also resolve completely, leaving an empty renal bed.

### Renal agenesis and ectopia

Bilateral renal agenesis is a lethal condition. Absence of the kidneys may be suspected in cases that present with low or absent amniotic fluid, and non-visualisation of the foetal urinary bladder. Adrenal hypertrophy is known to occur in such cases [17]. The adrenal glands may also be mistaken for the kidneys. Such changes can also occur in the ipsilateral adrenal gland in unilateral renal agenesis or ectopic kidney (Fig. 43.30).

### SKELETAL DYSPLASIAS

Ultrasonographical diagnosis of skeletal dysplasia in the foetus involves evaluation of the following areas:

1. Measurement and assessment of the long bones. Use of appropriate normograms for the population studied is essential due to variations in

bone length among the different ethnic groups. Although limb shortening may manifest in the early part of the second trimester, most cases, including lethal forms of dwarfism, are more accurately determined around the 20th week. The bone shortening found in heterozygous achondroplasia, however, is not evident before 24 weeks.

2. Thoracic diameters, configuration of the thoracic cage, shape and length of the ribs. The presence of a small thorax increases the likelihood of pulmonary hypoplasia and a lethal outcome.
3. Assessment of the cranium and spine. Poor mineralisation of the foetal cranium may be evident in osteogenesis imperfecta, congenital hypophosphatasia, and some cases of achondrogenesis. The poorly-mineralised skull results in unusual clarity of the intracranial structures in these cases.
4. Polydactyly. This may be seen in short-rib polydactyly syndrome, chondroectodermal dysplasia, and asphyxiating thoracic dysplasia.
5. Amniotic fluid volume. Polyhydramnios is usually seen in association with the more severe forms of skeletal dysplasias that result in small thoraces. This finding is useful for distinguishing skeletal dysplasia from intrauterine growth retardation.

The examination should also include the assessment of the other organ systems for associated anomalies in certain syndromes. Additional information, including the family history, should be available during the diagnostic work-up. As the antenatal US scans are unable to provide the definitive diagnosis of the skeletal anomaly in most cases, further evaluation after delivery or termination is necessary. This includes genetic assessment, skeletal survey and in the case of terminations, histological assessment. Most cases are managed on the basis of whether the condition is lethal or non-lethal.

Additional considerations for non-lethal cases include associated malformations and an expected degree of morbidity.

### Lethal dysplasias

The following are the more commonly detected types of lethal dysplasias:

- Thanatophoric dysplasia (Fig. 43.31)
- Achondrogenesis
- Type IIA osteogenesis imperfecta
- Congenital hypophosphatasia (Fig. 43.32)
- Homozygous achondroplasia

### Non-lethal dysplasia

Heterozygous achondroplasia is the best known non-lethal form of skeletal dysplasia. This abnormality is not detectable on the second trimester screening US scan, as the shortening of the long bones usually presents during the third trimester.



Figure 43.32: Transverse US scan shows a poorly-mineralised skull, resulting in clear visualisation of intracranial structures in a case of congenital hypophosphatasia.



Figure 43.31: Thanatophoric dysplasia. (A) Longitudinal US scan shows a markedly shortened femur (between cursors). (B) Longitudinal US of the trunk shows a narrowed foetal thorax (C) due to thanatophoric dysplasia. (H: head, A: abdomen).



Figure 43.33: Placenta praevia major. US scan shows the placenta (P) overlying the internal os of the cervix (CX). Foetus is in a cephalic presentation with head (H) seen just above the placenta.

The less severely affected types of osteogenesis imperfecta may present with some in-utero anomalies such as bowing of the femurs. These usually non-lethal dysplasias are:

- Chondroectodermal dysplasia
- Diastrophic dysplasia
- Spondyloepiphyseal dysplasia congenita

Although this group of skeletal dysplasias is often non-lethal, they may sometimes be associated with other anomalies that are serious, and sometimes lethal.

## MISCELLANEOUS CONDITIONS

### Placenta

The more commonly encountered abnormalities of the placenta include abnormal placental location (placenta praevia) (Fig. 43.33), abnormal shape, abnormal adherence to the uterine wall (placenta accreta), placental abruption, haematoma (Fig. 43.34) and placental tumours (e.g. chorioangioma). Abnormal thickening of the placenta may reflect underlying systemic conditions affecting the foetus such as hydrops, viral infections and diabetes. A common feature of placenta is that of hypoechoic areas found in the subchorionic layer or within the placental substance. These hypoechoic areas are generally not clinically significant [18].

### Amniotic fluid

The source of amniotic fluid changes with the gestational age of the pregnancy. During the first trimester, the fluid is produced by the foetal membranes and foetal skin. By mid-trimester, most of the fluid is produced by the foetal kidneys. Fluid volume is influenced by foetal swallowing during the third trimester. Obstruction in the proximal part of the foetal gastrointestinal tract results in polyhydramnios. Changes in amniotic fluid volume reflect the balance between production and removal.

There are several methods for assessing amniotic fluid volume. These include subjective assess-



Figure 43.34: US scan shows a subchorionic haematoma (between cursors) seen as a hypoechoic mass on the uterine wall.

ment, measurement of single deepest pocket of fluid, and a semi-quantitative technique of using the four-quadrant measurement of the largest free pocket of fluid in each quadrant, and adding up the measurements. Each technique has its limitations. Experience is necessary for accurate subjective assessment. While the methods involve measurements that may seem accurate, foetal size, position and movement can influence distribution of fluid and affect the measurements. For single deepest pocket measurement, oligohydramnios is suspected if the deepest pocket obtained measures less than 2 cm. Measurements of more than 8 cm indicate polyhydramnios. In the four-quadrant technique, the normal range is 8 cm to 20 cm.

## CONCLUSION

Obstetrical US provides the attending obstetrician with essential information for the management of a pregnancy. Based on findings obtained, deci-

sions are made regarding further tests e.g. amniocentesis, mode of delivery, termination of pregnancy in the case of lethal anomalies, and counselling for subsequent pregnancies. For more detailed accounts of the various topics on imaging of the foetus and obstetrical problems, the reader should refer to dedicated texts on the subject of obstetrical and gynaecological US.

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