Ultrasound has permanently changed imaging in perinatology, increasing expectations of success and improving outcomes in maternal and neonatal care. The method is safe, real time, relatively inexpensive, and readily available in hospitals and clinics worldwide. \(^1\) Fetal imaging is the most common ultrasound study. Identifying anomalies related to fetal and placental development or dating errors is crucial. Sonographic genetic screening, cervical assessment, Doppler vascular measurements, and three-dimensional (3D) examinations have evolved from investigational techniques to accepted diagnostic tools. In the past two decades, magnetic resonance imaging (MRI) has joined sonography for the prenatal diagnosis of a broad spectrum of disorders.

### Ultrasound Equipment

In an ultrasound exam, a transducer (probe) is placed directly on the skin, inside the vagina or other body cavity, or directly on the organ of interest intraoperatively. A thin layer of water-based gel acts as a coupling agent to potentiate sound wave transmission.

Obstetric studies may be performed by either the transabdominal or transvaginal approach, using transducers of the appropriate frequency, usually between 1.5 and 10 MHz. Choice of transducer is a trade-off between penetration with lower frequencies (essential in the obese patient) and resolution at the higher end (required for the smallest fetal structures).

### Fetal Imaging Techniques

Real-time ultrasound, in which image brightness varies with the intensity of returning signals (B-mode), is the standard method of fetal imaging (Fig. 11.1). It is required for confirmation of cardiac activity and fetal movement in living gestations. Brief signal bursts are followed by relatively long receptive intervals (1:10 ratios or greater); lower signal frequencies encounter less interference but reflect from fewer informative interfaces. Image quality varies with distance to the target, structure size, movement relative to the signal, and tissue transmission characteristics. Ideally, the transducer is close to its target; the transvaginal approach is preferable in early gestation for cervical studies and for gynecologic investigations. Suboptimal images are common with obesity, limited fluid interfaces, intervening structures, gas-filled viscera, scar tissue, and poor positioning relative to the sound beam. Diagnostic difficulty arises when different materials have similar echo characteristics, as do blood, urine, ascites, and the contents of many cysts.

M-mode ultrasound is a direct representation of beam reflection by moving edges (e.g., in cardiovascular imaging). Interpretation requires standardized, often hard-to-achieve, stable views. M-mode is useful in assessing arrhythmias, myocardial contractility, and pericardial effusions. M-mode “snapshots” efficiently document cardiac activity and rate (Fig. 11.2).

Doppler ultrasound uses the frequency shift that occurs when sound beams are reflected off moving objects to demonstrate the presence, velocity, and direction of blood flow (Fig. 11.3). Direct calculations from narrow, tortuous fetal and uterine vessels lack accuracy; to compensate, prenatal Doppler findings ideally are obtained at beam angles less than 35 degrees relative to umbilical vessels and 15 degrees for middle cerebral arteries. The flow indices are generally expressed as ratios, relative to median values. Color Doppler semiquantitatively assigns direction to blood flow; by convention, warm colors (red) denote movement toward the transducer, and saturation is keyed to velocity. Color Doppler illuminates cardiac, arterial, and venous structures (Figs. 11.4 and 11.5). Color Doppler energy (power Doppler) reflects signal intensity; amplitude corresponds to blood cell motion. Power Doppler is sensitive to very low flow and effective independent of angulation; it is helpful for mapping vascular beds and for quick visualization of any fetal vessel (Figs. 11.6 and 11.7).

Three-dimensional ultrasound analyzes returning echoes along a third axis. Images are manipulated electronically to render surfaces and volumes from multiple perspectives, both as static and real-time (“four-dimensional”) views. Surface rendering of subtle organ details enhance detection of anomalies, partially overcoming positional limits of standard scans and permitting a comprehensive review of fetal organs and skeleton (Fig. 11.8). Three-dimensional studies have improved volume calculations, facilitated analysis of complex spatial relationships, and have better explained...
Abstract

The field of perinatal imaging and diagnosis has been exponentially growing for the last 30 years using both ultrasound and magnetic resonance imaging (MRI). It has revolutionized diagnosis of fetal anomalies in different organ systems, enables fetal well-being assessment, accurately determines gestational age, and uncovers potential uterine, placental, and ovarian disorders that might negatively affect maternal and fetal outcome. Apart from imaging, other techniques, for example, DNA analysis from chorionic villus samples, amniocentesis, and free fetal DNA circulating in the maternal blood can be used to diagnose specified fetal anomalies. The aim of this chapter is to share with pediatrician and neonatologist the current modalities of prenatal imaging as well as its limitations. The chapter includes descriptions of abnormalities paired with their sonographic images. The template includes a brief description of the anomaly, the relevant findings, and information regarding management and prognosis.

Keywords

fetal imaging
ultrasound
magnetic resonance imaging (MRI)
prenatal diagnosis
fetal anomalies
• Fig. 11.1 Transabdominal B-mode, two-dimensional scan. Profile of 20-week fetus.

• Fig. 11.2 Transvaginal M-mode demonstration of embryonic cardiac activity at 5 5/7 weeks’ gestation. Upper frame: Embryo with cursor across thorax. Lower frame: M-mode display of wall movement during two cardiac cycles, 114 beats per minute (between vertical lines).

• Fig. 11.3 Upper frame: Color Doppler highlights a segment of the umbilical cord (trapezoid). The gate (transverse parallel lines) identifies the sampling site within the umbilical artery. Lower frame: Pulse Doppler waveform recorded from the umbilical artery.

• Fig. 11.4 Color flow Doppler demonstration of a right pelvic kidney, descending aorta, and inferior vena cava in a 28-week fetus. Flow toward the transducer is the color of the upper bar; flow away corresponds to the lower bar colors.

• Fig. 11.5 Transverse view of fetal pelvis, umbilical cord (UC), and urinary bladder (UB) with color Doppler showing bifurcation of the umbilical arteries (UA).

• Fig. 11.6 Color Doppler demonstration of the anterior cerebral, pericar- dal, and vertebral arteries (thin, first thick, and second thick arrows). Lower velocities are shown as more saturated hues. Turbulent flow and flow more rapid than the scale parameters demonstrate aliasing (mixture of colors). Compare with Fig. 11.7, power Doppler of same structures.
The Fetus

MRI is often used to elucidate problems initially identified by ultrasound examinations. Images are acquired in the axial, coronal, and sagittal planes relative to the fetus or orthogonal to the maternal pelvis. Gadolinium is placentally transferred and is contraindicated during pregnancy because of its exceptional persistence in tissue and known potential for adult renal injury. Its use rarely may be justified for assessment of placenta accreta or serious maternal disease.

Bioeffects and Safety

As part of the Food and Drug Administration’s (FDA’s) initiative to reduce unnecessary radiation exposure from medical imaging, health care providers are advised to consider techniques with little or no ionizing radiation, for example, ultrasound or MRI, and only if medically appropriate.

Diagnostic ultrasound energy has the potential for affecting tissues (bioeffects); two recognized mechanisms are heating and cavitation. Data collected during routine studies show that “gray-scale” B-mode ultrasound is associated with a negligible rise in temperature. To date, there has been no convincing evidence of harm to the human fetus.

By definition, ultrasound is inaudible to humans; moreover, oncogenic effects have not been identified. At nonclinical levels, ultrasound energy causes cell lysis, intracellular shearing, streaming effects, altered membrane permeability, and abnormal chromosome function. Heat exposure triples with each change in modality: from 2- or 3D, B-mode to M-mode, then color flow, before peaking during pulsed Doppler. Harmful levels should not be attained routinely but might occur during focal cranial pulsed Doppler.
interrogation or in a febrile patient, without unusually long exposures. Mechanical disruption from cavitating gas bubbles is improbable in the fetus. Both temperature and disruption risks are now displayed on equipment (Fig. 11.10); thermal index (TI), a ratio between transducer output and the energy needed to warm up tissue temperature by 1°C, with a desired value below 2, is also categorized by tissue type: TI soft tissues, TI cranial structures, and TI bone. Mechanical index (MI) references pulse amplitude effects of compression and decompression, ideally maintained below 0.4 in fetal studies. Publication standards now usually require that these indices be displayed on submitted images. For safety, only medically essential examinations should be performed; settings and duration should be the minimum required to achieve adequate views.

Strong magnetic fields and radiofrequency waves are used in MRI with no known harmful effects, but large longitudinal studies are lacking. As with ultrasound, heat delivery to the fetus is a recognized hazard; in MRI, however, the maternal surface receives the greater thermal exposure. Noise from the magnetic coils (up to 120 dB) is, in theory, capable of causing acute hearing damage; fortunately, maternal tissue attenuation decreases fetal intensities by 25% to relatively safe levels. Direct magnetic bioeffects remain unproven; the FDA states that safety to the fetus “has not been established.”

The FDA has recommended that health care providers attempt to minimize exposure while maintaining diagnostic quality when using ultrasound. The lowest possible ultrasound exposure settings that obtain adequate image quality and gain the necessary diagnostic information should be used, following the as-low-as-reasonably-achievable (ALARA) principle. Spectral or “flow” Doppler should not routinely be used to “auscultate” the fetal heart rate in the first trimester because of its higher energy delivery; instead, adequate documentation of viability can be obtained with use of M-mode or conventional two-dimensional real-time ultrasonography with video or cine archiving.

Who Should Perform Fetal Ultrasound Examination?

Evaluating the unborn fetus is an enormous responsibility. A true hazard of performing obstetrical ultrasound studies lies in the potential for error in image interpretation. Either under- or over-diagnosis may trigger a chain of events with tragic consequences. Some radiologists who perform ultrasound studies may have had relatively limited exposure during training to obstetrical imaging; in contrast, obstetrical services may offer extensive bedside experience but fewer didactic hours, particularly for complex cases. As a result, many practitioners prefer to delegate the task of scanning to sonographers, themselves of variable competencies, choosing instead to emphasize image interpretation and case management.

All practitioners who perform or supervise the performance of obstetric ultrasonography should be licensed medical practitioners, with specific training in obstetric ultrasonography; this is especially necessary when providing specialized obstetric ultrasound examinations. Certification programs, continuing education and credentialing for both sonologists and the interpreting sonologists, are important steps toward improving safety and diagnostic accuracy. To ensure the highest quality and accuracy in interpretation of obstetric imaging, the American Institute of Ultrasound in Medicine and the American College of Radiology offer ultrasound facility accreditation. The process involves a review of submitted case studies, practice volume, equipment use and maintenance, report generation, image storage, and qualifications of all providers. Practices, not individuals, are accredited for obstetrics, gynecology, or both; practices maintaining ultrasound accreditation have been shown to demonstrate improved compliance with published standards and guidelines for the performance of ultrasound examinations.

Ethical Considerations

The education of physician sonologists encompasses visual recognition and interpretative tasks common to diagnostic imaging but also demands mastery of specific mechanical skills (or, at least, an ability to assess the latter in sonographers). Both ultrasound and MRI have rapidly evolved; clinically relevant frontiers are often explored by collaborators with pooled data, prerelease technology, and funding for staff and statistical analysis. Nuchal translucency measurement, a deceptively simple sonographic method for aneuploidy screening, provides a cautionary example (Fig. 11.11). Its orderly dissemination, including certification courses, ongoing audits, professional society, and laboratory coordination, stands in sharp contrast to the viral dissemination of most techniques, yet erosion of competency, once achieved, remains a concern.
businesses with the impetus for rapid expansion. A number of ethical issues are raised, including conflicts of interest for the commercial enterprise, the fetus, and the parents with respect to long-term effects. Current epidemiologic evidence is not synchronous with advancing ultrasound technology; a lack of evidence of harm is not the same as lack of harm. Applying four major theories of ethics and principles (the precautionary principle, theories of consequentialism and impartiality, duty-based theory, and rights-based theories) leads to the conclusion that obstetric ultrasound practice is ethical only if the indication for use is based on medical evidence, rendering “keepsake studies” ethically unjustifiable.

Multiple gestations may result in a number of ethical dilemmas. Advances and regulation in assisted reproductive technology (ART) have decreased the incidence of multifetal pregnancies, but fetal reduction remains a painful choice for parents facing the prospect of extreme prematurity in higher order multiples. Management of twin–twin transfusion syndrome (TTTS), anomalous co-twins, discordant growth, or distress far from term also necessitates choosing among unsatisfactory alternatives. An excellent review of the psychosocial consequences and the ethical issues associated with selective termination of pregnancy has been published.25

Non-diagnostic studies, varying prognoses for a given diagnosis, and the inherent limitations of ultrasound and MRI studies lead to ethical issues in management and counseling. Patients and physicians alike may share unrealistic expectations for the predictive accuracy of targeted diagnoses.

Anomalies and variants linked to Down syndrome and other serious conditions (sonographic “markers”) identified during routine studies present patients and caregivers with unanticipated, unwelcome options, particularly if patients had previously explicitly refused serum screening or direct genetic testing.19 Ideally, informed consent discussions addressing risks, benefits, consequences, alternative strategies, and limitations of ultrasound and MRI should be provided to all patients before performing such imaging. Given the irreversible nature of both birth and abortion, prospective parents must ultimately judge for themselves their tolerance for uncertainty in diagnosis and for imperfection in their offspring.

**Classification of Fetal Sonographic Examinations**

**A. First-Trimester Examination**

First trimester transvaginal scans exclude ectopic implantation by demonstrating an intrauterine asymmetric or “double sac” gestational sac with yolk sac or embryo, ideally with visible cardiac activity; identifiable embryos in the fallopian tubes are uncommon. Heterotopic pregnancy occurs in less than 0.1% of spontaneous conceptions, leading to the pragmatic conclusion that finding an intrauterine pregnancy excludes an ectopic one, except after assisted reproductive technic (ART). Nevertheless, careful study of the

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**Fig. 11.11** Nuchal lucency measurement (calipers) in a 13-week fetus. The nasal bone is visible (down arrow), and the amnion (up arrow) is clearly distinguished from the skin fold.

**Fig. 11.12** Vulvar skin folds (arrow) of normal female fetus in the midtrimester.
adnexa, ovaries, and cervix is generally prudent. A “double sac” is usually seen transvaginally at levels of 1000-1500 international units of human chorionic gonadotropin before 5 1/2 menstrual weeks (see Fig. 11.10); but serum human chorionic gonadotropin (hCG) and visualization thresholds are variable. Finding a yolk sac or embryo confirms an hCG and visualization thresholds 5 1/2 menstrual weeks (see Fig. 11.10); but serum human international units of human chorionic gonadotropin before sac” is usually seen transvaginally at levels of 1000-1500 adnexa, ovaries, and cervix is generally prudent. A “double in younger women), the associated risk of miscarriages, over 35 years old, thereby missing abnormal pregnancies need for invasive testing (previously offered to every woman screening for early detection of abnormalities decreases the number of entities accurately (Fig. 11.13). First-trimester provisional; however, early fetal period scans diagnose a failed pregnancy until well past the accepted normal thresholds for appearance and progressive development of these structures.

Embryonic heart rates, slower at initiation, increase to more than 160 beats per minute (bpm) by the 9th week, declining slightly through the 13th week. Persistent rates below 100 bpm have been linked to risk for missed abortion, aneuploidy, and anomalies.15 When an embryonic heart rate (below 80 bpm) is detected between 6 and 7 weeks, a first-trimester demise is anticipated in approximately 25%, even if the rate subsequently normalizes. In such pregnancies, a follow-up scan is warranted. Prior to 6 weeks, an embryonic heart rate below 100 bpm is not necessarily indicative of a poor prognosis. Likelihood of survival into the second trimester is also significantly higher when there is concordance between biometrically calculated gestational age and menstrual dating.3

Embryonic anatomic surveys are limited and necessarily provisional; however, early fetal period scans diagnose a number of entities accurately (Fig. 11.13). First-trimester screening for early detection of abnormalities decreases the need for invasive testing (previously offered to every woman over 35 years old, thereby missing abnormal pregnancies in younger women), the associated risk of miscarriages, and enabled first-trimester invasive testing and diagnosis in high-risk pregnancies using chorionic villi sampling rather than a second-trimester amniocentesis. It also allowed the safer and more private option of first-trimester termination of an abnormal pregnancy before the pregnancy was visible, decreased anxiety, and provided reassurance about pregnancy well-being in both high-risk and low-risk situations. Progress in the field of first-trimester sonography continues in two intertwined directions: One is finding additional sonographic markers that increased the accuracy and sensitivity of detecting chromosomal abnormalities, especially Down syndrome. The other is improving the early detection of congenital anomalies. Midtrimester confirmation continues to be prudent for the majority of first-trimester findings (see Fig. 11.13). Later studies retain advantages with respect to the natural history of many anomalies and for visualization of heart, spine, and other problematic structures.

B. Standard Second- or Third-Trimester Examination
A standard second- or third-trimester sonogram includes an evaluation of fetal number, presentation, cardiac activity, fetal biometry, amniotic fluid and placental characteristics, and fetal organ survey. Second trimester transvaginal cervical measurement is encouraged for patients at risk for prematurity; however, the role of universal screening remains controversial. Examination of the maternal pelvic structures is otherwise performed transabdominally at this time, if feasible.

C. Limited Examination
A limited examination is performed to investigate a specific concern, usually under exigent circumstances. For example, a limited examination might identify fetal cardiac activity in the presence of bleeding or confirm presentation in early labor. Abbreviated sonographic studies are more acceptable when there has been a prior complete study; pragmatically, a full examination should be documented once the acute situation has stabilized for ongoing pregnancies.

D. Specialized Examinations
A detailed “targeted” anatomic examination is performed when an anomaly is suspected on the basis of history, biochemical or genetic results, or findings on prior scans. Other specialized examinations include fetal Doppler ultrasound, 3D imaging, biophysical profile, fetal echocardiogram, detailed neurosonography, and additional biometry or evaluation of organs not usually imaged on routine studies.

Applications of Ultrasound Studies

Genetic Screening

Genetic screening combines ultrasound study and biochemical testing to enhance the detection of chromosomal abnormalities. This approach has resulted in greater scrutiny of younger patients and less frequent age-based invasive

*Fig. 11.13* Eleven-week embryo with a thickened nuchal lucency and an omphalocele (arrow) containing fetal liver. The fetal liver does not undergo physiologic herniation. A study at 18 weeks identified a lumbarosacral spina bifida in addition to confirming the omphalocele.
Presently, noninvasive screening for fetal aneuploidy (trisomies 13, 18, 21) is encouraged for all low-risk patients. Common noninvasive screening options include: (1) first-trimester screening (nuchal translucency measurement and maternal serum biochemical marker algorithm), (2) first- and second-trimester cell-free fetal DNA fragment analysis from maternal blood, (3) second-trimester serum screening (maternal age and serum biochemical marker algorithm), or (4) two-step integrated screening, which includes first- and second-trimester serum screening with or without nuchal translucency (integrated prenatal screen, serum integrated prenatal screening only, contingent and sequential screening variations). Different algorithms noticeably affect sensitivity, specificity, and predictive values; cost or convenience may factor in choosing a strategy. Analysis of cell-free fetal DNA, found at concentrations almost 25 times higher than those from intact nucleated fetal blood cells extracted from similar volumes of maternal blood. Cell-free fetal DNA (cfDNA) noninvasive prenatal screening is now in wide use for specific targeted chromosomal abnormalities, especially trisomy (an extra copy of a chromosome) or monosomy (a missing chromosome), and numerous commercial products are currently marketed for this indication. Patient acceptance has been high, perhaps in part because of early identification of fetal sex. Insufficient fetal fractions and indeterminate results appear more frequently in obese patients but may also occur with chromosomal abnormalities. Direct prenatal diagnosis by chorionic villus biopsy or amniocentesis should be routinely recommended for at-risk individuals on the basis of age, family history, abnormal screening, or ultrasound findings.

One-third of fetuses with Down syndrome will have anomalies, characteristically endocardial cushion defects (Fig. 11.14), duodenal atresia (Fig. 11.15), and more subtly, small atrioseptal and ventriculoseptal cardiac defects (Fig. 11.16). Two-thirds may have second-trimester sonographic markers: for example, increased nuchal lucency, hypoplastic or absent nasal bones, and abnormal cardiovascular Doppler patterns in the first trimester, thickened nuchal fold (Fig. 11.17) and nasal hypoplasia, ventriculomegaly, choroid cysts (Fig. 11.18), hypoplasia of the fifth digit, decreased long bone ratios, enhanced echogenicity of pampillary muscles and bowel, and renal pyelectasis (Fig. 11.19). The predictive value of screening components is affected by ethnicity, habitus, maternal diet, and fetal sex, as well as by device and operator-dependent detection rates. The permutations may confound counselors attempting to elucidate (and patients trying to grasp) the difference between screening and diagnosis, and basic descriptions of risks...
and benefits. Recently described first-trimester evaluation of the posterior brain (intracranial translucency) provides an additional screening tool for open neural tube defects and other intracranial abnormalities (Fig. 11.20A and B).\textsuperscript{21} Second-trimester sonographic follow-up and genetic evaluation may add critical information to the assessment of these abnormal findings. First-trimester identification of tricuspid regurgitation and increased ductus venosus resistance seems a promising, albeit technically challenging, addition to early screening protocols.\textsuperscript{26}

**Assisted Reproduction**

Ultrasound is essential for timing and guiding oocyte retrieval and helpful in embryo transfer; its role in judging
endometrial receptivity is less clear. Saline ultrasound studies prior to fertility treatments routinely complement or replace hystero-salpingogram in assessment of the uterus and adnexa. Use of MRI may play an expanded role in structural and functional evaluation in the future.

Assisted reproductive technology results in more twins and higher-order multiples; early ultrasound study of embryos, amnioncity, and chronicity is essential to subsequent management. With the increasing popularity of ART, obstetricians and radiologists are more likely to encounter associated complications, especially in an emergency setting. These complications include ovarian hyperstimulation or torsion, ectopic or heterotopic pregnancy, and pregnancies of unknown location or undetermined viability. Ovarian hyperstimulation syndrome may occur following ovulation induction or ovarian stimulation and is characterized by bilateral ovarian enlargement, by multiple cysts, and third-spacing of fluids (Fig. 11.21). Additional clinical findings may range from gastrointestinal discomfort to life-threatening renal failure and coagulopathy. Ovarian torsion should be excluded in any woman undergoing ART who presents with severe abdominal pain. Ectopic pregnancy resulting from ART has a relatively increased frequency of rarer and more lethal forms, including interstitial and cervical locations. Heterotopic pregnancies, simultaneous intrauterine and ectopic implantations, are more common in ART patients.

Ultrasoundography is the first-line choice for identifying ART complications, although lack of specific symptoms may first trigger other approaches. Familiarity with characteristic strengths and limitations of these and other techniques will facilitate accurate, timely diagnosis and avert potentially serious consequences.

**Multiple Gestations**

Multiple gestation accounts for about 3% of all pregnancies. With an increase in the number of fetuses, scans become more complex, time consuming, and error prone; additionally, determination of zygosity is essential. The management of anomalous, discordant, or moribund co-twins differs significantly based on chorionicity (Fig. 11.22).

Monochorionic twins occur with a relatively constant frequency (1:250 pregnancies), unlike dichorionic twinning influenced by race, heredity, maternal age, parity, and ART. Ultrasound assignment of chorionicity is most accurate for different-sexed dizygotic twins, but by evaluating sac appearance in early gestation, approaches this accuracy in gender-concordant pairs. Successful identification may occur throughout gestation by examining the dividing membranes at their placental origin. Dichorionic diamniotic twins are usually (95%) dizygotic, with independent risks for anomalies and placental malfunction. Monochorionic pairs are predictably monozygotic; attrition rates exceed 30% from early abortion, anomalies, and prematurity. Matched and isolated anomalies are both more common in monochorionic gestations; because of shared vasculature, loss of a co-twin may kill its sibling outright or produce severe neurologic damage in up to one-third of survivors.

Monochorionic TTTS, more common in females, is characterized by unbalanced, shared perfusion that restricts
growth and amniotic fluid production in the donor and causes volume overload, cardiac dysfunction, and polyhydramnios in the recipient. Ultrasound staging has been used to time and to guide a variety of vascular ablation strategies. Serial amniocentesis may be helpful in milder cases. Management by these approaches has been modestly effective in decreasing stillbirths and prematurity in TTTS. Monoamniotic twins rarely experience TTTS but routinely encounter cord entanglements, resulting in high lethality rate of both fetuses (Fig. 11.23). Management usually consists of a scheduled preterm cesarean prior to onset of labor. For all multiple gestations, serial ultrasound monitoring of growth, well-being, placental performance, and cervical length is common practice.

Twin reversed arterial perfusion (TRAP) sequence is another complication of monochorionic twinning, complicating approximately 1% of monozygotic pregnancies. Placentation among TRAP cases has been predominantly reported to be monochorionic diamniotic and to a lesser degree monochorionic monoamniotic twins. The proposed pathogenesis is the association of paired artery-to-artery and vein-to-vein anastomoses through the placenta combined with delayed cardiac function of one of the twins early in pregnancy. This situation allows blood pumped from the healthy twin “pump twin” to perfuse retrogradely the heart of the other twin, also known as the “acardiac” twin or “parabiotic twin.” Thus, flow in the artery and vein are

![Fig. 11.22 Diagram (left) of variations in appearance of the dividing membrane for dichorionic, monochorionic, and monoamniotic twins. Upper-right image shows typical thick first-trimester appearance of the chorion (arrow) in dichorionic diamniotic twins. Lower image shows the peaked dividing membrane (arrow) of dichorionic twins in the latter half of pregnancy.](image1)

![Fig. 11.23 Monochorionic monoamniotic twins with entangled umbilical cords at 10 weeks’ gestation.](image2)
reversed in the umbilical cord of the acardiac twin, giving rise to the acronym TRAP. Retrograde perfusion interferes with normal cardiac development, which rarely goes beyond the stage of tubular heart. Common abnormal findings in the “acardiac twin” include impaired or absent development of the cephalic pole, rudimentary or absent heart, abnormal or absent upper limbs, relative preservation of the lower limbs although clubbing and abnormal toes are common, abnormal viscera, and single umbilical artery. A common finding is massive edema around the upper body including the neck of the acardiac twin (Fig. 11.24A-C).

**Pregnancy Evaluation**

One of the most powerful applications of prenatal ultrasound is using biometrics to establish or confirm gestational age. Through 22 weeks of gestation, most genetically normal individuals cluster closely on normographic curves. Sonographic measurements of fetal ultrasound parameters are the basis for accurate determination of gestational age and detection of fetal growth abnormalities. It has been shown that a fixed error of about 8% (plus or minus) can be anticipated when determining gestational age by ultrasound, consistent with the observation that the earlier gestational age is determined, the lower the margin of error in days. If accelerated or restricted growth supervenes, however, biometric markers are generally compromised accordingly.

Selection of the most useful single parameter depends on the timing and purpose of measurement and is influenced by specific limitations. Commercial equipment has a variety of preinstalled normograms, biometry-based dating, and weight estimation formulae. Embryonic pole and crown-rump lengths are considered most precise for early dating of pregnancy, becoming more variable with fetal flexion effects at the end of the first trimester. Biparietal diameter (BPD), obtainable after parietal bone calcification in week 12, maintains the closest correlation with gestational age in the second trimester. BPD is measured from the outer to the inner table of the skull, perpendicular to the parietal bones and central falx cerebri. The proper plane contains the cavum septum pellucidum, thalami, third ventricle, and tentorial hiatus, within the bony table of a complete head circumference (Fig. 11.25). In cases of variation in the shape of the skull, a head circumference (HC) measurement obtained in the same plane may be an effective alternative. Microcephaly may be suspected when the HC measurement is more than 3 standard deviations (SD) below the mean but is rarely diagnosed prenatally. Additionally, antenatal sonographic estimation of HC is associated with significant underestimation compared with the actual postnatal HC. This discrepancy may have important clinical implications and should be taken into account in the interpretation of sonographically measured HC. BPD and HC are relatively spared in nutritional and perfusional disorders of growth, although cranial measurements may also be distorted by compression effects. The abdominal circumference (AC), measured along the outer margin of the abdominal skin line at the level of the gastric bubble and the intrahepatic portion of the umbilical vein at the bifurcation of the portal veins, is the best single predictor of growth aberrations but is less helpful for dating (Fig. 11.26). Femoral length (FL) is the fourth measurement commonly included in biometric
CHAPTER 11  Perinatal Ultrasound

Fetal Growth

Serial measurements over time are the best way to judge fetal growth. The sac and embryo grow perceptibly each day; by the second trimester, intervals of 2 or 3 weeks between studies are more reliable. Problems arise when attempting simultaneously to assign age and weight percentile without reliable dating. The abdominal circumference (AC) is a better indicator of decreased perfusion or increased glycogen storage than the cranial measures; ratios of AC to the HC and FL amplify differences but have poor sensitivity and specificity. Strategies to identify small-for-date fetuses have included risk panels, Doppler ratios, amniotic fluid volume, placental scores, and biometry-based weights; none have had completely satisfactory results, although outcomes appear to have improved.

Correctly identifying fetal growth abnormalities remains an elusive goal despite their major contribution to adverse perinatal outcomes and stillbirth. Current clinical screening relies on the symphysis–fundal height measurement, but fewer than 25% of small-for-gestational-age (SGA) infants will be identified using this methodology in a low-risk population. Routine third-trimester ultrasound study for growth assessment has a better detection rate, ranging from 50% to 80%, but the impact on perinatal outcome is unclear.

Several common findings, in addition to risk panels, underlying maternal diseases (particularly with vascular components) and prior stillbirth or growth-restricted outcomes may raise concern for fetal growth restriction. These include a modest size–dates discrepancy (within method error) on a first-trimester study, abnormal maternal serum levels of pregnancy-associated plasma protein-A (PAPP-A), and free β-human chorionic gonadotropin at 9-13 weeks, usually obtained as part of genetic screens; early elevation of maternal blood pressure and abnormal uterine artery resistance by Doppler at 11 0/7 to 13 6/7 weeks; a hypoglycemic response to glucose testing may be an early third-trimester observation. By combining uterine artery Doppler findings and baseline maternal characteristics, detection rates for early-onset fetal growth restriction have reached clinically acceptable levels. Unfortunately, fetal growth restriction developing later in pregnancy still goes largely undetected.

Placental Abnormalities

Placental conditions affecting the mother and/or fetus include gestational trophoblastic diseases, placental hematoma, chorangioma, abruptio, placenta previa, placenta accreta/increta/percreta, vasa previa, choriocarcinoma, chorioamnionitis, viral and parasitic villitis and placentalitis, decidual and thrombotic vasculopathies, infarction, polyps, and retained products of conception. Although gross inspection and histopathology provide the ultimate diagnosis, ultrasonography is the definitive prenatal modality for the evaluating the majority of these conditions. MRI
and computed tomography are infrequent adjunctive measures, with the latter occasionally helpful in tumor staging or trauma cases.\textsuperscript{31}

**Placental Location**

Placental location can be confidently established by abdominal and transvaginal ultrasound studies. Low-lying placentation, present in up to 60% of early second-trimester studies, persists as placenta previa in only 1% to 2% of patients at term (Fig. 11.27). Fetal vessels near the cervix can be visualized using color Doppler, facilitating the diagnosis of funic (umbilical cord) presentation and vasa previa (fetal vessels overlying the os) (Fig. 11.28). Normally inserted centrally in the placenta, the umbilical cord may later, as a result of asymmetric placental growth, be located marginally or even on adjacent membranes; in the latter position, traumatic lacerations, hemorrhage, and compression-linked heart rate changes are potential consequences.

Abnormally invasive placenta is a spectrum disorder encompassing the histopathologic diagnoses of placenta accreta (a small focus or more generalized muscular invasion), placenta increta (deeper myometrial invasion up to the uterine serosal layer), and placenta percreta (through the serosa to adjacent visceral or vascular structures). It is potentially life threatening, as forced removal of an abnormally invasive placenta can lead to catastrophic maternal hemorrhage; management of all but the most circumscribed lesions usually requires hysterectomy (Fig. 11.29A and B).

Invasive placentation was previously diagnosed only when failed attempts to remove the placenta were followed by massive bleeding. Placental accretion is significantly more likely in women with the combination of placenta previa and a history of one or more cesarean sections,\textsuperscript{36} after myomectomy or curettage, and with high parity.\textsuperscript{18} The frequency of accretion, now complicating 1/2500

\[ \text{Placental Location} \]

\[ \text{Placental Location} \]

\[ \text{Low-lying placenta, present in up to 60% of early second-trimester studies, persists as placenta previa in only 1% to 2% of patients at term (Fig. 11.27). Fetal vessels near the cervix can be visualized using color Doppler, facilitating the diagnosis of funic (umbilical cord) presentation and vasa previa (fetal vessels overlying the os) (Fig. 11.28). Normally inserted centrally in the placenta, the umbilical cord may later, as a result of asymmetric placental growth, be located marginally or even on adjacent membranes; in the latter position, traumatic lacerations, hemorrhage, and compression-linked heart rate changes are potential consequences.} \]
deliveries, has increased more than 10-fold in the past 20 years, echoing rising cesarean rates. Sonographic criteria for placenta accreta were developed using conventional gray-scale, 2D, and 3D color and power Doppler transabdominal and transvaginal ultrasonography. The sonographic characteristics identified are loss/irregularity of the echo-free “clear space” between the uterus and the placental basal plate; thinning or interruption of the hyperechoic interface between the uterine serosa and the bladder wall (increasing concern for percreta), and Doppler findings that included the presence of turbulent placental lacunae with high-velocity flow, as well as hypervascularity of the uterine serosa—bladder wall interface and irregular intraplacental vascularization. Diagnostic ultrasound findings of indistinct placental margins, attenuated myometrium, and large turbulent placental vessels (mainly veins) have sensitivity and specificity for accretion in the range of 85%. The presence or absence of the listed findings has been shown to be very helpful in diagnosing placental accretion and in differentiation of placenta accreta from percreta. In a recent study, the European Working Group on Abnormally Invasive Placenta sought to increase diagnostic capabilities of abnormally invasive placenta. The researchers have suggested a total of 10 sonographic findings detected by 2D gray-scale and color Doppler study to be very helpful in diagnosing an abnormally invasive placenta. Similar sonographic findings with 81% sensitivity and 98.9% specificity were recently reported.

Antepartum diagnosis of abnormal attachment permits multidisciplinary planning for prematurity management, anesthesia, transfusion, hemostatic and uterotonic medications, balloon tamponade, arterial embolization, or scheduled preterm (around 34 weeks) cesarean-hysterectomy prior to onset of labor (Fig. 11.30). MRI mapping is most helpful when there is posterior placentation, suspected lateral extension, after myomectomies, for evaluation of adjacent viscera with percreta, or when ultrasound findings are ambiguous.

The diagnosis of placental abruption remains a clinical one. The role of imaging in this disorder is to exclude placenta previa, an equally common source of severe third-trimester bleeding. Subchorionic hematomas are often noted on transvaginal scans early in gestation; symptomaticity, size, and persistence have been linked to poorer outcomes. Later abruptions are more difficult to visualize; acute bleeding is isoechoic with placenta and can be mistaken for placentomegaly. Hypoechoic fluid collections and hyperechoic infarcted areas appear in more chronic presentations.

Grading placental appearance to detect disturbed growth or maturation is of limited benefit. Persistent immaturity is linked to hydrops fetalis, although not as often as increased echogenicity and thickening. Precociously mature placentas may presage growth restriction (Fig. 11.31).

**Amniotic Fluid Volume**

Amniotic fluid is initially secreted by the amnion; by the 16th week of pregnancy, fetal renal production accounts for the majority, with nearly complete turnover during a 24-hour period. Malformations of the esophagus and upper gastrointestinal tract, inhibited fetal swallowing, aneuploidy, intermittent renal obstruction, maternal diabetes, twin–twin transfusion syndrome, some forms of dwarfisms, and fetal hydrops are associated with marked polyhydramnios. Severe growth restriction with polyhydramnios carries a poor prognosis. Polyhydramnios is idiopathic in almost half of the cases; associated complications include maternal respiratory compromise, premature membrane rupture, preterm labor and delivery, malposition, abruption, cord prolapse, preeclampsia, amniotic fluid embolism, and puerperal hemorrhage. Maximum vertical pocket or summed depths across quadrants conveniently serve as proxies for volume calculation in clinical settings (Fig. 11.32). Oligohydramnios may occur after membrane rupture; after fetal renal compensation for placental hypoperfusion; from functional or obstructive urogenital anomalies; with maternal dehydration; or following exposure to some medications, including indomethacin and angiotensin-converting-enzyme inhibitors. Maternal obesity is associated with underestimation of amniotic volumes. Amniotic fluid measurements may be
competency, and, rarely, torsions or ruptures of the horn in which the gestational sac is located. Poorly vascularized septations are etiologically associated with abruption and pregnancy failure. Myomas complicate 1% to 2% of pregnancies with more frequent cesareans and prematurity, abruption, degeneration, and fetal malpresentation; less common complications include fetal deformation, dysmaturity, puerperal hemorrhage, and hysterectomy. Adverse obstetric outcomes are rare; studies are confounded by age, ethnicity, and other differences in those who develop myomas. Generally good maternal and neonatal outcomes are expected in most pregnancies with uterine fibroids.

**Cervical Length and Pelvic Structures**

Cervical length and appearance is frequently assessed during routine antenatal ultrasound; transvaginal measurements (Fig. 11.33) are more reliable and reproducible than transabdominal views. The closed endocervical canal length is positively correlated with duration of gestation in a continuous fashion. Moreover, once values fall below 25 mm, preterm deliveries increase. In patients with prematurity risks, shorter cervical length is strongly predictive of delivery before 36 weeks; in combination with fetal fibronectin and other biomarker assays, ultrasound aids in identifying those at highest risk for imminent delivery.

Attempts to prevent premature births by tocolysis have been ineffective; postponing delivery for 48 hours to permit steroid enhancement of lung maturity has proven more feasible. First-trimester ultrasound prediction of the need for cervical cerclage has not been reliable. Specific candidates may benefit from either preventive or “rescue” cerclage procedures on the basis of midtrimester cervical lengths. Progesterone prophylaxis against prematurity by intramuscular injection or vaginal preparation has increased in recent years; the former has supporting evidence for effectiveness after a short cervix has been noted by ultrasound.

The gravid uterus is conveniently studied by ultrasound. Congenital Müllerian anomalies, including duplications and septations, occur in about 0.5% of the population (Fig. 11.34). Patients with bicornuate uteri may experience irregular bleeding in early pregnancy, altered cervical
Evaluation during pregnancy, although often augmented by MRI and computed tomography (CT). Ovarian torsion is a rare but acute surgical emergency; it is more likely with ART, ovarian cysts, and adnexal masses, favoring first-trimester or puerperal onset. Once an adnexal mass has been discovered by ultrasound, better characterization by MRI may be essential in avoiding unnecessary surgery during pregnancy. Asymptomatic lesions, even when quite large, are now generally expectantly managed until delivery or the postpartum period if malignancy is not suspected. Ultrasound is frequently sufficient for a secure diagnosis of corpus luteal cysts, benign cystic teratomas, and endometriomas. Use of MRI for atypical lesions can confirm the presence of fat, key to the diagnosis of teratoma and the exclusion of endometrioma. MRI aids in identification of hydrosalpinges and nongynecologic lesions. Expectant management of many adnexal masses past delivery is now more common, based on benign ultrasound, Doppler, and MRI characteristics.\(^{31}\)

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**Second-Trimester Ultrasound Study of the Fetus**

During second-trimester studies, a number of normal fetal structures are routinely identified and anomalies excluded. Basic anatomic surveys usually include documentation of fetal cranial integrity and central nervous system anatomy: midline brain structures, cavum septum pellucidum and thalami, lateral and third ventricles, choroid plexus, cerebellum, and posterior fossa.

Views confirming facial symmetry, intact orbits, clear lenses, paired nares, intact lips and hard palate, and normal profile are part of comprehensive studies. Fetal swallowing, respiratory movements, and nuchal structures may be noted. The spinal column is normally imaged in the long-axis, transverse, and coronal views. Distinct advantages in the visualization of the facial features, small parts, and spine are provided by 3D imaging (Fig. 11.35). Views of the thorax, including the axis, site, and relative proportions of cardiac and mediastinal structures with respect to the lungs and pulmonary vessels, yield indirect support for the integrity of the diaphragm. Over time, the basic cardiac examination has expanded from obtaining the axis, laterality, and rate to requiring symmetric four-chamber apical views and, as feasible, images of normal outflows, ductal and aortic arches, and additional arterial and venous tracts (Fig. 11.36). Prenatal echocardiography adds M-mode rhythm, color flow Doppler, and structural studies capable of details and diagnostic accuracy approaching those of postnatal examinations, subject to predictable limitations. Fetal abdominal views confirm the closure of the ventral wall; presence, normal size, and site of hepatic, gastric, splenic, pancreatic, vascular, and choledochal structures; renal and adrenal contours; normal bowel dimensions and echogenicity; bladder filling; and umbilical cord appearance. Accuracy of fetal sex assignment exceeds 99% by the midtrimester (Figs. 11.12 and 11.37). Fetal digits, small parts, cerebellar vermis, and
palatal components are often imaged by a combination of 2D and 3D. As gestation progresses, neurosonography may identify additional features of intracranial architecture and cerebral maturation.

**Doppler Ultrasound**

Initially considered revolutionary, obstetric Doppler has been hampered by costs, time constraints, safety concerns, and lack of clear utility for most patients. Currently accepted applications, originally employed in research and referral centers, commonly include identification and localization of blood flow, resistance to flow, vessel patterns, and directionality and velocity in cardiopulmonary structures and in a wide range of additional vessels, most commonly the uterine arteries, umbilical vessels, ductus venosus, and middle cerebral artery.

Uterine blood flow patterns reflect both maternal vascular resistance and placental site. Abnormal Doppler results in these and other vessels precede growth restriction and maternal hypertensive complications. Paired umbilical cord arteries are readily shown by color Doppler to bifurcate around the bladder (see Fig. 11.5). Absence of one umbilical cord artery may have associated cardiac and renal anomalies, aneuploidies, or growth restriction. Abnormally high umbilical artery resistance is more often characteristic of uteroplacental constraints on fetal growth than of anomalies or aneuploidy. Doppler findings may suggest the origin of a size/date discrepancy, but weight estimation more reliably identifies the small-for-gestational-age fetus. Persistent absence or reversal of end-diastolic flow, albeit rare, is associated with severe growth restriction, and perinatal morbidity and mortality. Middle cerebral artery resistance and umbilical venous patterns usually change later in the course of compromised perfusion; paradoxical ductus venosus and umbilical venous patterns are considered premorbid. Schemes combining clinical risks, biometry, Doppler measurements of the umbilical and middle cerebral vessels, and biophysical testing appear to have improved outcomes for growth-restricted fetuses while reducing the frequency of iatrogenic prematurity.²

Doppler measurement of peak systolic velocity in the middle cerebral artery has become integral to detection of fetal anemia and management of isoimmunization...
Placental dysfunction leading to fetal growth restriction (FGR) is an important risk factor for neurodevelopmental delay. Observations clarify that FGR evolves prenatally from a preclinical phase of abnormal vascular, nutrient, and endocrine milieu to a clinical phase differing in characteristics in preterm versus term pregnancies. Relating childhood neurodevelopment to prenatal characteristics may aid in identifying mechanisms and timing of critical insults. Based on available studies, lagging head circumference, degree of FGR, gestational age, umbilical artery (UA), aortic and cerebral Doppler parameters may be independent prenatal predictors of infant and childhood neurodevelopment. Head circumference is meaningful independent of gestational age, but a generalized growth delay has greater impact in early-onset FGR. Gestational age at delivery becomes the chief determinant of neurodevelopment before 32 to 34 weeks’ gestation. Doppler evidence of placental resistance to flow is increasingly important after 27 weeks, with maximum concern when flow reversals are noted in the umbilical artery, aorta, ductus venosus, or the umbilical vein. These findings predominate in early-onset FGR; cerebral vascular impedance changes, sometimes expressed as a ratio between the pulsatility indices of the middle cerebral and umbilical arteries, are important in later FGR. Abnormal motor and neurologic delays occurring in preterm FGR give way to cognitive effects and abnormalities related to specific brain areas as gestation advances, suggesting different pathophysiologies or evolving vulnerabilities of the fetal brain. Current studies do not suggest that fetal deterioration has an independent effect on neurodevelopment in early-onset FGR; the role of intervention in late FGR requires more research: risks for damage and patterns of deterioration may differ in the third trimester.49

Ultrasound-Guided Procedures

Real-time ultrasound is well suited by virtue of safety, economy, and convenience for guiding placement of needles, cannulae, catheters, and other devices used in fetal diagnosis and therapy. The range of procedures includes oocyte retrieval and embryo transfers, embryonic and fetal reduction, chorionic villus sampling, placental and skin biopsies, amniocentesis and amnioreduction, cord blood sampling, intrauterine transfusion, aspiration of fluid from various fetal and maternal sites, and an adjunct to fetoscopy and vascular ablation. The effectiveness as well as complications from procedures are often apparent in real time. Determinations of fetal status; relative fetal and umbilical cord positions during external version for malpresentations, labors, and immediately before transfusions; surgical interventions; and the EXIT (EX utero intrapartum treatment) procedure are important accepted applications.

Fetal Well-Being Assessment

A tenet of antepartum testing is that more accurate predictions of fetal wellness are achieved in proportion to the number of variables considered. See also Chapter 12. The biophysical profile (BPP) is a noninvasive ultrasound-based clinical tool that integrates levels of dynamic biophysical activities into a usable standard. The BPP attempts to predict the presence or absence of fetal asphyxia to prevent progression to metabolic acidosis and fetal death. The BPP seeks to identify four parameters in a 30-minute period, each of which is assigned two points: (1) observation of continuous tidal fetal breathing for 30 seconds, (2) a vertical amniotic fluid pocket of at least 2 cm (or 2 cm in 2 planes), (3) three obvious fetal movements, and (4) fetal tone, manifested by brisk flexion of small parts or hands. Death or injury within 1 week is more likely with scores below six, with loss of tone considered a late finding. Conversely, scores of eight
are considered highly reassuring for well-being during the same period. A fetal non-stress test, sometimes included for an additional 2 points (total score of 10), does not usually affect the predictive value of the BPP.

Antepartum testing by BPP should not be performed prior to viability or when fetal or maternal conditions preclude successful intervention. Cautious interpretation is appropriate for extreme fetal prematurity; in the presence of anomalies; maternal metabolic abnormalities; during labor; and with some drugs, including opiates, sedatives, and antenatal corticosteroids.

**Fetal Anomalies**

Neonatal outcomes can be optimized by forewarning and timely preparation when fetal abnormalities are present. Unfortunately, more than one-third of birth defects remain unrecognized prenatally. False-positive diagnoses are less frequent, but up to 10% discordancy with autopsy findings highlights the inherent uncertainties of prenatal diagnosis. Familiarity with imaging pitfalls associated with common anomalies is imperative to avoid misdiagnosis. Suspected structural defects should always be visualized in the correct plane with attempted confirmation in other planes. Precise measurements using specific criteria, appropriate gain settings, and additional modalities may be required for correct interpretation and management. Expert second opinions by patient referral or image review are available for most findings. Despite acknowledged limitations, prenatal ultrasound is the standard method for recognition of fetal anomalies.

**Central Nervous System**

**Fetal Ventriloculomegaly**

Hydrocephalus and ventriculomegaly are not strictly synonymous. Hydrocephalus connotes raised intracranial pressure, a functional observation not directly amenable to ultrasound. Prenatal enlargement of lateral ventricles (Fig. 11.39) may occur without altered pressure because of developmental abnormalities. Enlargement of the atria and posterior horns of the lateral cerebral ventricles (colpocephaly) may be associated with agenesis of the corpus callosum or type II Chiari malformations. Dilated lateral ventricles may also result from obstructed cerebrospinal fluid flow by hemorrhage or from brain destruction by a variety of causes, including ischemia and infection (e.g., cytomegalovirus, toxoplasmosis, or Zika virus). A single measurement over 10 mm at the level of the posterior atria of the lateral ventricles defines ventriculomegaly.

Atrial widths tend to decrease slightly during pregnancy. Choroid plexus positioning is gravity-dependent; on axial views, the choroid, attached at the level of the foramen of Monro, rests on the dependent wall of the lateral ventricle, marking the limit of the lateral ventricle, even when the wall cannot be visualized. A “dangling” choroid plexus conveys the extent and severity of ventricular enlargement (Fig. 11.40).

Clinical outcomes of severe ventriculomegaly (atrial size greater than 15 mm) are discouraging; survivors are often mentally and physically impaired. Hydrocephalus associated with arachnoid cyst, atresia of Monro, absence of the corpus callosum, or minor intracranial hemorrhage may have a better prognosis. By contrast, hydrocephaly seen with holoprosencephaly, encephalocele, congenital syndromes, or infection generally leads to loss or greater disability. Formal diagnostic and management algorithms, combining ultrasound, MRI, genetic diagnosis, and infectious disease screening play an essential role in counseling and care when hydrocephaly is identified.
In utero shunting has no current role in the treatment of prenatal ventriculomegaly. Asymmetric or rounded lateral ventricles and mild ventriculomegaly (widths between 10 and 12 mm) convey an uncertain prognosis, as most infants are normal, particularly when resolution occurs; slightly wider dimensions (12-15 mm) carry an intermediate prognosis.

Agenesis of the corpus callosum can be isolated but is commonly associated with other anomalies of abnormal neuronal migration. Midtrimester diagnosis of many brain lesions is hampered by ongoing differentiation and growth of the central nervous system; third-trimester transvaginal cranial ultrasound and MRI are particularly helpful in assessing the progressive development of gyri and sulci of the brain (Fig. 11.41).

**Meningomyelocele and (Type II) Chiari Malformation**

Meningomyelocele (Fig. 11.42) may be associated with a Chiari II malformation, that is, downward displacement of the hindbrain, as well as with other brain anomalies, including ventriculomegaly (Fig. 11.43). Often, hydrocephalus does not develop in children with neural tube defects (NTD) until after birth or lesion repair. Open NTD (Fig. 11.44) is usually accompanied by markedly elevated alpha fetoprotein levels in both maternal serum and amniotic fluid. A meningomyelocele appears on ultrasound as splaying or divergence of the posterior ossification centers, best appreciated on axial spinal views. A fluid-filled sac may be seen and the integrity of the overlying skin can be visibly disrupted on axial and sagittal images. The level

• **Fig. 11.41** Fetal MRI at 35 weeks’ gestation demonstrating agenesis of corpus callosum. Axial scan shows parallel lateral ventricles enlarged posteriorly (colpocephaly) in a teardrop shape.

• **Fig. 11.42** MRI of sacral meningomyelocele (thick arrow) at 21 weeks’ gestation; normal posterior fossa including cerebellum and fourth ventricle (thin arrow).

• **Fig. 11.43** Fetal MRI at 19 weeks demonstrates Chiari II malformation. Note dilated lateral ventricle (straight arrow) and communication between lower lumbar spinal canal and amniotic fluid–open neural tube defect (curved arrow).
of the meningocele can be ascertained more accurately with 3D ultrasound and MRI (see Figs. 11.35 and 11.42), helpful for predicting the outcome in affected children. Severe cases may be associated with kyphoscoliotic spinal deformity or clubbing of one or both feet. Prenatally, extremity movement may seem to be preserved; however, absence of normal activity better correlates with postnatal motor function.

**The Skull and Brain in Spina Bifida**

The “lemon sign” refers to the altered appearance of the calvarium, similar in shape to a lemon, on an axial view of the skull and brain (see Fig. 11.40). Biconcave frontal bones produce the calvarial distortion. The sign is best demonstrated between 18 and 24 weeks but is not specific, appearing sometimes in otherwise normal children. The “banana sign” refers to abnormal cerebellar positioning, curled in a crescent (banana-like) around the brainstem (see Fig. 11.44). Obliteration of the cisterna magna and cerebellar distortion are secondary to type II Chiari malformation. One or both signs are seen in the majority of spina bifida cases during the midtrimester and should prompt a more detailed examination of the spine. Prenatal surgical closure of defects may improve outcome in appropriate candidates, particularly with respect to preservation of motor function and prevention of hindbrain displacement.

**Anencephaly**

Anencephaly, the absence of normal brain and calvarium superior to the orbits, can be detected as early as 10 weeks but is recognizable throughout gestation (Fig. 11.45). Maternal serum alpha fetoprotein is usually very elevated. About half of cases have associated anomalies, for example, meningocele, cleft palate, or clubfoot. Occasionally, the typical appearance is altered by the presence of echogenic material superior to the orbits, identified pathologically as angiomatous stroma (“area cerebrovasculosa”). Polyhydramnios and malpresentation are common later findings. Abnormal prolongation of pregnancy may occur, presumably secondary to lack of fetal neurohormonal cues for labor. Anencephaly is considered incompatible with
meaningful survival; most affected live-borns succumb in the immediate neonatal period. Intervention for purely fetal indications is futile and is not recommended.

**Encephalocele**

The extracranial protrusion of brain tissue within a meningeal sac is a straightforward ultrasound diagnosis, although dependent on site and size of lesion and appropriate visualization. Most encephaloceles in the Western world are occipital and midline and should be distinguished from soft tissue edema or lymphangiomata of the neck. The identification of a calvarial defect allows a specific diagnosis (Fig. 11.46). Encephaloceles may be isolated or associated with amniotic band syndrome (when off midline) and genetic syndromes such as Meckel-Gruber. Prognosis is affected by site, size/amount of extruded tissue, and adversely affected by ventriculomegaly, microcephaly, and syndromic etiology. Anterior sites are more common in Asian populations; small nasal or frontoethmoidal encephaloceles may initially elude even postnatal detection.

**Holoprosencephaly**

Holoprosencephaly, a malformation resulting from early failure of division of the embryologic prosencephalon or forebrain, is frequently associated with facial abnormalities (Fig. 11.47A). Its incidence ranges from 0.6 to 1.9 per 10,000 live births, although this is likely an underestimate because many cases spontaneously abort and milder variants can be unrecognized. In cases without chromosomal abnormalities, recurrence risk is estimated to be 6%. The etiology of holoprosencephaly is heterogeneous and not fully known with most cases being sporadic. Environmental, mechanical, and genetic factors have all been implicated. Several chromosomal disorders are linked to this anomaly, most notably trisomy 13, with 70% exhibiting holoprosencephaly. Holoprosencephaly in association with extra-cephalic malformations strongly suggests aneuploidy; there are also autosomal dominant and recessive familial forms. The condition has been induced by a variety of teratogenic agents in animal models. In alobar holoprosencephaly, the most severe form, there is a complete failure of cleavage of the forebrain into two hemispheres. The cerebral hemispheres are fused and enclose a single prosencephalic ventricle. In addition to a single ventricular cavity, the thalami are fused, and the cavum septum pellucidum (CSP), corpus callosum, falx cerebri, optic tracts, and olfactory bulbs are all absent. The alobar condition is often associated with fetal loss; the vast majority of live-borns experience severe neurologic dysfunction or death in early infancy; less severe variants have a wider, less predictable range of outcomes. Partial cleavage results in semilobar holoprosencephaly, with posterior separation of the cerebral hemispheres, variable degrees of thalamic fusion, and absent olfactory bulbs and corpus callosum. In lobar holoprosencephaly, the abnormalities may be confined to absence of the CSP and corpus callosum with fusion of the lateral ventricles and cingulate gyrus; hemispheres are separated anteriorly and posteriorly. The mildest form, septal optic dysplasia, lacks only the CSP. Severe forms of holoprosencephaly may exhibit cyclopia (median mono-ophthalmia) with or without proboscis, cebocephaly (ocular hypotelorism and a blind single nostril), ethmocephaly (ocular hypotelorism with proboscis) (see Fig. 11.47B), and median facial clefts. The diagnosis of holoprosencephaly was typically made during the midtrimester anatomic survey by noting fused lateral ventricles and thalami and the absence of normal midline structures. Early prenatal detection of holoprosencephaly can occur between 11 and 14 weeks’ gestation by obtaining transverse cranial images in which the normal “butterfly sign” of choroid and ventricles is absent (see Fig. 11.47C).

**Dandy-Walker Malformation**

The Dandy-Walker malformation is a fluid-filled posterior fossa cyst, with variable occurrence of lateral ventriculomegaly; the posterior fossa is always enlarged and the tentorium uplifted. The cerebellar hemispheres, separated by the cyst, are rudimentary. Agenesia of the cerebellar vermis is difficult to confirm before 20 weeks because wide variations in developmental timing mean that full formation may not yet be present. The Dandy-Walker variant consists of direct communication between the fourth ventricle and cisterna magna, without posterior fossa enlargement and with only mild hypoplasia of the inferior vermis. The subtle ultrasound findings can be associated with serious abnormalities.
part 2

The Fetus

Sacrococcygeal teratomas (SCTs) are congenital germ cell tumors, usually benign but often large, arising posteriorly at the medial spinal base. Posterior bony elements of the lumbosacral spine are typically intact. Often SCT is associated with polyhydramnios, prematurity, and more rarely with anemia, high output cardiac failure, and hydrops. Maternal serum alpha fetoprotein is usually elevated. Generally, SCTs are not related to aneuploidy, although rare cases of chromosomal abnormalities have been reported. The typical teratoma is a complex cystic and solid mass protruding from the fetal rump, with arterial and venous flow seen by color Doppler. Uncommonly, it may be entirely cystic or solid, extend into the fetal pelvis (Fig. 11.50) and abdomen, or undergo malignant degeneration. Some SCTs have unique characteristics, including those found through volumes along any given orientation, and precisely locate abnormalities by use of stable anatomic reference points (Fig. 11.49).

Choroid Plexus Cyst

Cysts in the choroid plexus, noted in 1% to 2% of normal second-trimester pregnancies, usually resolve by the early third trimester (see Fig. 11.18). Cyst number, size, or persistence impart no additional clinical significance. Isolated choroid plexus cysts are generally associated with normal karyotype, do not require follow-up to confirm resolution, and do not affect postnatal development.\textsuperscript{14} Choroid cysts have been associated with trisomy 18.

Spine

3D imaging is extremely useful for studying fetal spine, vertebrae, ribs, pelvic bones, and spinal cord. Operators can simultaneously visualize three orthogonal planes, scroll through volumes along any given orientation, and precisely locate abnormalities by use of stable anatomic reference points (Fig. 11.49).

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immature teratomas, convey elevated risks for recurrence or malignancy.33,34

Head and Neck

After 14 weeks, visualization of the nose, nares, orbits, forehead, lips, eyes, and ears is feasible. Orbits are clearly seen axially; the ocular diameters, and interocular and binocular distances (defining hypertelorism and hypotelorism) are measurable. The profile reveals the forehead, nose, and jaw sagittally (contrast Fig. 11.1 with Fig. 11.51). The coronal view, best for facial structures, includes the orbits (and lenses), eyelids, nose, and lips. Coronal and 3D views identify facial clefting, including cleft lip and palate; clefts may be central or lateral, unilateral or bilateral, involving nasal structures or, more subtly, the soft palate. Sagittal views should include the nasal bones; nasal bone hypoplasia or absence is associated with an increased risk for Down syndrome. The complex anatomy of the fetal face is shown exquisitely by 3D sonography. The distinction between
normal and abnormal facial features can be seen clearly by comparing Fig. 11.8 with Fig. 11.52.

Cystic hygroma is a septate, cystic mass arising from the neck and occipital region secondary to lymphatic malformation; it may extend over the entire trunk (Fig. 11.53A). Posterior septation of the nuchal ligament and bony integrity distinguish this lesion from neural tube defects. Associated aneuploidies, particularly trisomy X (45,X), are common. When associated with generalized edema, cystic hygromas carry a grim prognosis; when small, isolated, and with normal karyotype, more favorable outcomes, including regression, are well documented. Less common neck lesions include teratomas, hemangiomas, branchial cleft cysts, and anteriorly, thyroid goiters and thymic absence.

Increased nuchal translucency (see Fig. 11.11) and nuchal fold thickness (see Fig. 11.17) are associated with aneuploidies, including trisomies 21 and 18, cardiac malformations, diaphragmatic herniation, dwarfsisms, a wide range of other anomalies and genetically inherited disorders (Noonan syndrome), lymphatic obstruction sequence, and fetal loss. With normal karyotypes and reassuring midtrimester surveys, outcomes are generally good.47

Heart

Optimal timing of the fetal heart study is a compromise between finding defects as soon as possible and accurately assessing complex anatomy, recognizing that some lesions manifest late in pregnancy. Basic heart evaluation is now an integral part of anatomic surveys at 18 to 22 weeks’ gestation. Initial appreciation of first-trimester cardiac anatomy is feasible by either a transvaginal or transabdominal approach, augmented by color Doppler; however, the best timing for early imaging of the four chambers and great arteries is usually at 13 to 14 weeks. In experienced hands, first-trimester fetal echocardiography is quite sensitive for the detection of major structural cardiac abnormalities. Some features useful in combination with nuchal measurements for aneuploidy screening include abnormal flow patterns in the ductus venosus and tricuspid valve; these are also early markers for cardiac abnormalities and should prompt more detailed assessment. A focused cardiac examination should be performed for known medical or historical risks, with teratogen exposures, and when any significant extra-cardiac anomaly or chromosomal defect has been identified. Cardiac defects should also be excluded in the presence of an aberrant right subclavian artery or other vascular anomalies.47

Screening for fetal cardiac abnormalities was introduced more than 30 years ago, yet prenatal detection remains challenging. Most major congenital heart defects are amenable to prenatal identification, with tertiary centers reporting high diagnostic accuracy; however, overall detection rates remain low and highly operator dependent. Pregnancies
chapter 11
perinatal ultrasound

Gastrointestinal Tract
Normal Bowel Appearance

Physiologic migration of the gut into the proximal umbilical cord occurs between gestational weeks 7-10 and may be seen on first-trimester scans (Fig. 11.55) up to 12 weeks. The normal liver always remains intra-abdominal; bowel migration should not be mistaken for a ventral abdominal wall defect.

The fetal stomach, seen transvaginally by 9-10 weeks of gestation, is an echo-free structure in the upper left abdomen, varying in size as it empties and fills.

Fetal small bowel is not undifferentiated in appearance early in gestation but appears as central abdominal fluid-filled loops by the late second trimester (Fig. 11.56). Fetal colon is clearly evident by the third trimester as a hypoechoic, haustrated tube at the abdominal periphery. Meconium in the large bowel is normal in the third trimester; second-trimester meconium normally may have transiently increased echogenicity. Hyper-echogenic bowel, bright as adjacent osseous structures, is a concerning but nonspecific finding associated with trisomy 21, cystic fibrosis,
bowel atresia, congenital infections (e.g., cytomegalovirus), growth restriction, placental abruption, and poor outcome. Gastrointestinal tract obstruction can often be diagnosed by ultrasound. Blockage of the proximal alimentary canal interferes with normal amniotic fluid regulation by fetal swallowing and gut absorption. In complete proximal obstructions, assuming intact membranes and normal renal production, polyhydramnios is invariable.

**Esophageal Atresia and Tracheoesophageal Fistula**

A nonvisualized fetal stomach combined with polyhydramnios should alert the examiner to the possibility of esophageal atresia. Unfortunately, these signs coincide in only 40% of cases. Nonvisualization of the stomach is not specific for esophageal atresia; proximal esophageal pouches are rarely visualized. Polyhydramnios, although present in two-thirds of cases, may not develop until the third trimester. The VACTERL complex consists of vertebral, anal, cardiovascular, tracheal, esophageal, renal, and limb malformations; these systems should be scrutinized if tracheoesophageal fistula is suspected. The stomach may also fail to fill when there is little fluid to swallow (e.g., as in oligohydramnios, or secondary to neuromuscular conditions like Pena-Shokeir syndrome that interfere with swallowing).

**Small Bowel Obstruction**

In duodenal atresia, the distended stomach and proximal duodenum produce a “double-bubble” sign akin to the neonatal radiologic finding (see Fig. 11.15). A strong (25%) association with trisomy 21 warrants genetic testing when duodenal atresia is suspected. Other etiologies include annular pancreas, duodenal web, malrotation, and severe duodenal stenosis. Ileal and jejunal atresias usually cause multiple distended bowel loops, often exhibiting exaggerated peristalsis (Fig. 11.57). More distal obstruction, as seen in meconium ileus, Hirschsprung disease, and anal atresia, may not result in visible bowel dilation until third trimester or postpartum. Meconium peritonitis, a sterile but morbid chemical response to intrauterine bowel perforation, is characterized by echogenic peritoneal calcifications and free intraperitoneal fluid (Fig. 11.58); some cases progress to echogenic pseudocysts.

**Anterior Abdominal Wall Defects**

A midline ventral abdominal wall defect involving the base of the umbilical cord is characteristic of omphalocele (Fig. 11.59). A sac surrounds the herniated viscera and may include the liver (Figs. 11.60 and 11.61). Omphalocoles are often associated with other anomalies (Beckwith-Wiedemann syndrome, pentalogy of Cantrell) and aneuploidies (trisomies 13 and 18). In gastroschisis, herniated bowel loops float freely in the amniotic cavity without a covering membrane (Fig. 11.62). Defects are usually to the right of the umbilical insertion; aneuploidies, extra-intestinal abnormalities, familial forms, and recurrence are not customary with gastroschisis. Gastroschisis is thought to be caused by a vascular accident with wall infarction; young maternal age, use of vasoactive substances, and NSAIDs have been related to increased risk for this lesion.

**Diaphragmatic Hernia and Thoracic Lesions**

Congenital diaphragmatic hernia (Fig. 11.63) is diagnosed when abdominal organs (usually stomach and bowel) are seen in the thorax. Fluid-filled intrathoracic bowel loops with visible peristalsis help confirm the diagnosis. Associated congenital heart disease, other structural defects, and chromosomal abnormalities worsen a guarded prognosis. Hernia contents produce a mass effect, shifting the mediastinum and compressing the lungs (Fig. 11.64); the resulting pulmonary hypoplasia and secondary pulmonary hypertension are major determinants of mortality. Polyhydramnios and lagging abdominal circumference are common associations. Estimation of lethality is usually based on lung measurements at the level of the four-chamber heart (expressed as
observed to expected lung/head circumference); the process is unreliable, largely because most lesions are in an intermediate prognostic category. Calculated 3D lung volumes by ultrasound study and MRI provide more precise measurements of the lungs but with only minor impact on prognostic accuracy. Liver herniation into the chest (Fig. 11.65) is common but carries a worse prognosis; right-sided hernias are harder to recognize and may also increase mortality. Bilateral herniation is generally lethal with few documented exceptions.

Congenital pulmonary adenomatoid malformations (Fig. 11.66) may have cysts with a similar appearance to bowel but lack peristalsis and displacement of identified abdominal organs. Microcystic forms are echogenic and resemble sequestrations; the latter usually have systemic arterial feeding vessels (Fig. 11.67), although blended forms have been described. Growth is variable, with spontaneous regression during pregnancy commonly reported. “Vanishing” lesions, not identified by postnatal x-rays, are always present on chest CT. Fetal hydrops may develop; fetal surgery

• Fig. 11.59 Three-dimensional views of an omphalocele (star) at 18 weeks’ gestation.

• Fig. 11.60 Color Doppler view of a massive omphalocele (outline) that includes the liver and hepatic vessels (color). The extruded portion and the sac are almost equal in diameter to the torso (to the right).

• Fig. 11.61 Fetal MRI of omphalocele at 24 weeks’ gestation. Note membrane (thin arrow) covering the omphalocele that includes the liver. Fetal stomach and bladder are shown by thick arrows. Note normal smooth brain surface at 24 weeks without gyri or sulci.
The Fetus

Gallbladder and Bile Ducts

A choledochal cyst is a localized circular dilation of the biliary system, often involving the common bile duct and located in the upper right abdominal quadrant, separate from the gallbladder. Echogenic gallstones and sludge are noted in the gallbladder lumen on third-trimester scans (Fig. 11.68) in one out of every 200 fetuses. The prognosis of fetal gallstones and biliary sludge is favorable. Follow-up studies in neonates generally confirm resolution once separated from the maternal hormonal milieu; infants are usually asymptomatic. The fetal gallbladder can often be seen after 14 weeks' gestation; its rare absence, suspected by persistent nonvisualization, is associated with a variety of abnormal conditions, most notably biliary atresia and cystic fibrosis.16

may be indicated to avert this potentially fatal complication. Gestational age-normalized congenital pulmonary airway malformation volume and congenital cystic adenomatoid malformation volume ratios have been proposed as methods for anticipation of hydrops. The latter ratio has been used to identify the need for fetal intervention. In predominantly microcystic lesions, maternal betamethasone therapy has been reported to lower risks for polyhydramnios and fetal hydrops.15

• Fig. 11.62 Gastrochisis. Transverse abdominal image of the spine (left, short arrow) demonstrates free bowel loops (thick arrow) to the right of the umbilical cord insertion (long arrow). This lesion is more common in younger gravidas and is usually isolated.

• Fig. 11.63 MRI of posterior Bochdalek diaphragmatic hernia at 28 weeks’ gestation. Note the loops of bowel occupying the left hemithorax and compressing the lung.

• Fig. 11.64 Fetal MRI of posterior Bochdalek diaphragmatic hernia at 28 weeks’ gestation. Note the loops of bowel occupying the left hemithorax and compressing the lung.

• Fig. 11.65 MRI of right-sided diaphragmatic hernia at 32 weeks’ gestation. Liver (thick arrow) has herniated into right thorax with compressed right lung. Thin arrow shows incidental nuchal cord.

• Fig. 11.65 Fetal MRI of right-sided diaphragmatic hernia at 32 weeks’ gestation. Liver (thick arrow) has herniated into right thorax with compressed right lung. Thin arrow shows incidental nuchal cord.
arteries (Fig. 11.70). Normal fetal ureters are not visible by ultrasound. Fetal adrenal glands are usually identifiable because of a relatively large cortex and unique, contrasting echo textures. Lethal renal malfunctions, including bilateral multicystic dysplasia, bilateral agenesis, and some cases of congenital

Genitourinary Tract

The kidneys, readily visible by 12-14 weeks’ gestation, are located lateral to the fetal spine (Fig. 11.69) below the diaphragm and prominent fetal adrenal glands. Normal renal size and volume can be plotted against gestational age. By the late third trimester, fat deposition creates an echogenic border, enhancing visualization. The bladder, may be seen by 11 weeks of gestation, flanked by paired umbilical

• Fig. 11.66 Congenital pulmonary adenomatoid malformation. MRI at 26 weeks in the fetal coronal plane showing both lungs. Multiple large fluid-filled cysts are present.

• Fig. 11.67 Pulmonary sequestration is shown. Power Doppler highlights the feeding vessel (arrow).

• Fig. 11.68 Fetal gallbladder (£GB$) (arrow) with echogenic sludge and small calculi in the third trimester. Findings usually resolve once maternal hormonal levels decrease. Compare with normal echo-free appearance in Fig. 11.19.

• Fig. 11.69 Transverse view of the fetal abdomen, spine up, demonstrating the normal renal appearance (arrows).

• Fig. 11.70 Power Doppler demonstration of two umbilical cord arteries (arrows) outlining the fetal bladder (B).
autosomal recessive polycystic kidney disease result in failure of fetal urine production. After 16 weeks, amniotic fluid is almost entirely fetal urine, but oligohydramnios may not become apparent until 18-20 weeks of gestation. Infants born after prolonged oligohydramnios have a characteristic facial appearance, limb deformities, and pulmonary hypoplasia, termed Potter syndrome (or sequence). The finding of oligohydramnios should direct attention to the fetal urinary tract with rescanning at 30-minute intervals for confirmation if the fetal bladder is not initially visualized. Anhydramnios (no measurable fluid), a persistently empty bladder, and an inability to identify fetal kidneys or renal arteries are strongly suggestive of bilateral renal agenesis. Unfortunately, absence of amniotic fluid may impede making a secure diagnosis with its associated implications for management, particularly because fetal adrenal glands, losing their normal angulation in renal agenesis, may be mistaken for kidneys. Fetal MRI can be ideal in establishing this lethal diagnosis. Care must be exercised when suspecting unilateral renal agenesis to exclude renal ectopy (pelvic kidney); color Doppler may reveal the aberrant course of the renal artery (see Fig. 11.4).

Pyelectasis (dilation of the renal pelvis), occurring in up to 5% of pregnancies, is a variant that should not be misinterpreted as hydronephrosis. Pyelectasis is considered a minor marker for trisomy 21 but when isolated is not usually an indication for amniocentesis; transient urinary reflux is a common underlying cause. Measurements of anteroposterior views of the kidneys and renal pelves in the transverse plane are used to identify both pyelectasis and hydronephrosis. Upper limits of 4 mm and 7 mm in second and third trimesters respectively define pyelectasis, with normalization by the third trimester routinely anticipated. Pelvic dilation above 1 cm is suspicious for hydronephrosis, particularly with calyceal involvement. Vague correlation exists between the magnitude of fetal renal pyelectasis and renal abnormalities. Persistent pyelectasis should be confirmed neonatally and followed until either resolved or an etiology established.44

Urinary tract obstruction may have a variable sonographic appearance, depending mainly on timing of onset. Renal dysplasia, sometimes with cyst formation, is the consequence of early obstruction; obstruction occurring later in gestation is more likely to result in dilation of the collecting system (Fig. 11.71).

Hydronephrosis may be unilateral, resulting from obstruction of the ureteropelvic or ureterocystic junctions, or bilateral, secondary to lower tract obstruction by posterior urethral valves or urethral agenesis. A distended, thick-walled bladder may be seen in males with posterior urethral valve, along with hydroureretes; urinary ascites may occur as a result of calyceal or bladder rupture with intraperitoneal accumulation of urine. “Keyhole” dilation of the posterior urethra helps in distinguishing this lesion from Eagle-Barrett (prune-belly) syndrome (Fig. 11.72).

Multicystic dysplastic kidney disease (MCDKD) is commonly an incidental unilateral finding on prenatal examination. Bilaterality is less frequent but lethal. In unilateral cases, abnormalities of the contralateral kidney are common.24 Associated nonrenal abnormalities occur frequently with both unilateral and bilateral MCDKD, increasing risks for chromosomal abnormality. Males predominate in unilateral MCDKD (ratio of 2.4:1), but females are four times more likely to have chromosomal abnormalities and twice as likely to have nonrenal anomalies or lethal bilateral disease.24 The option of chromosomal analysis should be discussed whenever there are extrarenal abnormalities or bilaterality. Unilateral isolated MCDKD with normal amniotic fluid is not associated with abnormal genetic studies and usually has a favorable outcome; however, a neonatal work-up to exclude reflux in the unaffected kidney should be performed.37
Severe hydronephrosis, in contrast to MCDKD, is an orderly arrangement of the enlarged renal pelvis with connecting, dilated calyces. Bilateral renal anomalies (including bilateral secondary multicystic renal dysplasia) are common (4%) in the fetus (Fig. 11.73). Dilated ureters occur in ureterovesical junction obstruction, primary megaureter, or secondary to reflux. Infantile autosomal recessive polycystic kidney disease causes bilaterally enlarged, echogenic reniform kidneys (Fig. 11.74). Other renal and urogenital anomalies identified prenatally include cysts, masses, duplicated collecting systems, and bladder extrophy; in the last, a nonvisualized bladder coincides with normal amniotic fluid and renal appearance. A short or bifid male phallus is sometimes associated.

Reliable identification of male and female sex is anticipated after 16 weeks’ gestation (see Fig. 11.37) by external genital examination, with interposition of uterus noticeably separating the fetal bladder from the rectum. Fetal ovarian cysts, fluid-filled or separtate, are often abdominal and hard to distinguish from gastrointestinal and other lesions. Hydrometrocolpos may also produce a complex fluid-filled pelvic structure (see Fig. 11.37C).

Identification of genes involved in sex differentiation, noninvasive prenatal genetic screening, measurements of...
key mediating hormones, combined with ultrasound, have allowed multidisciplinary teams to identify cases of ambiguity, conduct detailed prenatal evaluations, and provide informed parental counseling.

Following the characterization of the sex-determining region on the Y chromosome in 1990, there have been a number of genes found to play a role in sex development. The most common disorders of sex development (DSD) result from disruption of androgen levels and activity affecting later embryonal development, for example, congenital adrenal hyperplasia and androgen insensitivity syndrome. Available powerful genetic techniques can interrogate the entire genome for causative changes; it is important to critically assess the flood of genetic data for meaningful information. Recent discoveries have clarified roles of various transcription factors in DSD, including SOX9, SF1, and WT1 genes. Disruption of signaling molecules, such as hedgehog, WNT, cyclin-dependent kinase, and Ras/MAP kinase is known to cause some DSD. Dosage dependence of genes involved in gonadal development is a recurrent theme; genetic changes in promoter and repressor regions are being probed by microarray analysis and other techniques. Multiple phenotypes result from deletion, duplication, homozygous, heterozygous, and regulatory region changes in the same gene. Ongoing studies may yield clinically applicable insights into DSD and its underlying genetic basis.23

Musculoskeletal System

Examination of all four extremities and determination of femur length is routine in prenatal studies. Normograms exist for all fetal long bones, feet, and many other skeletal components. Measurements falling more than three standard deviations below the mean for age are very suspicious for skeletal dysplasia. Because normal individuals by definition populate the fifth percentile, it is reassuring that most skeletal dysplasias are dramatic; an abnormal femur-to-foot ratio (<0.89) provides additional diagnostic support. The biparietal diameter usually continues to reflect gestational age unless the skull is also involved.

Careful measurement of all bones in the peripheral skeleton is required in suspected cases, first to define the category of dysplasia and then, if possible, to make a diagnosis. Extreme generalized limb reduction or absence is termed phocomelia; overall limb shortening is micromelia; rhizomelia refers to proximal (femurs and humeri) reduction; mesomelia to more distal (forearms and lower legs); and acromelia to the most distal (feet and hands) dysplasias. Fractures and curvatures; altered bone density; the appearance of the spine, skull, and ribs; and extraskeletal anomalies may be keys to more specific diagnoses. Polyhydramnios may also be present and is often a poor prognostic sign.

Pulmonary hypoplasia from restrictive thoracic deformation is often the cause of death in severe skeletal dysplasias. Lung volume can be assessed by MRI or by 3D imaging, but a ratio of FL/AC below 0.16 functions as a convenient proxy for more cumbersome estimators for lethality.37

Thanatophoric dysplasia is uniformly fatal and relatively common, often recognizable in utero by severe micromelia, curved femurs, and narrow thorax (Fig. 11.75). Useful for diagnosis, when present, is a cloverleaf-shaped skull. Achondroplasia, also lethal, manifests severe micromelia with poor vertebral ossification. Lethal type II osteogenesis imperfecta presents with short, angulated fractures of long bones and ribs (Fig. 11.76). Bone density, particularly calvarial, is decreased. Fatal forms of hypophosphatasia may have similar severe demineralization with strikingly clear resultant central nervous system imaging and easy compressability of the skull. Short-ribbed polydactyly syndrome, also lethal, is characterized by a small chest with hypoplastic truncated ribs, postaxial polydactyly, and polyhydramnios (Fig. 11.77).

Heterozygous achondroplasia is the most common non-lethal skeletal dysplasia; homozygous status is incompatible with survival. Shortening of the femur, frontal bossing, trident hand shape and increased amniotic fluid volume characterize this autosomal dominant rhizomelic disorder, rarely apparent prior to the third trimester. Prenatal genetic diagnosis is limited to known carriers. Eighty percent of
cases are new mutations; some are associated with advanced paternal age.

Other abnormalities of the musculoskeletal system include the malformation or absence (dysostosis) of various skeletal portions; for example, limb reduction anomalies including radial-ray syndromes (hypoplasia or absence, often affecting thumb development) and hemimelia. Amniotic band syndrome (Fig. 11.78) asymmetrically amputates or truncates an extremity, although the narrow strand may not persist. Valgus deformities of the feet (Fig. 11.79) may be isolated, familial, positional, or secondary to central nervous system and spinal cord abnormalities.

Pitfalls in the diagnosis of skeletal dysplasia are appreciable. Complete current classification from the International Skeletal Dysplasia Society is available online.\(^{45}\) Classification of skeletal dysplasias is constantly being altered by new genetic information.

**Two-Vessel Umbilical Cord**

A single umbilical artery may be identified on transverse views of free cord loops or at the bladder bifurcation based on color Doppler (Fig. 11.80). Single umbilical arteries have been linked to other anomalies, unfortunately without a consistent pattern. Lack of normal coiling and short cords are also linked to abnormalities, particularly those affecting movement. It is unclear whether an isolated single umbilical artery is associated with aneuploidy, nor does the laterality of the absent vessel have significance. Being small-for-gestational age is more common in neonates with single umbilical artery. Counseling, selective use of amniocentesis, and serial third-trimester examinations to document fetal growth may be helpful in management.
• Fig. 11.79 Valgus deformity of the fetal foot may be isolated, familial, positional, or secondary to central nervous system and spinal cord abnormalities.

Summary

As sonography nears maturity, it has had gratifying success in fulfilling its early potential. Initial limitations have yielded to advances in technology and technique or have been augmented by genetic and biochemical breakthroughs. MRI is now well established as a versatile partner to ultrasound in prenatal diagnosis. The future of imaging holds immense promise for continued progress in improving neonatal outcomes.

Key Points

• All practitioners who perform or supervise the performance of obstetric ultrasonography should be licensed medical practitioners, having received specific training in obstetric ultrasonography.
• Gray-scale B-mode ultrasound is associated with a negligible rise in temperature. Strong magnetic fields and radiofrequency waves are used in MRI with no known harmful effects. Large longitudinal studies confirming the safety of ultrasound and MRI are lacking.
• A standard ultrasound study should follow the guidelines published by the American Institute of Ultrasound in Medicine.
• An important application of prenatal ultrasound is using biometrics to establish gestational age and the utility of the biophysical profile to confirm fetal well-being.
• Genetic screening of the fetus should combine ultrasound study and biochemical testing or cell-free fetal DNA to enhance the detection of chromosomal abnormalities.
• Neonatal outcomes can be optimized by forewarning and timely preparation when fetal abnormalities are present.
• Nondiagnostic studies, varying prognoses for a given diagnosis, and the inherent limitations of ultrasound and MRI studies might lead to ethical issues in management and counseling.

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