Ultrasound has become the standard method for the diagnosis of fetal congenital abnormalities and is quite frequently used for that purpose. The reasons for its widespread use are attributed to improvement in the skill of ultrasonographers and the introduction of high-resolution equipment.

In this chapter, we first discuss the types of ultrasound examinations performed during pregnancy. Subsequently, we present the criteria for the diagnosis of the major abnormalities involving the chest and gastrointestinal, cardiac, and renal systems. Where appropriate, we discuss the accuracy of the ultrasound diagnosis and its impact on pregnancy management.

The Ultrasound Examination

Unlike the practice in some European countries, antenatal ultrasound is not routine in the United States. Rather, the examination is requested only on indication and the decision to order the test is reached by agreement between the health care provider and the patient. The rationale for this policy emanates from concern about exposing the entire pregnant population to ultrasound and lack of sufficient studies defining the cost/benefit ratio of routine ultrasound.

It is important to note, however, that no biologic ill effects have been noted in humans from the use of diagnostic ultrasound. Nevertheless, the issues of safety and cost benefit ratio will remain areas that will linger with us for some time to come. Directly related to these issues are two factors: the length of the ultrasound examination and the amount of data that should be included in the examination.

Constraint on the time necessary to conduct a complete ultrasound study is directly linked to the issues of safety and cost effectiveness. First, common sense dictates that unnecessary long exposures are best avoided. Second, prolonged studies designed to gather detailed data on every fetus require additional time in terms of personnel and equipment utilization. Thus, studies requiring a long time to conduct are costlier.

The additional cost of prolonged studies may be difficult to justify because the potential benefit of gathering voluminous data routinely is also not clear. As a result, it is logical to assume that the length of ultrasound studies should be proportional to the nature of the indication. For example, a detailed anatomic fetal survey would be justified in a pregnancy at high risk for fetal anomalies, but not necessarily in one where such risk factors are nonexistent.

Participants in the 3rd International Genetic Conference, held in 1980 at Scarborough, Maine, were the first to address issues of this nature. They defined clear objectives for two types of ultrasound examinations based on relative risk factors.
The two-tier system included two levels of study, one more detailed than the other. The first was a level I or stage I examination, basically directed toward assessment of dates, fetal growth, number of fetuses, placental location, amniotic fluid volume, and major anomalies such as anencephalus and obvious masses in the fetal trunk, chest, and abdomen. The other was a level II or stage II examination directed toward detailed imaging of fetal anatomic structures in women at high risk for delivering infants with congenital anomalies.

In the last few years, however, the level I examination evolved into a more complete examination, referred to as a *standard study*. The level II examination also evolved into a more sophisticated study for pregnancies at high risk for birth defects, and is referred to as detailed or targeted imaging for fetal anomalies (TIFFA).² The TIFFA studies are usually performed in tertiary centers by ultrasonographers with extensive experience in the diagnosis of anomalies.

**Standard Study**

The standard study includes assessment of the biparietal diameter (BPD), head circumference (HC), abdominal circumference, femur length (FL), cerebellar diameter, nuchal fold thickness, orbital diameter, amniotic fluid volume, placental position, and, when indicated, umbilical Doppler velocimetry and biophysical profile. These data generate information relative to pregnancy dates, fetal growth, and fetal well-being.

Specific anatomic views are also examined in a standard study. In particular, attention is directed (1) to the shape of the head, to rule out abnormalities such as the lemon and banana signs; (2) to increased thickness of the nuchal fold as in fetuses with Down syndrome; (3) to the size of lateral ventricles and posterior fossa; and (4) to the four-chamber view of the heart. These four areas represent specific anatomic views that screen for spina bifida, Down syndrome, hydrocephalus, and congenital heart disease, respectively, discussed elsewhere.

**TIFFA Study**

In the TIFFA study more detailed anatomic views are obtained, particularly those necessary to rule out more subtle anomalies, including small spina bifidas located in the sacral area or congenital heart lesions that are not visualized in the four-chamber view of the heart.

The indications for TIFFA include elevation of α-fetoprotein (AFP) in maternal serum or amniotic fluid, polyhydramnios or oligohydramnios, history of a previous defect, maternal diabetes mellitus, intrauterine growth retardation, breech presentation at term, suspicion of an anomaly on a standard study, and abnormal Doppler velocimetry, in the form of absent end-diastolic velocity (AEDV).

In conducting a TIFFA examination the ultrasonographer targets specific anatomic views, the image of which is clear in his or her mind. Each of these views is then recorded either on tape or on photograph. The specific planes or anatomic views looked for are listed in Figure 13.1.

For each of the views in Figure 13.1, the ultrasonographer should indicate if the specific plane is clearly or poorly visualized and whether the anatomic relationships appear normal or abnormal.² If an abnormality is detected a descriptive synthesis of the findings should be presented in the “comment” section (Fig 13.1).

**Fetal Anomalies: Diagnosis and Management**

**Abnormalities of the Chest and Gastrointestinal Tract**

**Chest**

Ultrasonically recognizable anomalies of the chest mainly include esophageal atresia, cystic adenomatoid malformation, and primary as well as secondary pulmonary hypoplasia.

**Esophageal Atresia**

Esophageal atresia is a rare anomaly, occurring at the rate of 1:25,000 births. The presence of a