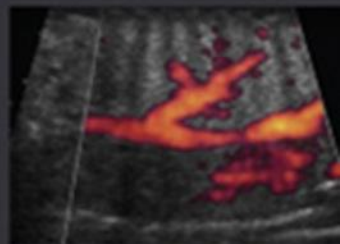
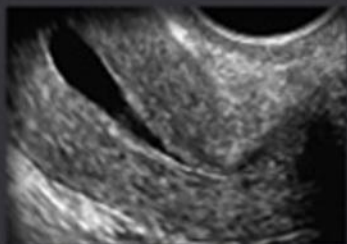


CalLEN'S
ULTRASONOGRAPHY
IN OBSTETRICS
AND GYNECOLOGY

SIXTH EDITION



MARY E.
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To our families for their unwavering support and encouragement, to our residents and fellows who inspire us to be better teachers, to all the contributing authors for their hard work and excellent chapters, to our sonographers for their dedication to patient care, and to our colleagues who constantly stimulate us through their passion for the field of ultrasound.

M.E.N., L.M.S., V.A.F.

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The first edition of this textbook was published well over 30 years ago, when Peter Callen was a young faculty member, we were in medical school, and obstetric sonography was an emerging subspecialty. Whereas the book has grown and evolved through the subsequent editions, it has remained one of the most highly regarded texts in obstetric and gynecologic ultrasound. The three of us grew up reading and learning from Callen's *Ultrasonography in Obstetrics and Gynecology*. Therefore, when we were invited to edit the sixth edition, it was an opportunity that we were eager to embrace, daunting though it was to follow in Peter's footsteps.

Over the years, many aspects of obstetric and gynecologic ultrasound have changed. Ultrasound technology and equipment continue to improve. The anatomic detail that can be visualized and the physiologic parameters that can be assessed have expanded the utility, as well as the complexity, of ultrasound imaging. As in other areas of medicine, care of the obstetric and gynecologic patient in today's practice has advanced to involve much more collaboration across disciplines. This sixth edition reflects this change and was truly a multidisciplinary effort reflecting contributions from experts in obstetrics and gynecology, maternal-fetal medicine, and diagnostic radiology and biomedical imaging. Mary recruited authors from the fields of obstetrics and maternal-fetal medicine and edited chapters on obstetric and fetal sonography. Leslie served as the gynecology editor, managing the recruitment of authors and editing of chapters focused on gynecologic imaging. Vickie helped ensure that the ultrasound images, diagrams, and medical illustrations were of the highest quality. The three of us have been fortunate to have been trained by, and subsequently worked alongside, some renowned leaders in the field of

obstetric and gynecologic imaging. Many of these colleagues and friends, names well known to those in this arena, have contributed to this textbook. The updated information highlights the clinical context and impact of imaging findings. We believe this approach has added to the richness, breadth, and depth of the content of this new edition.

The prior editions of this textbook were edited by Peter with meticulous care and attention. He was always mindful of including authors who are respected experts in their fields and whose writing is authoritative and clear. He ensured that each edition was a substantial update from the previous version and that each chapter was well and liberally illustrated with high-quality sonographic images, drawings, and diagrams to help clarify concepts and illustrate ideas. We aimed to follow his lead and have greatly benefited from his experience, advice, and help throughout this process.

In addition to Peter, who taught us so much and had faith that we could successfully carry on this tradition, there are many others to whom thanks is due. First and foremost, we want to acknowledge and thank all the authors who contributed such outstanding chapters, representing many hours of dedication and effort. The staff at Elsevier, including Taylor Ball in particular, was very helpful and patient as we moved through the production process. Many sonographers and sonologist colleagues helped us collect high-quality images. Finally and importantly, we must thank our families who tolerated so many late nights, early mornings, and weekends spent writing, editing, and tending to this text.

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and Vickie A. Feldstein, MD**

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ULTRASOUND OF THE EARLY FIRST TRIMESTER

Sonography Showing Heartbeat

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CALLEN'S
ULTRASONOGRAPHY
IN OBSTETRICS
AND GYNECOLOGY

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SECTION I

Obstetrics

Obstetric Ultrasound Examination

Peter W. Callen, Mary E. Norton

SUMMARY OF KEY POINTS

- Recent years have seen dramatic advances in ultrasound technology, including improved spatial and contrast resolution, routine use of three-dimensional (3D) and four-dimensional (4D) imaging, volumetric scanning, expanded indications for color and spectral Doppler, new and improved ultrasound scanning probes, and improved digital review workstations.
- With improved imaging comes the complicating corollary as to what minor findings should be reported to the patient and which merely lead to unnecessary anxiety.
- Although there is high-quality evidence that ultrasound is safe for the fetus when used appropriately, consensus statements conclude that Doppler examination of fetal vessels in early pregnancy should not be performed without a clinical indication.
- The nonmedical use of ultrasound for psychosocial or entertainment purposes is strongly discouraged by professional organizations such as the American Institute of Ultrasound in Medicine (AIUM).
- Only those with adequate training in a conventional training program should be performing and interpreting ultrasound examinations.
- Consensus guidelines and criteria for transvaginal sonographic diagnosis of pregnancy failure in a woman with an intrauterine pregnancy of uncertain viability have been established and should be followed.
- Although early detection of a morphologic abnormality is useful, the confident unequivocal detection of an abnormality is even more important. Unless one is extremely confident of the existence of an abnormality in the first trimester, a follow-up examination should be performed.
- Measurements made early in pregnancy, for the most part, are more accurate than those made near term.
- Although a diagnosis of oligohydramnios and polyhydramnios can be made subjectively, the extremes of amniotic fluid volume should also be assessed objectively using either the deepest vertical pocket (DVP) or amniotic fluid index (AFI).
- It is preferable to report the distance from the inferior edge of the placenta to the internal cervical os rather than relying on terms that may have differing meanings (e.g., marginal placenta).
- If a single obstetric ultrasound or a targeted examination is performed, it should be done at a gestational age of 18 to 20 weeks.
- Obstetric ultrasound examinations represented the majority of medical malpractice cases involving ultrasound.
- Ultrasound examination is a noninvasive, safe procedure that has a high degree of patient acceptance and can yield a wealth of information.

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It has now been over 4 decades since sonography was first used to evaluate the obstetric patient. At first, the questions this modality sought to answer were very basic: Is there a pregnancy? Is the fetus alive? Is there a singleton or a twin gestation? What is the location of the placenta? What is the gestational age? Probably, few envisioned the day when ultrasonography would be used to identify subtle anatomic defects such as cleft lip or palate, to predict obstetric complications such as placenta accreta, or to accurately detect the presence of fetal anemia. It is hard to believe that, at its inception, it was difficult to convince clinicians as to the usefulness of this new diagnostic modality in obstetric management. Now, it is routine for a patient to have at least one, and often several, ultrasound examinations during her pregnancy. The technologic advances in ultrasound imaging, including 3D/4D and volumetric measurements, the use of high-frequency transvaginal probes, and the utility for chromosomal screening in early pregnancy (e.g., nuchal translucency) have only expanded the indications for sonographic imaging in the obstetric patient.

Since the last edition of this textbook, there have been dramatic advances in ultrasound technology, including improved spatial and contrast resolution, routine use of 3D and 4D imaging, volumetric scanning, expanded indications for color and spectral Doppler, new and improved ultrasound scanning probes, and improved digital review workstations, to name a few. Likewise, our knowledge of normal fetal anatomy and pathology, and the pathophysiology of disease in general, has increased substantially. The Internet has made communication among and between researchers and clinicians easier. In addition, there have been many collaborative studies and refinements of the guidelines for the performance of the obstetric ultrasound examination. However, there are still differences in the approach to the obstetric ultrasound examination from one group to the next. Although guidelines have improved consistency in the conduct and reporting of obstetric examinations, several issues are often hotly debated: for example, what constitutes a basic ultrasound examination, what structures should be evaluated, what is the ideal timing of the examination, what is the appropriate role of the first trimester anatomic survey, who should perform and interpret the examination, how safe is ultrasound, how should it be recorded and documented, how should it be reported, and last, how should the patient be told the results of the examination? With improved images comes the complicating corollary as to what minor findings should be reported to the patient and which merely lead to unnecessary anxiety. These issues are addressed later in the text and some of them are discussed here.

SAFETY OF ULTRASOUND EXAMINATION

It was not long after the inception of ultrasound imaging that questions were raised as to the safety of this new modality. Despite numerous claims for the safety of ultrasound to the mother and fetus, a number of studies have noted possible adverse effects of diagnostic ultrasound to the developing fetus. These studies have focused primarily on thermal and cavitation mechanisms leading to possible injuries to the developing fetus.¹⁻⁵

Absorption of the ultrasound wave's energy by soft tissue and bone, and its conversion to heat, are measured by the thermal index (TI). A TI of 1 means an increase of 1° C. Several studies have suggested a general threshold of temperature elevation of 1.5° to 2° C above maternal core temperature before any evidence of a developmental effect occurs. With modern ultrasound machines, there is only a negligible rise in temperature, usually less than 1° C. The World Federation for Ultrasound in Medicine and Biology has stated that "a diagnostic exposure that produces a maximum in situ temperature rise of not more than 1.5° C above normal physiological levels may be used

without reservation on thermal grounds."⁶ However, this organization further stated that "a diagnostic exposure that elevates embryonic and fetal in situ temperature above 41° C for 5 minutes should be considered potentially hazardous."⁶ The conclusion overall is that it is unlikely that there is a deleterious effect of ultrasound in the first trimester during embryogenesis with routine gray-scale ultrasound.

However, when Doppler ultrasound is used during the first trimester, it is likely that temperature increases of over 1.5° C may occur.¹ Studies of the effect of Doppler on soft tissues adjacent to bone and nerve conductance demonstrated a significant rise in temperature when the ultrasound Doppler beam was held for more than 30 seconds.^{1,7} The European Federation for Societies in Medicine and Biology in 1998 concluded that "until further scientific information is available, investigations using pulsed or color Doppler should be carried out with careful control for output levels."^{1,8} It is recommended that when performing Doppler imaging in early pregnancy, the displayed TI should be 1.0 or less and exposure time should be kept as short as possible, usually no longer than 5 to 10 minutes and not exceeding 60 minutes.⁹ A thermal index for soft tissue (TIs) should be used at earlier than 10 weeks' gestation, and a thermal index for bone (TIb) should be used at 10 weeks' gestation or later when bone ossification is evident. In keeping with the ALARA (As Low As Reasonably Achievable) principle of prudent scanning, M-mode imaging should be used instead of spectral Doppler imaging to document embryonic/fetal heart rate.¹⁰ Transvaginal ultrasound is not more harmful than transabdominal scanning; again, the risk is dependent on the TI.

Although the potential for embryonic effects from Doppler imaging exists, there is little evidence that ultrasound is teratogenic. As stated in one editorial on the subject, "Many of the studies to date have shown the embryo to be remarkably resilient to ultrasound exposure. Logic would suggest that Doppler techniques should not affect the embryo if the pulses are applied at a low level."¹¹ In one study, Zhu and associates¹² insonated pregnant rats with diagnostic levels of color Doppler ultrasound energy and studied the cell cycles of newborn rats by flow cytometry and factorial analysis. They found that the deoxyribonucleic acid content was not affected in any phase of the cell cycle in newborn rats by any of the different insonation times and frequencies. In another animal study, Pellicer and colleagues examined cellular damage in rats following exposure to low-intensity ultrasound for as long as 10 minutes. These investigators found that the longer the exposure time, the greater the liver cell damage that was observed.¹³ Other animal studies have likewise demonstrated a relationship between the length of exposure to Doppler ultrasound and potential effects on the developing brain.^{14,15} Although such studies support caution and minimizing unnecessary exposure, it is unclear whether such animal models can be extrapolated to humans and if such findings are important. However, at the present time, consensus statements conclude that Doppler examination of fetal vessels in early pregnancy should not be performed without a clinical indication.¹⁶

Cavitation involves the occurrence of gaseous bubble formation in an air-water interface.¹ One concern is that stress from the fluid adjacent to the gaseous body during the process of cavitation may disrupt cell membranes.^{1,17} Cavitation has been difficult to document in mammalian fetuses, because, for the most part, there is not an air-water interface, which is needed for the cavitation mechanism.¹ The mechanical index (MI) is an onscreen indicator that provides a rough guide to the likelihood that ultrasound will induce an adverse biologic effect by a nonthermal mechanism, including cavitation. For all practical purposes, this index is probably not relevant for obstetric scanning owing to the relative absence of gas bubbles (air) in the fetus.¹⁸

A number of studies have evaluated the effect of prenatal ultrasound on neonatal and infant outcome in animal models. Although some studies have documented lower birth weights, shorter heights, and lowered white blood cell counts in neonates who were scanned in utero compared with control subjects, the size differences disappeared when studied after 3 months. In addition, hematologic parameters normalized by this time.¹⁹ Neurodevelopmental studies revealed no significant differences in motor or cognitive tasks, or in learning skills.^{1,19} Studies evaluating the human fetus and neonate have reached similar conclusions. Studies have found either no difference in birth weights between exposed and nonexposed fetuses, or a difference that, although present at birth, was not present at 6 to 7 years of age.^{20,21}

The information on an association between ultrasound and congenital malformations is limited. Studies evaluating chromosomal aberration and ultrasound exposure have demonstrated little or no change.^{1,12,22}

The major difficulties with the studies investigating a possible deleterious effect of diagnostic ultrasound evaluation are threefold: (1) experimental ultrasound exposure levels or time of exposure often far exceeded those that are normally used diagnostically; (2) the systems used to show ultrasound effect (plants, cell culture, laboratory animals) may not be applicable to humans; and (3) many studies that have demonstrated adverse effects in vitro have not been reproducible.²³

One study evaluating the effect of diagnostic ultrasound on neuronal migration in mice raised much attention in the media.¹⁵ Although this is an interesting study in mice, for the reasons stated earlier, it has little to no applicability in humans. There are two major criticisms of this study. Although the study did use commercially available equipment in which only slightly greater ultrasound frequency than normal was used (6.7 MHz vs. 3.5-5.0 MHz), the fixed duration of exposure far exceeded what would normally be used in humans. The study did not demonstrate statistically significant abnormal results until 30 minutes of exposure. More than 10 years ago, when a sonologist wished to determine whether an embryo was nonviable, the recommended evaluation of the embryo was 3 minutes of observation, demonstrating no evidence of embryonic or cardiac activity. Two minutes of evaluation, let alone 3 minutes, seemed like an eternity, and most examiners, in our experience, stopped after 1 minute. In the slightly older embryo and early fetus, greater than 5 to 10 minutes of sustained evaluation of the fetal brain would be excessive. In most cases, the transducer would be moving around the brain rather than being in a fixed position during the examination.

The second criticism relates to the timing of embryology and the relative sizes of the mouse and human brains. As stated by the authors of this study: "The duration of neuronal production and the migratory phase of cortical neurons in the human fetus lasts approximately 18 times longer than in mice (between 6 and 24 weeks of gestation in humans, with the peak occurring between 11 and 15 weeks, compared with the duration of only approximately 1 week (between E11 and E18) in a mouse).^{15,24,25} Thus an exposure of 30 minutes represents a much smaller time dedicated to the development of the cerebral cortex in the human than in the mouse, and thus could have a lesser overall effect, making human corticogenesis less vulnerable to ultrasound waves."

The AIUM statement on the clinical safety of diagnostic ultrasound reiterates previous findings that no confirmed bioeffects caused by exposure at intensities typical of present diagnostic instruments have ever been reported in patients or instrument operators.²⁶ This statement acknowledges the possibility that bioeffects may be identified in the future but emphasizes the current data indicating that the benefits of the prudent use of diagnostic sonography outweigh the risks, if any.^{26,27} At a recent consensus conference on fetal imaging, it was

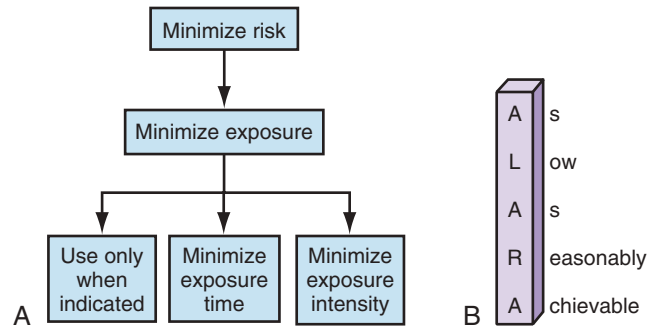


FIG 1-1 Minimizing risk by minimizing exposure (A) is the cornerstone of the ALARA principle (B). (From Kremkau FW [ed]: *Diagnostic Ultrasound: Principles and Instruments*, 7th ed. Philadelphia, WB Saunders, 2006.)

concluded that there is high-quality evidence that ultrasound is safe for the fetus when used appropriately.²⁸ However, as stated by Kremkau,²⁹ "even if this risk is so minimal that it is difficult to identify, prudent practice dictates that routine measures be implemented to minimize the risk while obtaining the necessary information to achieve the diagnostic benefit. This is the ALARA (as low as reasonably achievable) principle of prudent scanning" (Fig. 1-1).

The sonographer's knowledge of ultrasound and its safety is crucial to the safe implementation of this modality. Merritt, in an editorial, summarized it best: "In view of the rapid growth of sonography and its proliferation into the hands of minimally trained clinicians, it is likely that more patients are harmed each day by misdiagnosis resulting from improper indications, poor examination technique, and errors in interpretation than from all bioeffects."²⁷

INDICATIONS FOR OBSTETRIC ULTRASOUND EXAMINATION

National guidelines from many organizations in the United States and elsewhere, including the American College of Obstetricians and Gynecologists (ACOG), the Royal College of Obstetricians and Gynaecologists, and the Society of Obstetricians and Gynaecologists of Canada, highlight the benefits of obstetric ultrasound examination, including accurate determination of gestational age, fetal number, cardiac activity, placental localization, and diagnosis of major fetal anomalies. Because of these benefits, and because most congenital anomalies occur in patients with no known risk factors, these organizations agree that second trimester ultrasonography should be offered routinely to all pregnant women and should follow specific guidelines.³⁰⁻³² In addition, ACOG recommends that all pregnant women should be offered first trimester screening for aneuploidy, which may include nuchal translucency sonography.³³

The benefit of routine sonography in the detection of fetal anomalies has been debated. Large studies and systematic reviews report detection rates of 16% to 44% of anomalies prior to 24 weeks of gestation.^{31,34,35} Higher detection rates of major and lethal anomalies, as high as 84%, have been reported.³¹ The sensitivity of anomaly detection has been noted to vary with respect to the type of abnormality, patient factors, gestational age, and expertise of the imager.²⁸ Possible explanations for the variance in the detection rate of anomalous fetuses may include (1) differences in neonatal assessment, (2) differences in the definition of a major anomaly, (3) a differing risk status of the population, (4) differences in what is considered a routine or standard sonogram, and (5) the expertise of the examiner.³⁶

Who Should Perform the Ultrasound Examination and How Should It Be Performed?

Theoretically, the answer to who should perform the ultrasound examination should be extremely easy. In fact, it is one of the most controversial issues relating to the ultrasound examination. The answer should be that only those persons who have had adequate training (including didactic as well as supervised “hands-on” experience) should perform and interpret an ultrasound examination.

More than 30 years ago, the Joint Task Group on Training for Diagnosis in Obstetrical and Gynecologic Ultrasound developed guidelines for the post-resident physician who had completed residency programs in either radiology or obstetrics and gynecology that did not provide formal training in obstetric and gynecologic ultrasound evaluation.³⁷ These guidelines have been continuously updated, most recently in 2014, and include a recommendation of a minimum experience in obstetric and gynecologic ultrasound evaluation as well as training that includes basic physics, technique, performance, and interpretation. In addition, the physician should obtain practical and supervised experience (at least 300 examinations) before offering services as a physician competent in diagnostic ultrasound examination. Ongoing experience with at least 170 examinations per year is also recommended.³⁸

The “turf” battles between radiologists and obstetricians as to who should perform the examination are unfortunate. As long as the examining physician is adequately trained and performs the minimum standard obstetric ultrasound examination, as per the guidelines of the American College of Radiology (ACR), AIUM, and ACOG, the specialty of the examiner does not matter.^{39,40} However, we do not believe in the practice of self-referral. Self-referral examinations tend to be performed more and more frequently⁴¹ and are often less “complete” and of a lower quality than when they are performed by a dedicated ultrasound practitioner. Except in localities where there are no diagnostic ultrasound specialists, patients should be referred to practitioners whose major practice is ultrasonography.

Guidelines for the performance of obstetric ultrasound examinations have been published by ACR, ACOG, and AIUM, and components of the standard fetal examination at 18 to 20 weeks of gestation were published in a consensus report by National Institute of Child Health and Human Development (NICHD), Society for Maternal-Fetal Medicine (SMFM), ACOG, ACR, AIUM, Society of Pediatric Radiology (SPR), and Society of Radiologists in Ultrasound (SRU) in 2014.²⁸ AIUM has likewise published guidelines for performance of a detailed fetal anatomic survey, referred to by the billing code 76811.⁴² Although there may be sonologists who exceed these guidelines, the guidelines serve as a minimum standard for practitioners of basic and detailed obstetric ultrasonography.

Nonmedical Use of Ultrasonography

The AIUM has published a “prudent use” statement, which was also endorsed by ACOG. The AIUM advocates the responsible use of diagnostic ultrasonography and strongly discourages its nonmedical use for psychosocial or entertainment purposes. The use of either 2D or 3D ultrasound imaging only to view the fetus, obtain a picture of the fetus, or determine the fetal sex without a medical indication is inappropriate and contrary to responsible medical practice. Although there are no confirmed biologic effects on patients caused by exposures from present diagnostic ultrasound instruments, the possibility exists that such biologic effects may be identified in the future. Thus, ultrasound imaging should be used in a prudent manner to provide medical benefit to the patient.⁴³ This position has been ethically defended.⁴⁴

Terminology

The latest classification⁴⁵ of fetal sonographic examinations by the AIUM, ACR, and ACOG groups the examinations into four major categories: (A) the first trimester ultrasound examination, (B) the standard second or third trimester examination, (C) the limited examination, and (D) specialized examinations. The standard second and third trimester obstetric examination is often referred to as a routine examination, basic examination, Level 1 examination, or complete ultrasound examination. Specialized examinations might include a detailed anatomic examination, as well as fetal Doppler ultrasound, a biophysical profile, a fetal echocardiogram, and additional biometric measurements. A detailed anatomic examination is generally performed when a patient is at high risk for a fetal anomaly, or when an anomaly is suspected on the basis of the history, biochemical abnormalities or abnormal results on other screening tests, or the results of either the limited or standard scan.

It is important to note that although those individuals performing detailed anatomic examinations must be proficient in evaluating patients for congenital anomalies, it is not acceptable for the Level 1 examiner to be unskilled. In an excellent editorial on the subject, Filly⁴⁶ notes that unfortunately some examiners have chosen to use the term “Level 1” as a shield for incompetency. As he states, the Level 1 sonogram is not defined by the technical capability of the examiner, nor by the cost of the sonographic instrumentation employed. In fact, the Level 1 examination “requires a high degree of competency” and should follow the standard second or third trimester obstetric sonographic examination as described in the AIUM/ACR/ACOG guidelines.¹⁰

The specialized examination (CPT 76811) has been referred to as the Level 2 examination, survey examination, or targeted examination. As the AIUM/ACR/ACOG guidelines state, this is a detailed anatomic examination that is performed when an anomaly is suspected on the basis of history, abnormalities detected on prenatal screening tests, or the results of either a limited or standard examination previously performed.⁴⁵

The individuals performing the sonographic examination are referred to as either sonographers or sonologists. Traditionally, the technical component and initial production of images has been the responsibility of the sonographer (nonphysician), and the professional component and interpretation of images has been the responsibility of the sonologist (physician). The degree of collaboration between the two and their degree of involvement in the ultrasound examination vary from locality to locality. In many parts of the world, examinations are performed predominantly by physicians. Although the contribution of sonographers to the ultrasound examination is invaluable, it should be remembered, as stated by the AIUM, that “Ultrasound studies shall be supervised and interpreted by a physician with training and experience in the specific area of sonography. Findings must be recorded and results communicated in a timely fashion to the health care provider responsible for care. Although a sonographer may play a critical role in extracting the information essential to deriving a diagnosis, the rendering of the final diagnosis of ultrasound studies represents the practice of medicine, and, therefore, is the responsibility of the supervising physician.”⁴⁷

Perhaps the least controversial aspect of this discussion should be who should interpret the ultrasound examination. This, we believe, is straightforward. Only those with adequate training in a conventional training programs (e.g., residency) in which there are didactic lectures, hands-on scanning, and physician supervision in the performance and interpretation of cases should be performing and interpreting ultrasound findings. Training by manufacturer’s training specialists or 1- to 2-week mini-courses do not constitute adequate training in ultrasound.

Ultrasound Lexicon

Undoubtedly, hundreds of terms are used in obstetrics and ultrasonography that are either incorrect or confusing. Many of these terms are addressed later in this chapter and in other chapters in this text. Two areas in which terminology is often either misused or misunderstood in obstetric ultrasonography are fetal life and age. The term *viability* is defined as the ability to survive in the extrauterine environment. Even in cases of very late third trimester examinations, this statement cannot be used with complete certainty. We prefer to state that the embryo or fetus is living, if that is the case, and use the term *nonviable* for those embryos or fetuses that either are dead or are not capable of living in the extrauterine environment. Early pregnancy failure is another, perhaps even better, way of communicating this information.

The second often-confused term is gestational age. Taken as it sounds, this term would seem to imply the actual age of the fetus from conception to the present. In fact, this term, which is widely used by obstetricians and sonologists, is most often meant to be synonymous with menstrual age. Menstrual age refers to the length of time calculated from the first day of the last normal menstrual period to the point at which the pregnancy is being assessed. The true age of the embryo or fetus, *fetal age*, is rarely known accurately unless the patient has had assisted fertilization or has extremely regular menstrual periods and the day of conception is known. In general, the fetal age is 2 weeks less than the menstrual age.

In this text, the terms gestational age and menstrual age are used interchangeably. The important point for any examiner to remember is not which term is necessarily preferable but rather that the person interpreting the examination and the physician who ordered the examination both use the same terminology.

Another often-misused term is *fetal pole*. This term should be abandoned. It is most often used to describe the presence of the embryo in the early first trimester sonogram. The embryonic period lasts until the end of the 10th menstrual week; during this time the developing conceptus should be referred to as the embryo. Thereafter, the conceptus should be referred to as a fetus.

THE AMERICAN INSTITUTE OF ULTRASOUND IN MEDICINE GUIDELINES

In 2013 the Practice Guideline for the Performance of an Antepartum Obstetric Ultrasound Examination was updated by the AIUM in collaboration with the ACR, the SRU, and ACOG.⁴⁵ It is a modification of previously developed guidelines that were first published in 1986. The actual ACR/AIUM/ACOG guidelines are presented in [Table 1-1](#). What follows is our own bias as to what constitutes an appropriate ultrasound examination. In some respects, this discussion is an expansion of the guidelines previously mentioned. Because this multiauthor text is essentially a detailed review of the obstetric ultrasound examination, we recognize that our viewpoint in this chapter and those of the authors of the subsequent chapters may differ.

ULTRASOUND EQUIPMENT AND DOCUMENTATION

It seems that there will always be differences of opinion as to which ultrasound machines produce the best images. With the present state of ultrasound technology, these differences are often subjective, particularly when discussing state-of-the-art machines. Most ultrasound machines use phased-array real-time technology and include 3D/4D ultrasound technology and color and pulsed Doppler flow capabilities and cine recording.

An often-debated issue is which transducer should be used for the ultrasound examination. The answer is as many transducers as are necessary should be used to answer the question for which the patient is referred. There is a misconception that the newest transducer introduced by a manufacturer may be the only one that is needed. When sector and, ultimately, transvaginal probes were first introduced, many practitioners believed that these transducers alone could be used for the entire examination. Many learned that using only a single transducer restricts the field of view or visualization of detail, making diagnosis more difficult.

The most common transducers, which are the workhorses of the ultrasound laboratory, are a convex linear array, a sector transducer (3 to 5 MHz), and a transvaginal probe (5 to 10 MHz). The higher frequency transducers are most useful in achieving high-resolution scans, particularly in the near-field, and the lower frequency transducers are useful in those circumstances in which increased penetration of the sound beam is necessary. Variations of transducer technology include convex linear transducers and multifrequency probes as well as probes allowing harmonic and 3D imaging and Doppler flow imaging.⁴⁸

Whatever technology is used, images from the examination should be documented and stored. The purpose of documentation is twofold. First, the identification of normal structures is important so they can be viewed retrospectively and compared with later images if pathologic processes are ultimately demonstrated. Second, if a pathologic problem is identified, it can be shown to referring examiners, who will be doing further examinations.

Most imaging centers utilize picture archiving and communication systems (PACS). These systems allow for the storage of digital ultrasound images on computers and transmission of complete studies to computer workstations for viewing and interpretation. The quality of the images is excellent with these systems. Digital images can be also transmitted to remote locations (telemedicine) for review and consultation.⁴⁹

In addition to digitally stored images, a written report of the ultrasound examination should be included in the patient's medical record. When significant pathologic processes are present, the referring physician should be notified immediately. This immediate communication should occur not only in cases of fetal malformations but also in cases of serious obstetric complications, such as oligohydramnios, diminished fetal movement, macrosomia, and fetal growth restriction. Physician notification should be documented.

THE FIRST TRIMESTER ULTRASOUND EXAMINATION

Identification of an Intrauterine Pregnancy

Patients referred for first trimester ultrasound evaluation often have vaginal bleeding, which raises the question of an ectopic pregnancy or a threatened abortion. The primary goal of ultrasound evaluation in the first trimester is to determine whether the pregnancy is intrauterine and whether the embryo is living. With present-day equipment, particularly transvaginal transducers, both of these tasks should be readily accomplished at very early stages of gestation. The same care taken in concluding that a pregnancy in the second or third trimester has a lethal malformation should be applied in deciding that an early pregnancy is nonviable. If there is reasonable doubt about embryonic life, a repeat examination in as few as 7 to 10 days will invariably make the conclusion unequivocal. In 2012 a consensus conference from the SRU established guidelines and criteria for transvaginal ultrasonographic

Text continued on p. 12

TABLE 1-1 Guidelines for Performance of the Antepartum Obstetric Ultrasound Examination**I. Introduction**

The clinical aspects contained in specific sections of this guideline (Introduction, Classification of Fetal Sonographic Examinations, Specifications of the Examination, Equipment Specifications, and Fetal Safety) were revised collaboratively by the American Institute of Ultrasound in Medicine (AIUM), the American College of Radiology (ACR), the American College of Obstetricians and Gynecologists (ACOG), and the Society of Radiologists in Ultrasound (SRU).

Recommendations for personnel qualifications, written request for the examination, procedure documentation, and quality control vary among the organizations and are addressed by each separately.

This guideline has been developed for use by practitioners performing obstetric sonographic studies. Fetal ultrasound should be performed only when there is a valid medical reason, and the lowest possible ultrasonic exposure settings should be used to gain the necessary diagnostic information. A limited examination may be performed in clinical emergencies or for a limited purpose such as evaluation of fetal or embryonic cardiac activity, fetal position, or amniotic fluid volume. A limited follow-up examination may be appropriate for reevaluation of fetal size or interval growth or to reevaluate abnormalities previously noted if a complete prior examination is on record.

While this guideline describes the key elements of standard sonographic examinations in the first trimester and second and third trimesters, a more detailed anatomic examination of the fetus may be necessary in some cases, such as when an abnormality is found or suspected on the standard examination or in pregnancies at high risk for fetal anomalies. In some cases, other specialized examinations may be necessary as well.

While it is not possible to detect all structural congenital anomalies with diagnostic ultrasound, adherence to the following guidelines will maximize the possibility of detecting many fetal abnormalities.

II. Classification of Fetal Sonographic Examinations**A. First-Trimester Examination**

A standard obstetric sonogram in the first trimester includes evaluation of the presence, size, location, and number of gestational sac(s). The gestational sac is examined for the presence of a yolk sac and embryo/fetus. When an embryo/fetus is detected, it should be measured and cardiac activity recorded by a 2-dimensional video clip or M-mode imaging. Use of spectral Doppler imaging is discouraged. The uterus, cervix, adnexa, and cul-de-sac region should be examined.

B. Standard Second- or Third-Trimester Examination

A standard obstetric sonogram in the second or third trimester includes an evaluation of fetal presentation, amniotic fluid volume, cardiac activity, placental position, fetal biometry, and fetal number, plus an anatomic survey. The maternal cervix and adnexa should be examined as clinically appropriate when technically feasible.

C. Limited Examination

A limited examination is performed when a specific question requires investigation. For example, in most routine nonemergency cases, a limited examination could be performed to confirm fetal heart activity in a bleeding patient or to verify fetal presentation in a laboring patient. In most cases, limited sonographic examinations are appropriate only when a prior complete examination is on record.

D. Specialized Examinations

A detailed anatomic examination is performed when an anomaly is suspected on the basis of the history, biochemical abnormalities, or the results of either the limited or standard scan. Other specialized examinations might include fetal Doppler ultrasound, a biophysical profile, a fetal echocardiogram, and additional biometric measurements.

III. Qualifications and Responsibilities of Personnel

See the AIUM Official Statement *Training Guidelines for Physicians Who Evaluate and Interpret Diagnostic Abdominal, Obstetric, and/or Gynecologic Ultrasound Examinations* and the *AIUM Standards and Guidelines for the Accreditation of Ultrasound Practices*.

IV. Written Request for the Examination

The written or electronic request for an ultrasound examination should provide sufficient information to allow for the appropriate performance and interpretation of the examination. The request for the examination must be originated by a physician or other appropriately licensed health care provider or under the provider's direction. The accompanying clinical information should be provided by a physician or other appropriate health care provider familiar with the patient's clinical situation and should be consistent with relevant legal and local health care facility requirements.

V. Specifications of the Examination**A. First-Trimester Ultrasound Examination****1. Indications**

Indications for first-trimester sonography include but are not limited to:

- a. Confirmation of the presence of an intrauterine pregnancy;
- b. Evaluation of a suspected ectopic pregnancy;
- c. Defining the cause of vaginal bleeding;
- d. Evaluation of pelvic pain;
- e. Estimation of gestational (menstrual) age;
- f. Diagnosis or evaluation of multiple gestations;
- g. Confirmation of cardiac activity;
- h. Imaging as an adjunct to chorionic villus sampling, embryo transfer, and localization and removal of an intrauterine device;
- i. Assessing for certain fetal anomalies, such as anencephaly, in high-risk patients;
- j. Evaluation of maternal pelvic masses and/or uterine abnormalities;
- k. Measuring the nuchal translucency (NT) when part of a screening program for fetal aneuploidy; and
- l. Evaluation of a suspected hydatidiform mole.

Comment

A limited examination may be performed to evaluate interval growth, estimate amniotic fluid volume, evaluate the cervix, and assess the presence of cardiac activity.

TABLE 1-1 Guidelines for Performance of the Antepartum Obstetric Ultrasound Examination—cont'd

2. Imaging Parameters

Comment

Scanning in the first trimester may be performed either transabdominally or transvaginally. If a transabdominal examination is not definitive, a transvaginal scan or transperineal scan should be performed whenever possible.

- a. The uterus (including the cervix) and adnexa should be evaluated for the presence of a gestational sac. If a gestational sac is seen, its location should be documented. The gestational sac should be evaluated for the presence or absence of a yolk sac or embryo, and the crown-rump length should be recorded when possible.

Comment

A definitive diagnosis of intrauterine pregnancy can be made when an intrauterine gestational sac containing a yolk sac or embryo/fetus with cardiac activity is visualized. A small, eccentric intrauterine fluid collection with an echogenic rim can be seen before the yolk sac and embryo are detectable in a very early intrauterine pregnancy. In the absence of sonographic signs of ectopic pregnancy, the fluid collection is highly likely to represent an intrauterine gestational sac. In this circumstance, the intradecidual sign may be helpful. Follow-up sonography and/or serial determination of maternal serum human chorionic gonadotropin levels are/is appropriate in pregnancies of undetermined location to avoid inappropriate intervention in a potentially viable early pregnancy.

The crown-rump length is a more accurate indicator of gestational (menstrual) age than is the mean gestational sac diameter. However, the mean gestational sac diameter may be recorded when an embryo is not identified.

Caution should be used in making the presumptive diagnosis of a gestational sac in the absence of a definitive embryo or yolk sac. Without these findings, an intrauterine fluid collection could represent a pseudo-gestational sac associated with an ectopic pregnancy.

- b. The presence or absence of cardiac activity should be documented with a 2-dimensional video clip or M-mode imaging.

Comment

With transvaginal scans, while cardiac motion is usually observed when the embryo is 2 mm or greater in length, if an embryo less than 7 mm in length is seen without cardiac activity, a subsequent scan in 1 week is recommended to ensure that the pregnancy is nonviable.

- c. Fetal number should be documented.

Comment

Amnionicity and chorionicity should be documented for all multiple gestations when possible.

- d. Embryonic/fetal anatomy appropriate for the first trimester should be assessed.

- e. The nuchal region should be imaged, and abnormalities such as cystic hygroma should be documented.

Comment

For those patients desiring to assess their individual risk of fetal aneuploidy, a very specific measurement of the NT during a specific age interval is necessary (as determined by the laboratory used). See the guidelines for this measurement below.

NT measurements should be used (in conjunction with serum biochemistry) to determine the risk of having a fetus with aneuploidy or other anatomic abnormalities such as heart defects. In this setting, it is important that the practitioner measure the NT according to established guidelines for measurement. A quality assessment program is recommended to ensure that false-positive and false-negative results are kept to a minimum.

Guidelines for NT Measurement:

- i. The margins of the NT edges must be clear enough for proper placement of the calipers.
- ii. The fetus must be in the midsagittal plane.
- iii. The image must be magnified so that it is filled by the fetal head, neck, and upper thorax.
- iv. The fetal neck must be in a neutral position, not flexed and not hyperextended.
- v. The amnion must be seen as separate from the NT line.
- vi. The + calipers on the ultrasound must be used to perform the NT measurement.
- vii. Electronic calipers must be placed on the inner borders of the nuchal line space with none of the horizontal crossbar itself protruding into the space.
- viii. The calipers must be placed perpendicular to the long axis of the fetus.
- ix. The measurement must be obtained at the widest space of the NT.

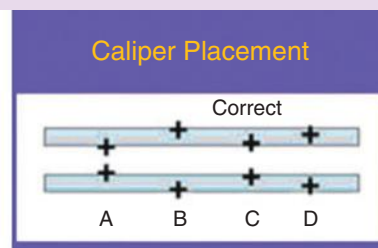


TABLE 1-1 Guidelines for Performance of the Antepartum Obstetric Ultrasound Examination—cont'd

- f. The uterus including the cervix, adnexal structures, and cul-de-sac should be evaluated. Abnormalities should be imaged and documented.
- Comment
The presence, location, appearance, and size of adnexal masses should be documented. The presence and number of leiomyomata should be documented. The measurements of the largest or any potentially clinically significant leiomyomata should be documented. The cul-de-sac should be evaluated for the presence or absence of fluid. Uterine anomalies should be documented.

B. Second- and Third-Trimester Ultrasound Examination**1. Indications**

Indications for second- and third-trimester sonography include but are not limited to:

- a. Screening for fetal anomalies;
- b. Evaluation of fetal anatomy;
- c. Estimation of gestational (menstrual) age;
- d. Evaluation of fetal growth;
- e. Evaluation of vaginal bleeding;
- f. Evaluation of abdominal or pelvic pain;
- g. Evaluation of cervical insufficiency;
- h. Determination of fetal presentation;
- i. Evaluation of suspected multiple gestation;
- j. Adjunct to amniocentesis or other procedure;
- k. Evaluation of a significant discrepancy between uterine size and clinical dates;
- l. Evaluation of a pelvic mass;
- m. Evaluation of a suspected hydatidiform mole;
- n. Adjunct to cervical cerclage placement;
- o. Suspected ectopic pregnancy;
- p. Suspected fetal death;
- q. Suspected uterine abnormalities;
- r. Evaluation of fetal well-being;
- s. Suspected amniotic fluid abnormalities;
- t. Suspected placental abruption;
- u. Adjunct to external cephalic version;
- v. Evaluation of premature rupture of membranes and/or premature labor;
- w. Evaluation of abnormal biochemical markers;
- x. Follow-up evaluation of a fetal anomaly;
- y. Follow-up evaluation of placental location for suspected placenta previa;
- z. History of previous congenital anomaly;
 - aa. Evaluation of the fetal condition in late registrants for prenatal care; and
 - bb. Assessment for findings that may increase the risk for aneuploidy.

Comment

In certain clinical circumstances, a more detailed examination of fetal anatomy may be indicated.

2. Imaging Parameters for a Standard Fetal Examination

- a. Fetal cardiac activity, fetal number, and presentation should be documented.

Comment
An abnormal heart rate and/or rhythm should be documented. Multiple gestations require the documentation of additional information: chorionicity, amnionicity, comparison of fetal sizes, estimation of amniotic fluid volume (increased, decreased, or normal) in each gestational sac, and fetal genitalia (when visualized).
- b. A qualitative or semiquantitative estimate of amniotic fluid volume should be documented.

Comment
Although it is acceptable for experienced examiners to qualitatively estimate amniotic fluid volume, semiquantitative methods have also been described for this purpose (eg, amniotic fluid index, single deepest pocket, and 2-diameter pocket).
- c. The placental location, appearance, and relationship to the internal cervical os should be documented. The umbilical cord should be imaged and the number of vessels in the cord documented. The placental cord insertion site should be documented when technically possible.

Comment
It is recognized that the apparent placental position early in pregnancy may not correlate well with its location at the time of delivery. Transabdominal, transperineal, or transvaginal views may be helpful in visualizing the internal cervical os and its relationship to the placenta. Transvaginal or transperineal ultrasound may be considered if the cervix appears shortened or cannot be adequately visualized during the transabdominal sonogram.
A velamentous (also called membranous) placental cord insertion that crosses the internal os of the cervix is vasa previa, a condition that has a high risk of fetal mortality if not diagnosed before labor.

Continued

TABLE 1-1 Guidelines for Performance of the Antepartum Obstetric Ultrasound Examination—cont'd

d. Gestational (menstrual) age assessment.

First-trimester crown-rump measurement is the most accurate means for sonographic dating of pregnancy. Beyond this period, a variety of sonographic parameters such as biparietal diameter, abdominal circumference, and femoral diaphysis length can be used to estimate gestational (menstrual) age. The variability of gestational (menstrual) age estimation, however, increases with advancing pregnancy. Significant discrepancies between gestational (menstrual) age and fetal measurements may suggest the possibility of a fetal growth abnormality, intrauterine growth restriction, or macrosomia.

Comment

The pregnancy should not be redated after an accurate earlier scan has been performed and is available for comparison.

- i.** The biparietal diameter is measured at the level of the thalami and cavum septi pellucidi or columns of the fornix. The cerebellar hemispheres should not be visible in this scanning plane. The measurement is taken from the outer edge of the proximal skull to the inner edge of the distal skull.

Comment

The head shape may be flattened (dolichocephaly) or rounded (brachycephaly) as a normal variant. Under these circumstances, certain variants of normal fetal head development may make measurement of the head circumference more reliable than biparietal diameter for estimating gestational (menstrual) age.

- ii.** The head circumference is measured at the same level as the biparietal diameter, around the outer perimeter of the calvarium. This measurement is not affected by head shape.
- iii.** The femoral diaphysis length can be reliably used after 14 weeks' gestational (menstrual) age. The long axis of the femoral shaft is most accurately measured with the beam of insonation being perpendicular to the shaft, excluding the distal femoral epiphysis.
- iv.** The abdominal circumference or average abdominal diameter should be determined at the skin line on a true transverse view at the level of the junction of the umbilical vein, portal sinus, and fetal stomach when visible.

Comment

The abdominal circumference or average abdominal diameter measurement is used with other biometric parameters to estimate fetal weight and may allow detection of intrauterine growth restriction or macrosomia.

e. Fetal weight estimation.

Fetal weight can be estimated by obtaining measurements such as the biparietal diameter, head circumference, abdominal circumference or average abdominal diameter, and femoral diaphysis length. Results from various prediction models can be compared to fetal weight percentiles from published nomograms.

Comment

If previous studies have been performed, appropriateness of growth should also be documented. Scans for growth evaluation can typically be performed at least 2 to 4 weeks apart. A shorter scan interval may result in confusion as to whether measurement changes are truly due to growth as opposed to variations in the technique itself.

Currently, even the best fetal weight prediction methods can yield errors as high as $\pm 15\%$. This variability can be influenced by factors such as the nature of the patient population, the number and types of anatomic parameters being measured, technical factors that affect the resolution of ultrasound images, and the weight range being studied.

f. Maternal anatomy.

Evaluation of the uterus, adnexal structures, and cervix should be performed when appropriate. If the cervix cannot be visualized, a transperineal or transvaginal scan may be considered when evaluation of the cervix is needed.

Comment

This will allow recognition of incidental findings of potential clinical significance. The presence, location, and size of adnexal masses and the presence of at least the largest and potentially clinically significant leiomyomata should be documented. It is not always possible to image the normal maternal ovaries during the second and third trimesters.

g. Fetal anatomic survey.

Fetal anatomy, as described in this document, may be adequately assessed by ultrasound after approximately 18 weeks' gestational (menstrual) age. It may be possible to document normal structures before this time, although some structures can be difficult to visualize due to fetal size, position, movement, abdominal scars, and increased maternal abdominal wall thickness. A second- or third-trimester scan may pose technical limitations for an anatomic evaluation due to imaging artifacts from acoustic shadowing. When this occurs, the report of the sonographic examination should document the nature of this technical limitation. A follow-up examination may be helpful. The following areas of assessment represent the minimal elements of a standard examination of fetal anatomy. A more detailed fetal anatomic examination may be necessary if an abnormality or suspected abnormality is found on the standard examination.

i. Head, face, and neck:

Lateral cerebral ventricles;

Choroid plexus;

Midline falx;

Cavum septi pellucidi;

Cerebellum;

Cistern magna; and

Upper lip.

Comment

A measurement of the nuchal fold may be helpful during a specific age interval to assess the risk of aneuploidy.

TABLE 1-1 Guidelines for Performance of the Antepartum Obstetric Ultrasound Examination—cont'd

- ii. Chest:
 - Heart:
 - Four-chamber view;
 - Left ventricular outflow tract; and
 - Right ventricular outflow tract.
- iii. Abdomen:
 - Stomach (presence, size, and situs);
 - Kidneys;
 - Urinary bladder;
 - Umbilical cord insertion site into the fetal abdomen; and
 - Umbilical cord vessel number.
- iv. Spine:
 - Cervical, thoracic, lumbar, and sacral spine.
- v. Extremities:
 - Legs and arms.
- vi. Sex:
 - In multiple gestations and when medically indicated.

VI. Documentation

Adequate documentation is essential for high-quality patient care. There should be a permanent record of the ultrasound examination and its interpretation.

Images of all appropriate areas, both normal and abnormal, should be recorded. Variations from normal size should be accompanied by measurements. Images should be labeled with the patient identification, facility identification, examination date, and side (right or left) of the anatomic site imaged. An official interpretation (final report) of the ultrasound findings should be included in the patient's medical record. Retention of the ultrasound examination should be consistent both with clinical needs and with relevant legal and local health care facility requirements.

Reporting should be in accordance with the *AIUM Practice Guideline for Documentation of an Ultrasound Examination*.

VII. Equipment Specifications

These studies should be conducted with real-time scanners, using a transabdominal and/or transvaginal approach. A transducer of appropriate frequency should be used. Real-time sonography is necessary to confirm the presence of fetal life through observation of cardiac activity and active movement.

The choice of transducer frequency is a trade-off between beam penetration and resolution. With modern equipment, 3- to 5-MHz abdominal transducers allow sufficient penetration in most patients while providing adequate resolution. A lower-frequency transducer may be needed to provide adequate penetration for abdominal imaging in an obese patient. During early pregnancy, a 5-MHz abdominal transducer or a 5- to 10-MHz or greater vaginal transducer may provide superior resolution while still allowing adequate penetration.

VIII. Fetal Safety

Diagnostic ultrasound studies of the fetus are generally considered safe during pregnancy. This diagnostic procedure should be performed only when there is a valid medical indication, and the lowest possible ultrasonic exposure setting should be used to gain the necessary diagnostic information under the ALARA (as low as reasonably achievable) principle.

A thermal index for soft tissue (Tis) should be used at earlier than 10 weeks' gestation, and a thermal index for bone (Tib) should be used at 10 weeks' gestation or later when bone ossification is evident. In keeping with the ALARA principle, M-mode imaging should be used instead of spectral Doppler imaging to document embryonic/fetal heart rate.

The promotion, selling, or leasing of ultrasound equipment for making "keepsake fetal videos" is considered by the US Food and Drug Administration to be an unapproved use of a medical device. Use of a diagnostic ultrasound system for these purposes, without a physician's order, may be in violation of state laws or regulations.

IX. Quality Control and Improvement, Safety, Infection Control, and Patient Education

Policies and procedures related to quality control, patient education, infection control, and safety should be developed and implemented in accordance with the *AIUM Standards and Guidelines for the Accreditation of Ultrasound Practices*.

Equipment performance monitoring should be in accordance with the *AIUM Standards and Guidelines for the Accreditation of Ultrasound Practices*.

X. ALARA Principle

The potential benefits and risks of each examination should be considered. The ALARA principle should be observed when adjusting controls that affect the acoustic output and by considering transducer dwell times. Further details on ALARA may be found in the AIUM publication *Medical Ultrasound Safety*, Second Edition.

Modified from American Institute of Ultrasound in Medicine: AIUM practice guideline for the performance of obstetric ultrasound examinations. *J Ultrasound Med* 32(6):1083-1101, 2013.

TABLE 1-2 Guidelines for Transvaginal Ultrasonographic Diagnosis of Pregnancy Failure in a Woman With an Intrauterine Pregnancy of Uncertain Viability*

Findings Diagnostic of Pregnancy Failure	Findings Suspicious for, But Not Diagnostic of, Pregnancy Failure [†]
Crown-rump length of ≥ 7 mm and no heartbeat	Crown-rump length of < 7 mm and no heartbeat
Mean sac diameter of ≥ 25 mm and no embryo	Mean sac diameter of 16-24 mm and no embryo
Absence of embryo with heartbeat ≥ 2 wk after a scan that showed a gestational sac without a yolk sac	Absence of embryo with heartbeat 7-13 days after a scan that showed a gestational sac without a yolk sac
Absence of embryo with heartbeat ≥ 11 days after a scan that showed a gestational sac with a yolk sac	Absence of embryo with heartbeat 7-10 days after a scan that showed a gestational sac with a yolk sac
	Absence of embryo ≥ 6 wk after last menstrual period
	Empty amnion (amnion seen adjacent to yolk sac, with no visible embryo)
	Enlarged yolk sac (> 7 mm)
	Small gestational sac in relation to the size of the embryo (< 5 mm difference between mean sac diameter and crown-rump length)

*Criteria are from the Society of Radiologists in Ultrasound Multispecialty Consensus Conference on Early First Trimester Diagnosis of Miscarriage and Exclusion of a Viable Intrauterine Pregnancy, October 2012.

[†]When there are findings suspicious for pregnancy failure, follow-up ultrasonography at 7 to 10 days to assess the pregnancy for viability is generally appropriate.

diagnosis of pregnancy failure in a woman with an intrauterine pregnancy of uncertain viability (see [Table 1-2](#) and [Chapter 4](#)).

Embryonic/Fetal Number

With a careful examination, the true number of embryos can be accurately determined even early in the first trimester. The literature has emphasized that it is important not to overestimate the number of developing gestations by misinterpreting findings such as a double sac sign, fluid in the uterine cavity, the yolk sac, or the presence of the amnion as evidence of multiple sacs or embryos and thus multiple gestations. However, the examiner may be just as likely to underestimate the number of developing gestations and embryos if a thorough evaluation of the gestational sac is not made for all embryos.⁵⁰ It is our experience that when multiple gestations are missed using ultrasound assessment, it is usually from a less than optimal first trimester examination. The head (crown) of one embryo may be added to the body (rump) of an adjacent embryo and measured as a singleton. This misdiagnosis, of course, occurs only in monochorionic gestations. For these reasons some investigators prefer that if one ultrasound examination is to be done concentrating on fetal number, it should be done in the early to middle second trimester of pregnancy.

Estimating Gestational Age

The subject of estimating gestational age is covered in detail in [Chapter 6](#). An estimate of gestational age should be made when an ultrasound examination is performed in the first trimester, as this is the most accurate time to determine gestational age. The two most common methods of gestational age estimation are mean gestational sac diameter and crown-rump length. For many years, the crown-rump length has been acclaimed as the most reliable method of estimating gestational age in utero. The crown-rump length is a highly accurate method of estimating gestational age using ultrasound evaluation (accuracy within 3 to 7 days). Other measurements, such as the head circumference or femur length, performed in the second trimester, are nearly as accurate and have the added benefit of allowing one to assess fetal morphologic features to a better advantage in a larger fetus. We believe that the first trimester ultrasound examination should not be done for the sole purpose of obtaining more accurate measurements if there is not a clinical reason why it cannot be done in the second trimester, and this approach was also affirmed in the NICHD Fetal Imaging Workshop consensus.²⁸

Morphologic Abnormalities

Recent years have seen continuous improvement in the resolution of ultrasound and many reports documenting morphologic abnormalities detected in the first trimester. Abnormalities involving virtually every organ system have been reported. In light of these reports, we are frequently asked when is the earliest time that a particular abnormality can be detected. Our reply is often that although early detection of a morphologic abnormality is useful, the confident unequivocal detection of an abnormality is even more important. Unless one is extremely confident of the existence of an abnormality in the first trimester, a follow-up examination should be performed.

One should be aware of four potential pitfalls in diagnosis in the first trimester: (1) the normal extra-abdominal position of the embryonic intestine simulating an abdominal wall defect, (2) the prominence of the developing cerebral vesicles (rhombencephalon), (3) the potential false negative diagnosis of anencephaly, and (4) the false positive diagnosis of cerebellar vermian and callosal abnormalities because of these structures not being fully developed at an early gestational age.⁵¹⁻⁵³

Placenta

In early pregnancies, it may be difficult to ascertain the site of the developing placenta. If, however, the examiner can confidently identify the site of placentation, either anterior or posterior, this information should be documented. It should be noted that the placenta either overlies the cervix or just reaches the cervix in up to 2% of pregnancies imaged transvaginally in the early second trimester.^{54,55} Placental "migration," or resolution of placenta previa as pregnancy progresses, occurs in most cases, probably as a result of the faster growth of the placenta-free uterine wall relative to the uterine wall covered by the placenta.⁵⁴ Factors such as prior cesarean delivery and the degree to which the placenta overlies the cervix affect whether placenta previa in the second trimester will resolve prior to delivery. In general, the placenta commonly extends to the cervix before 16 weeks, and a placenta previa should not be reported. At 16 weeks and beyond, if the placental edge is within 2 cm of the internal os, a diagnosis of "low-lying placenta" is made and follow-up at 32 weeks of gestation is recommended.²⁸ If one is uncertain as to the location of the inferior edge of the placenta, a transvaginal scan will help clear up any confusion and prevent a patient being labeled as having a placenta previa.

Uterus and Adnexa

The maternal uterus should be examined carefully for evidence of uterine abnormalities, particularly in high-risk patients. Late in pregnancy, these anomalies may be more difficult to detect. If uterine myomas are detected, their size, site, and relationship to the cervix should be recorded. It should be remembered that transient myometrial contractions may simulate myomas.

The adnexa should be carefully searched for the presence of cysts as well as ovarian neoplasms, both benign and malignant. Again, later in pregnancy, as the adnexal areas are displaced superiorly, they may be more difficult to evaluate adequately.

THE SECOND AND THIRD TRIMESTER ULTRASOUND EXAMINATIONS

Fetal Number and Fetal Life

Evaluating the number of fetuses should be extremely easy and accurate in the second and third trimesters. The increased perinatal morbidity and mortality risks of multiple gestations make it mandatory that a “surprise twin” at delivery be a rare event in any patient who has had a second or third trimester ultrasound examination. The major potential error in determining the number of fetuses is one of underestimation. This mistake, when made, is likely due to either not evaluating the fundal region or not making sure that the fetal head is associated with its body rather than that of a twin. When a multiple gestation is identified, it is important to determine the number of placentas and the number of amniotic sacs (the chorionicity and amnionity).

In the ultrasound report, a statement should be made that the fetus was living, if this was the case, by virtue of cardiac motion being identified. If there is any doubt about fetal life, a confirmatory examination by another examiner should take place. The lack of fetal movement should not be interpreted as representing fetal death. Slow fetal heart rates often portend a poor prognosis; however, this observation alone should not be considered evidence of a nonviable pregnancy. Many cases of fetal heart rates less than 80 beats per minute result in normal outcomes.

Fetal Position

Once fetal life and number have been identified, then the fetal lie and presenting part should be determined in patients beyond 20 weeks of gestation. Fetal lie refers to the relationship of the long axis of the fetus to the long axis of the uterus. Presentation defines the presenting fetal part closest to the cervix. The most common fetal lie is longitudinal, and the most common presenting part is the fetal head. Fetal lie or presentations other than these are referred to as malpresentations. Their significance lies in increased perinatal morbidity during delivery.

The advent of real-time ultrasound evaluation has placed an additional demand on the sonographer. If the sonologist interpreting the scans has not performed the examination, he or she must be able to deduce the lie and presentation from the sonographer's images. This may be done only by understanding the normal fetal anatomy and applying it to the scanning position (Figs. 1-2 and 1-3). Likewise, some congenital anomalies (e.g., dextrocardia, abnormal right-sided abdominal cystic mass) are recognized only fortuitously if a structure is identified as abnormal by virtue of its abnormal position related to the lie and presentation of the fetus.

As mentioned previously, the most common presenting part is the fetal head (cephalic presentation). (We prefer the term *cephalic* rather

than *vertex* because the latter term may also be used to describe a location on the fetal head.) When the head is adjacent to the lower uterine segment, it is likely that the fetus is in cephalic presentation; however, one must see all images before coming to that conclusion. The fetal body may also be low in the uterus with the fetal head, and, thus, the fetus would be in a transverse lie rather than in a cephalic presentation.

Fetal malpresentation requires that the sonographer extend the examination to answer two additional questions important to the referring obstetrician. First, what specifically is the presenting part (i.e., foot, buttocks in the case of a breech presentation, or shoulder in the case of a fetus in transverse lie) (Figs. 1-4 and 1-5)? Second, if a malpresentation persists into the latter third trimester, is there an associated fetal malformation or placental abnormality that may be causally related to the abnormal lie?⁵⁶

Assigning Gestational Age and Weight

The assignment of gestational age and weight is covered in detail in Chapter 6. It is important to remember several concepts when assigning gestational age using ultrasonography. First, measurements made early in pregnancy, for the most part, are more accurate than those made near term. In most cases, the measurements of the fetal head, body, and femur will be concordant with one another, within a week in the early to mid-second trimester. This is often not true in the late third trimester, when the femur may lag behind the other measurements and more variation is common and normal. In the early to mid-second trimester, if the femur or the head measurements are greater than 1 week less than the other measurements, this should raise a red flag and alert one to the possibility of either short-limbed bone dysplasia, trisomy 21, or microcephaly. Follow-up in these cases may be indicated. Second, pathologic states should be taken into consideration when deciding which body parts to use in assigning gestational age or weight. Most ultrasound machines allow the user to eliminate from the gestational age calculation those body parts that are abnormal. The abdominal circumference measurement is likely to be inaccurate in the presence of fetal ascites, and the femur length measurement is unreliable in fetuses with short-limbed dwarfism. Third, every obstetric ultrasound report should relate the calculated sonographic age to the patient's menstrual age or clinical gestational age. Because menstrual histories are frequently inaccurate, there is often a tendency to not believe a woman's menstrual history in deference to the calculated sonographic age. In doing so, however, one runs the risk of assigning an earlier gestational age to a fetus that is in fact older but growth restricted. Likewise, there is the possibility of assigning an earlier gestational age to a pregnancy that is post term, placing the fetus at risk for fetal postmaturity syndrome or in utero death. Fourth, the calculated fetal weight should be stated not only in grams but also as a percentile based on the patient's clinical gestational age or best obstetric estimate. Again, if the patient's menstrual dates are inaccurate, the obstetrician can make the decision not to become alarmed at a reported low-weight percentile. This is far better than misinterpreting a growth-restricted fetus as normal by relating the estimated weight only to the ultrasound-determined age. (Remember that although the formulas are different for the calculations of fetal age and weight, they are based upon the same biometric measurements and when compared to one another will often be near the 50% percentile.) Fifth, if there has been a previous ultrasound examination, there should be some statement in the report as to whether interval fetal growth has been normal or abnormal. Finally, sonograms attempting to assess normal or abnormal interval growth should have an interval of no less than 2 weeks. It may be difficult to determine whether there has been a growth abnormality versus a measurement error if scans are done with a shorter interval.

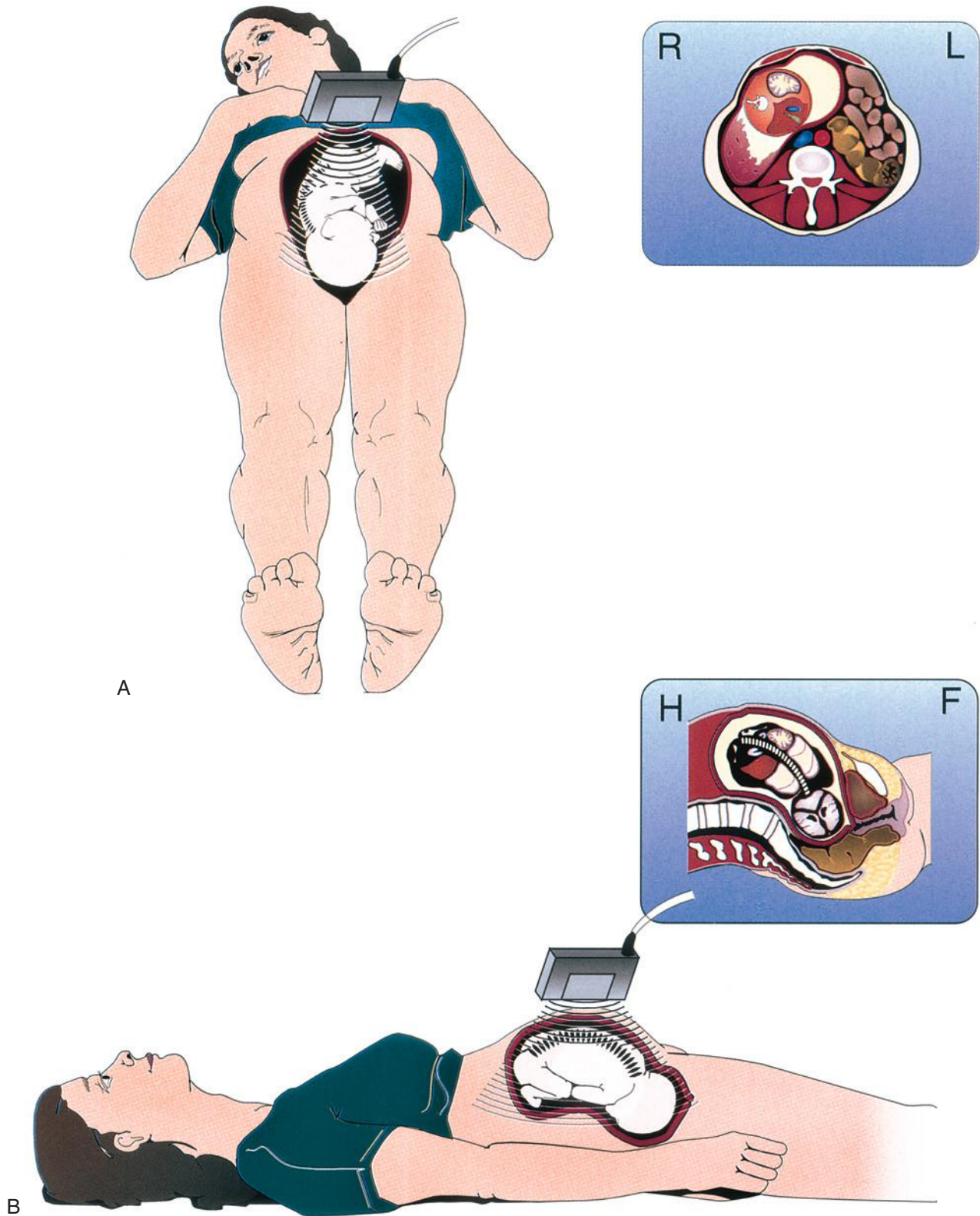


FIG 1-2 A, Illustration of a transverse plane of section of the gravid uterus. The fetus is in cephalic presentation, so this scan transects the fetal abdomen transversely. **B**, Longitudinal plane of section of the same fetus. These images are viewed with the maternal head to the left of the recorded image. F, foot; H, head; L, left; R, right.

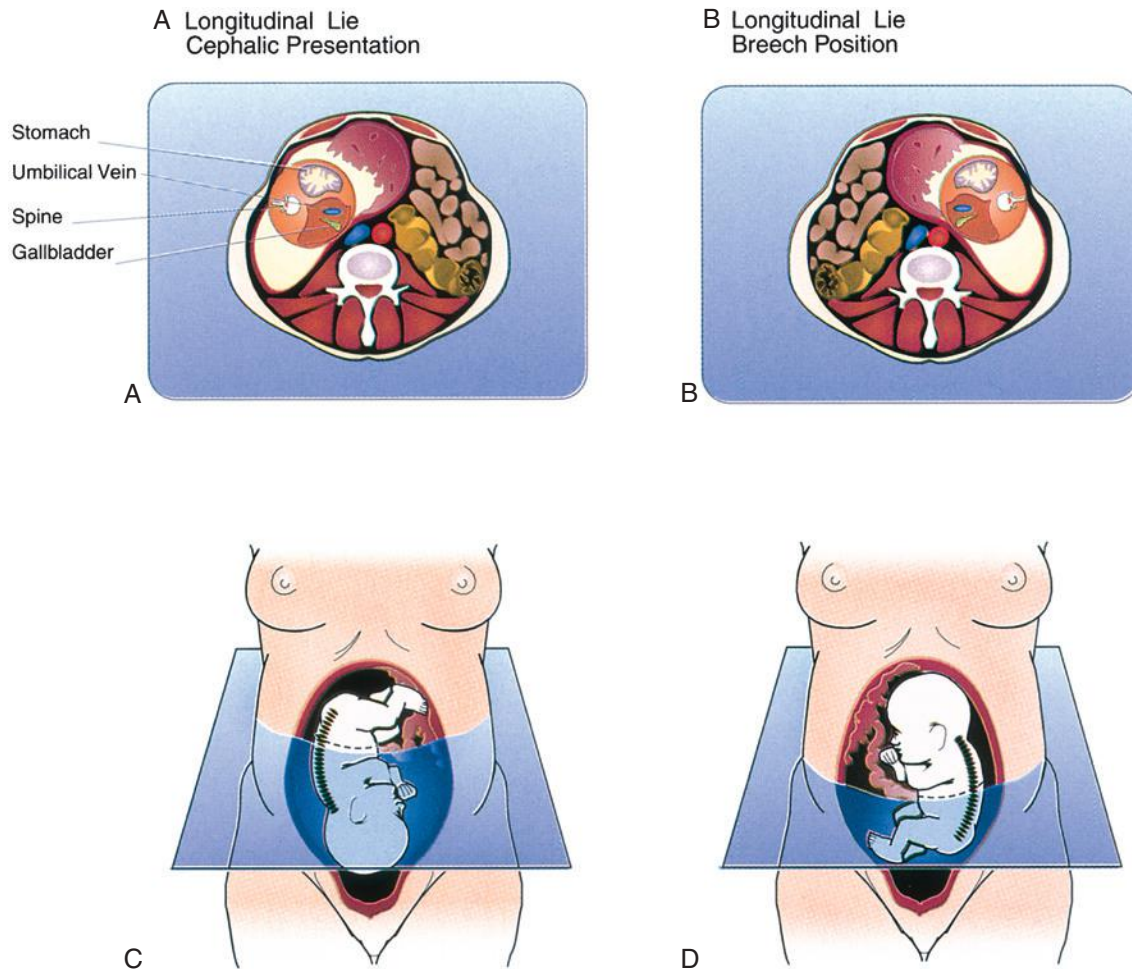


FIG 1-3 Knowledge of the plane of section across the maternal abdomen (longitudinal or transverse) as well as the position of the fetal spine and left-sided (stomach) and right-sided (gallbladder) structures can be used to determine the fetal lie and presenting part. **A**, This transverse scan of the gravid uterus demonstrates the fetal spine on the maternal right with the fetus lying with its right side down (stomach anterior, gallbladder posterior). Because these images are viewed looking up from the patient's feet, the fetus must be in a longitudinal lie and cephalic presentation. **B**, When the gravid uterus is scanned transversely and the fetal spine is on the maternal left, with the right side down, the fetus is in a longitudinal lie and breech presentation. **C**, When a longitudinal plane of section demonstrates the fetal body to be transected transversely and the fetal spine is nearest the lower uterine segment with the fetal right side down, the fetus is in a transverse lie with the fetal head on the maternal left. **D**, When a longitudinal plane of section demonstrates the fetal body to be transected transversely and the fetal spine is nearest the uterine fundus with the fetal right side down, the fetus is in a transverse lie with the fetal head on the maternal right. Although real-time scanning of the gravid uterus quickly allows the observer to determine fetal lie and presentation, this maneuver of identifying specific right- and left-sided structures within the fetal body forces one to determine fetal position accurately and identify normal and pathologic fetal anatomy.

Amniotic Fluid Volume

Amniotic fluid is an important consideration in assessing fetal development and well-being. Although there is relatively good agreement on the significance of extremes of amniotic fluid volume, there remains some controversy over the methodology used to make the diagnosis of either too much or too little amniotic fluid. Amniotic fluid volume should be assessed subjectively at all ultrasound examinations. Although a diagnosis of oligohydramnios and polyhydramnios can be made subjectively, the extremes of amniotic fluid volume should also be assessed objectively using either the DVP or AFI. The ability to assess amniotic fluid volume subjectively at different stages of gestation is readily learned and should not be difficult for most examiners. Two

points should be remembered when assessing amniotic fluid volume. First, amniotic fluid volume is large compared with fetal volume at early stages of gestation and should not be misinterpreted as polyhydramnios. Conversely, in term patients, the normal volume of amniotic fluid is quite small so that only small pockets may be seen. Second, patients who are obese often appear to have less than normal volumes of amniotic fluid. This may be due in part to scattering of sound with resultant artifactual echoes within the amniotic fluid.

In making the diagnosis of oligohydramnios, one should remember two points. First, because in many cases, it will imply the likelihood of a fetal renal malformation or severe growth restriction in the absence of ruptured membranes, this diagnosis should be made only when there is almost no amniotic fluid. An exception to this is when there is

Breech Presentation



FIG 1-4 Illustration of the types of breech presentation. In a frank breech presentation (the most common), the thighs are flexed at the hips with the legs and knees extended. In complete breech (the least common), the thighs are flexed at the hips, and there is flexion of the knees as well. One or both hips and knees are extended in the footling breech. The risk of cord prolapse is greatest with a footling breech and least with a frank breech. (Illustration copyright © 2006 Nucleus Medical Art, www.nucleusinc.com. All rights reserved.)



FIG 1-5 Longitudinal scan of a footling breech presentation. In this scan, the leg (*arrow*) and foot extend into the lower uterine segment and cervix.

a small amount of fluid in an early or mid-second trimester examination (when normally large amounts of amniotic fluid would be anticipated). Second, because of the association of severe diminution of amniotic fluid in a compromised fetus with ultimate fetal demise, the obstetrician should be alerted immediately if this diagnosis is made, before the patient leaves the ultrasound evaluation area.

The diagnosis of polyhydramnios, although seeming to be less serious, in many cases may in fact be associated with significant complications to the mother and fetus. In the mother, preterm labor and ruptured membranes may occur as a result of polyhydramnios, and in the fetus, fetal anomalies may be present. Although many cases of polyhydramnios ultimately result in a normal fetus, the high number of anomalous fetuses with this condition reported in the literature should alert the sonographer to perform a thorough evaluation when this diagnosis is suggested.⁵⁶⁻⁵⁸

Amniotic Fluid Volume in Multiple Gestations

If one looks at a list of causes of polyhydramnios in many obstetric texts, multiple gestations will most likely appear. Although increased

amniotic fluid volume may appear in twin gestations, in most cases the cause is some abnormality of pregnancy.⁵⁹ Many of these cases are due to twin-twin transfusion syndrome.⁶⁰

Placenta

As mentioned earlier, whenever the placenta is identified in pregnancy, its position and relationship to the cervix should be noted in the interpretation. The literature has emphasized the large number of false positive diagnoses of placenta previa that are made either early in pregnancy or in the presence of an overdistended urinary bladder.^{61,62} Although this is true, one must not be lulled into a sense of security in thinking that all low-lying placentas will “go away” and be clinically unimportant. If the placenta is low lying during a second trimester examination, every effort should be made to answer the question as to whether there is or is not a placenta previa; this may require a transvaginal examination. However, if after a variety of maneuvers and transducers (Trendelenburg position, emptying the bladder, translabial or transvaginal scanning) one is still unsure about the relationship of the edge of the placenta to the cervical os, the placenta should be interpreted as low lying, and a placenta previa cannot be excluded. Therefore, these patients should have follow-up examination at 32 weeks of gestation. We prefer to report the distance from the inferior edge of the placenta to the internal cervical os rather than relying on terms that may have differing meanings (e.g., marginal placenta). In patients with a prior cesarean delivery and a placenta previa, the placenta should further be assessed for evidence of placenta accreta, as described in [Chapter 19](#).

Abruptio placentae is a diagnosis that is often difficult to make using ultrasonography. One should remember that the myometrium and its vessels, as well as a transient myometrial contraction, may simulate a hematoma and that these potential false positive diagnoses should be avoided. Because most clinicians are aware that abruptio placentae is a difficult diagnosis, they often refer patients for ultrasound evaluation to exclude a placenta previa rather than to specifically view the abruptio. Patients with a true placental abruptio may not ever be seen in the ultrasound laboratory and go straight to labor and delivery.

Vasa previa, which is a variation of umbilical cord anatomy rather than a placental abnormality, is a serious and often overlooked condition. It occurs when fetal vessels cross the internal cervical os in an

attempt to reach the main substance of the placenta. Vasa previa can result in fetal exsanguination during delivery and should be suspected in cases of a velamentous cord insertion or a succenturiate lobe of the placenta, as well as in cases of “resolved” placenta previa.

Fetal Malformations

The subject of fetal malformations is among the most emotionally charged issues that either the parents or a diagnostician may have to face. Over time, ultrasound evaluation has undergone a transformation that has allowed us to answer not only the basic question as to whether the patient is pregnant but also whether a fetal anomaly is present. As smaller and smaller abnormalities are identified, the question now becomes what degree of assurance should a patient expect from a report that no anomaly was seen during a routine ultrasound examination. This is a complex issue. To examine every patient for all anomalies would be highly impractical. Fortunately, most major anomalies can be detected as part of a routine evaluation.

Major genetic or structural birth defects affect approximately 3% of infants born in the United States.⁶³ Congenital malformations are the single leading cause of infant death in the United States, accounting for more than 21% of all infant deaths.⁶⁴ In the United States, it has been estimated that 100,000 to 150,000 children are born each year with major congenital malformations, and approximately 8000 of these babies die before completing their first year of life.⁶⁴ Children with congenital malformations account for approximately 30% of pediatric admissions, and the total cost of health care is estimated at more than \$1.4 billion annually.⁶⁴⁻⁶⁶

As was mentioned in the discussion of the first trimester ultrasound examination, fetal anomalies have been described in virtually every organ system at almost every gestational age. There is much controversy as to when a comprehensive scan of a pregnancy should occur. There has been a desire from many to accomplish this task at a time just before an amniocentesis (14 to 16 weeks) or even earlier, at the time of nuchal translucency measurement.⁶⁷ In particular, three advantages to earlier anatomic evaluation have been cited: (1) transient abnormalities such as an increased nuchal translucency and echogenic bowel (which may serve as markers for chromosomal and structural abnormalities) may disappear if scanning first occurs after 16 weeks⁶⁸; (2) structures such as the fetal hands may be more readily seen, particularly with fingers extended, earlier than later in pregnancy⁶⁸; and (3) if necessary, termination may be easier to accomplish and safer earlier than later in gestation.⁶⁸ Although it is true that many morphologic abnormalities will be detected particularly when using a transvaginal probe, certain abnormalities of the face, heart, and skeleton will not be detected at early gestational ages. Likewise, certain embryologic developmental stages, such as the development of the cerebellar vermis and corpus callosum, are not complete until the mid to late second trimester. If one has the economic luxury of performing several sonographic examinations during pregnancy, a scan at 11 to 14 weeks' gestation followed by a scan at 22 to 24 weeks' gestation might be ideal. It is certainly not unreasonable to exclude gross and potentially lethal abnormalities during the time of the nuchal translucency scan. This would obviously affect the timing of the next scan. However, it is our recommendation, and that of professional organizations (Reddy and coworkers²⁸), that if a single ultrasound or a targeted (Level 2) examination is performed, it should be done at a gestational age of 18 to 20 weeks. The reason for this is that the fetus will be of a sufficient size to exclude most abnormalities and still allow time for a follow-up examination, if necessary. The slight loss of accuracy in assigning gestational age at this time is typically of limited clinical significance and is well worth the gain in visibility of fetal anatomy and pathologic malformation.

The patient and the referring obstetrician should be made aware that during the standard ultrasound examination, although many abnormalities may be detected fortuitously, more subtle lesions are likely to be detected only when the fetus is known to be at risk for a specific malformation. Anatomic malformations are likely to grow during pregnancy just as the fetus does; a defect seen at birth may have been too small to be detected earlier in pregnancy. Some lesions, such as duodenal atresia and achondroplasia, may not manifest until late in the second trimester. Finally, it is important for sonologists to know the limits of their expertise. If a malformation is suspected and the examiner has had little experience with the abnormality in question, the case should be referred to a more experienced examiner. Only in this way will patients be served best.

Uterus and Adnexa

Evaluation of the uterus and adnexa becomes more difficult the later in gestation the examination occurs. The most common abnormalities that are likely to be detected are uterine myomas. As stated earlier, it is important to measure the size of the myoma, record the location of the myoma, and define the relationship of the myoma to the cervix. If ovarian abnormalities are suspected, patients should have a postpartum examination.

VERBIAGE USED IN THE AIUM/ACR/ACOG GUIDELINES

The committees and individual members of the AIUM, ACR, and ACOG who helped develop the guidelines described earlier did a remarkable job. It is not easy producing a document such as this that will be widely applicable to ultrasound practitioners. In a few areas in which the words chosen were not clear we would like to give our own suggestions for interpretation.

It is understandable that the guidelines attempted to give the practitioner latitude in requirements for the obstetric ultrasound examination. They attempted to take into account the differences in maternal and fetal anatomy from one patient to the next as well as technical limitations at times. There are a number of instances in which the guidelines state that a structure or structures should be imaged. Unfortunately, additional wording is added that states “when possible” or “can also be attempted” or “when technically feasible.” As stated in the editorial mentioned earlier, “One could reasonably state that any of the views defined in the guidelines can only be obtained ‘if technically feasible.’” Indeed, the introduction to “Fetal Anatomic Survey” states that one may anticipate technical limitations: “some structures can be difficult to visualize due to fetal size, position, movement, abdominal scars, and increased maternal wall thickness.”²⁸ Adding these additional words, mentioned earlier, gives the examiner a “way out” to not examine important fetal or maternal anatomy. This issue occurs in discussion of the first trimester, when these modifying statements are made with respect to the cervix and amnionity and chorionicity in multiple gestations. We do not know of a reason why the cervix cannot be assessed in the first trimester in any patient using a variety of methods available. Chorionicity and amnionity can best be assessed in the first trimester of pregnancy; although the differentiation of monoamniotic from diamniotic twins may be difficult later in gestation, there is no reason chorionicity cannot be determined at this time. In the extremities section, the legs and arms are mentioned. Medically, this would mean only the tibia/fibula and the humerus. We believe the intent was for the femur, humerus, tibia, fibula, radius, and ulna to be evaluated.

The fetal sex was stated as, “Medically indicated in low-risk pregnancies only for evaluation of multiple gestations.” Although we

appreciate the intent, we suggest that it should be noted that fetal sex should also be demonstrated in singleton gestations when medically indicated for diagnosis and counseling. Examples of this type of indication would include hemophilia or distal urinary obstruction, attempting to differentiate posterior urethra valves (predominantly male disorder) from a cloacal abnormality (predominantly female disorder). As for multiple gestations, there are many who would take issue with saying that any multiple pregnancy is low risk. It perhaps would have been better stated that “when chorionicity is difficult to determine in multiple gestation pregnancies, fetal sex determination, when different between the twins, will be helpful in excluding monozygosity.”

For weight determination, a statement is made that “Currently, even the best fetal weight prediction methods can yield errors as high as $\pm 15\%$.” Although we agree with this in concept, we would not have put what appears to be an upper limit on errors in weight estimation. This seems to imply that errors greater than this would be below the standard of care. Particularly in macrosomic fetuses, errors as high as 25% to 30% can be seen from what seemed to be reasonable biometry.

A statement regarding technical difficulties reads: “A second- or third-trimester scan may pose technical limitations for an anatomic evaluation due to imaging artifacts from acoustic shadowing. When this occurs, the report of the sonographic examination should document the nature of this technical limitation. A follow-up examination may be helpful.” Although we fully understand this limitation, which all of us have encountered, the recommendation is problematic. There are certain anatomic structures or situations in which the anatomy has to be imaged or an abnormality reported. The statement “follow-up examination may be helpful” is too vague. In far too many cases, failure to visualize normal anatomy was judged to be due to fetal position, when in fact the structure was abnormal. If the brain, heart, or kidneys and bladder (in the presence of oligohydramnios) are not seen, a short-interval follow-up examination should be performed. If failure to adequately visualize the anatomy is a result of maternal body habitus, follow-up in 2 to 4 weeks may be useful. If the structure is still not seen, the referring obstetrician should be notified and the conversation documented. Further follow-up should be recommended as clinically indicated.

INTERPRETATION OF THE ULTRASOUND EXAMINATION

This is, of course, the focus of the remainder of this book; however, we would like to make several comments. Although the subspecialty of obstetric ultrasound seems to attract new biometric applications daily, we have never been much of advocates of the sole use of measurements to achieve a diagnosis. It seems that every day, someone has developed and published a new chart for the measurement of a fetal anatomic structure. We fully recognize that there are many measurements that are necessary for accurate ultrasound interpretation, fetal biometry for size and weight, cervical length, and fetal ventricular size, to name a few. For many of the abnormalities that can be recognized sonographically, we would prefer that the sonologist give more credence to his or her subjective eye than to a measurement when images or individual structures “just don’t seem right.” Although a measurement may appear to be within normal limits, with experience, there are times when subjectivity, in our opinion, wins out. It is acceptable to say that despite an AFI of 6, there is oligohydramnios or that the fetal bowel appears dilated without an objective measurement of the bowel lumen.

Another pitfall is making a measurement of an anatomic structure and not interpreting the significance of the measurement. For example,

some ultrasound laboratories believe that adding numerous measurements, for example, transcerebellar, renal, or intraorbital measurements, in addition to standard biometry (biparietal diameter [BPD], head circumference [HC], abdominal circumference [AC], femur length [FL]), will make the examination more complete. Although we will not argue the necessity of doing these additional measurements in some select cases, what is puzzling is that often the interpreter of the examination will not check these measurements against standard tables or nomograms to determine if they are normal or abnormal. In our opinion, this is a serious mistake.

Without launching into a discussion of statistics, suffice it to say that no measurement will likely be 100% accurate without false positive or false negative results. There are some situations in which false positive results may be acceptable (e.g., screening examinations for a serious abnormality). Every practitioner abhors the notion of being labeled an “over-reader” of examinations. The sonologist needs to consider whether the goal is to not miss any patients with a condition (thus, resulting in more potential false positive results), which may mean additional testing or intervention, or whether they wish fewer false positive results and thus risk missing the detection of an abnormality in some patients. With this in mind, one can set the threshold level for the test, either lower or higher. If the sonologist is calling a referring clinician or scheduling follow-up examinations four to six times a week with a suspected abnormality (pelviectasis, echogenic bowel, and so on), one should perhaps reevaluate the criteria for defining something as abnormal. However, if one calls a referring physician once every 2 to 4 weeks when one is “bothered” by a finding and wishes to call attention to it, one should not feel as if he or she is “over-calling” an abnormality. Likewise, one should not be embarrassed about calling the referring physician about a concern that something might be abnormal even if it ultimately proves to be normal. Only in this way will patients be best served.

Reporting of Ultrasound Results

One would anticipate that this might be the least contentious topic when discussing the obstetric ultrasound examination. However, it is controversial. In concert with recommendations by various organizations (e.g., AIUM, ACR, ACOG) a written report should be produced at the completion of the ultrasound examination and should be placed in the patient’s medical records. With widespread implementation of electronic medical records, this report typically uses one of a number of available reporting packages. Such reporting packages have been developed to make reporting easier: they include but are not limited to obstetric worksheets or checklists, computer templates or canned reports, computer voice recognition, digital transcription systems accessed by telephone or computer, and traditional voice dictation that is typed by transcriptionists (on site or remotely). All of these methods have the potential for producing an accurate and readable report. However, in our experience, the easier the reporting method, the more likely that observations made during the examination will not be conveyed accurately. Too often, sonographers and sonologists finish an examination and quickly enter checks into boxes on a worksheet indicating that a particular structure was observed or was normal. It is inconceivable, watching the speed with which the worksheet is completed, that they ever asked themselves the question, Did I really see those structures? Although it is becoming antiquated, conventional dictation systems that require the examiner to pick up the recording device and say “the following structures were seen ...” or some similar device or mechanism probably had a better chance of succeeding in conveying accurate information.

There are mixed benefits to templates that contain standard paragraphs such as the following structures were seen: lateral ventricles,

cerebellum, and so on. Although this information may be necessary for reimbursement purposes, it is likely that sonologists or referring physicians rarely read these paragraphs. They also have the potential to be confusing when abnormalities are detected, particularly if the paragraph ends with “and these structures were normal.” If one does not alter the paragraph, the referring physician will read in this standard paragraph a sentence that says: “the fetal kidneys were normal” and in the next paragraph a sentence that reads “bilateral fetal hydronephrosis was seen.”

It is our practice and recommendation that when fetal abnormalities are detected, the referring physician should be contacted in person or by telephone and the discussion should be documented in the report.

A question often raised concerns what to do when a structure normally seen on routine (basic) sonograms (and listed in the guidelines) is not seen. Or what to do when one identifies a structure that has an unusual appearance but that one suspects is probably normal. Often, it is assumed that failure to see a structure or structures are secondary to technical limitations, such as shadowing or poor fetal position or fetal physiology (the fetus just urinated or fluid in the stomach passed into the duodenum). If the examination is performed in the early second trimester, the patient should return within 2 to 4 weeks for another evaluation. As was mentioned earlier, if a structure in question relates to the heart or brain or kidneys and bladder (when oligohydramnios is present), the study should be considered incomplete. If a follow-up examination is warranted, the referring physician should be contacted and the conversation documented. The rationale for these recommendations is the concern that adequate follow-up and evaluation will not occur, or if they do occur, they will not be performed in a timely fashion. Statements indicating “clinical correlation recommended” are appropriate when the abnormality seen needs to be correlated with patient’s medical condition, laboratory tests, family history, and the patient’s age, but not when an isolated abnormality is seen (dilated bowel loop).

Discussing the Examination With the Patient

This topic is also controversial. The patient obviously wants to know that the fetus is healthy and to know the results at the time of the examination. As a general rule, the referring obstetrician knows the patient best and also often has important information about the patient’s menstrual history, family history, laboratory values, and emotional state. However, the performing sonologist may be more knowledgeable about the significance of a given finding and may be better able to explain the results to the patient and to counsel her effectively. In cases of suspected morphologic or genetic abnormalities, the advice of a reproductive geneticist may prove invaluable. As is discussed later, often the first words that are said to the patient are the things that she remembers. Despite what is said later, it may be difficult to undo what was said initially. It is appropriate to say that the diagnosing physician needs to evaluate all of the images together and possibly compare them with previous studies. At that time, the final report will be generated.

Counseling is often straightforward if the case is normal or when the diagnosis is unequivocally lethal (anencephaly). It becomes more complex in cases in which the outcome is less than 100% certain (e.g., mild isolated ventriculomegaly). It is important not to insert our own bias and values about raising children with disabilities. One cannot assume that if a serious malformation is detected that a patient will desire an abortion rather than to deliver a baby with disabilities. In some cases, the person informing the patient of the results is uncertain of the significance of the findings,⁶⁹⁻⁷¹ and the first information the patient hears may be unclear or misleading. This confusion may color

how patients perceive later information and may result in dissatisfaction with the bearer of the news.^{69,71}

It is often assumed that women weigh equally the risk of delivering a baby with a congenital abnormality versus a procedure-related pregnancy loss (amniocentesis). In fact, studies have demonstrated that most women see the long-term consequences of raising a disabled child as worse than a miscarriage, although women vary widely in this regard.⁷⁰ It is important to remember that we do not know better than our patients what is best for them. Our challenge is to help them reach a decision that is best for them given their particular background, experience, and values.

Even when precise and correct information is transmitted in a counseling session, it may not be interpreted with the same intended meaning by the patient. Certain words tend to have more serious and worse connotations to the patient than alternative words.⁷¹ The words “rare abnormality” are often interpreted as serious (even if it is a mild abnormality). The word “abnormality” is often interpreted as worse than a variation of normal. Likewise, technical genetic words often have a worse connotation. “Trisomy” sounds worse to most patients than “an extra chromosome.”⁷¹ Choroid plexus cysts (CPCs) and echogenic intracardiac foci (EIF) are discussed in [Chapter 3](#). One should be aware that even if the sonologist firmly believes that the finding of an isolated CPC or an EIF is likely a normal variant and of no consequence, for a patient, the fact that these structures are in the brain and heart is no small matter. The fact that there may not be a significant increased risk in that individual patient based on other findings or that the abnormality is of small size may not alleviate the patient’s anxiety once she is told of these findings.

There has been much debate regarding whether physicians should disclose that an isolated sonographic “soft marker” for a chromosomal abnormality has been detected in a fetus in the absence of other risk factors for aneuploidy.⁷²⁻⁷⁴ A 2007 study by Lee and associates⁷⁵ found that the detection and communication of isolated aneuploidy markers (CPCs, EIF, renal pyelectasis, echogenic bowel) is associated with increased maternal anxiety and perhaps unnecessary amniocenteses. Their conclusion: “given the amount of maternal anxiety generated with detection of aneuploidy markers, serious consideration should be given to offering pre- and post-ultrasound genetic counseling or otherwise, nothing should be mentioned about ultrasound markers that may be normal variants in patients who have no other risk factors for aneuploidy.”⁷⁵

The likelihood of having a normal child when an abnormality is suspected or a patient found to be at increased risk based on screening is almost always received more favorably by the family than being told of the small percentage of having an abnormal child. That is, a 99% chance of normal sounds less worrying than 1% chance of abnormal. When risk is given as a proportion, it often sounds worse than when it is given as a rate; in other words, 1/X sounds worse than X%.⁷¹ One should be aware that a large portion of the public lacks functional knowledge of fractions, large numbers, or percentages.^{76,77} In one study,⁷⁸ a third of adult women with less than a college education did not recognize that 1/1000 is less than 1%. It is interesting that although in most medical specialties, risk or prognosis is given as a percentage, for example, 10% chance of a cure or bad outcome, in prenatal genetic counseling, risk is often given as a proportion, which most patients do not fully understand. In a study by Grimes and Snively,⁷⁶ women of varying ages, education levels, and languages were asked to identify which proportion of bladder infection was higher: 1 in 384 or 1 in 112 persons. The same women were then asked to identify which rate of infection was higher: 2.6 per 1000 women or 8.9 per 1000 women. Overall, 73% correctly identified the higher risk in rate format (X/1000), in contrast with 56% who correctly answered the same

question framed as proportions (1/X).⁷⁶ Clearly, women understood rates better than proportions.

Perhaps the most important point of this discussion is what to do when a slight variation of normal or an unusual finding is seen. The first answer would be to consult your colleagues. If that does not answer the question, then one should discuss the case further with more experienced experts or refer the patient to a university or specialty center. In some cases, one will simply need to report that there is a finding and that you are uncertain of the significance. When the physician feels pressure to always give a black and white, or normal versus abnormal, answer without honest indecisiveness, this approach does a disservice to the patient.

Evaluating the Obstetric-Gynecologic Ultrasound Literature

Whereas this text serves as a reference to many, as an aid to ongoing clinical problems, new and useful clinical information is reported constantly in the medical literature. It is appropriate that the reader keep current with new advances. It is also important to be vigilant for poorly constructed studies and conclusions. Although virtually every report will have some mention of the sensitivity and specificity of a new test, this is only a small part of an adequate analysis of the utility of a new technique. There are a number of areas that should be considered when evaluating a new report in the literature; these areas are outlined in Table 1-3.⁷⁹ Perhaps the most common error is that authors do not state the prevalence of “disease or abnormality” in their population or that readers do not take this into account when applying the report to

their own practice. This also holds true for both equipment and techniques used for analysis.

When the authors discuss a new technique or potentially helpful finding, it is invariably depicted in their first figure (i.e., Fig. 1-1). If after looking at Figure 1-1 in the new publication, as well as its legend and text description, the reader does not understand what is being demonstrated, the publication is likely to be of little value in clinical practice.

Malpractice and the Obstetric Ultrasound Examination

It is likely that each person reading this text has been touched in some way by the ongoing malpractice crisis. For most of us, this crisis has resulted in increased costs of goods and services, and for some, it has meant being the defendant in a malpractice suit.

Medical malpractice actions typically arise from a patient's allegation of negligent diagnosis or treatment. In order to prevail, the patient must show that the physician fell below the applicable standard of care. Standard of care is established most commonly by the testimony of an expert witness. Although guidelines promulgated by various organizations (AIUM, ACR, ACOG) alone do not establish the standard of care introduced at trial, they do describe the general practice of obstetric ultrasound in many communities and are often referred to by medical experts.⁸⁰ The definition of standard of care varies slightly from state to state. California's instruction to juries regarding standard of care in medical malpractice cases is listed here:

CALIFORNIA CIVIL JURY INSTRUCTIONS (CACI)

501. Standard of Care for Health Care Professionals

[A/An] *[insert type of medical practitioner]* is negligent if [he/she] fails to use the level of skill, knowledge, and care in diagnosis and treatment that other reasonably careful *[insert type of medical practitioners]* would use in the same or similar circumstances. This level of skill, knowledge, and care is sometimes referred to as “the standard of care.”

[You must determine the level of skill, knowledge, and care that other reasonably careful *[insert type of medical practitioners]* would use in the same or similar circumstances, based only on the testimony of the expert witnesses *[including [name of defendant]]* who have testified in this case.]

Data on the number of claims of malpractice and their settlements are difficult to obtain. In one report, Sanders⁷⁸ reported malpractice claims in diagnostic ultrasonography in 228 cases. Obstetric ultrasound examinations represented the majority (78%) of the cases.

Some of the more common reasons for the initiation of malpractice suits (whether legitimate or not) include the following:

- Unreasonable expectations of the ultrasound examination on the part of the patient and the referring physician
- Physician performing the examination having inadequate training or equipment
- Failure to seek consultation in difficult cases
- Inadequate or incomplete study
- Misinterpretation of the ultrasound examination (resulting in the inability to terminate before the legal state limit, a wrongful termination, or preterm or postterm delivery)
- Poor communication with referring clinicians (improper wording, lack of timely communication)
- Failure to maintain ultrasound equipment
- Failure to supervise personnel adequately

It is our desire that this text, through the education process of the sonographer and sonologist, will help alleviate errors in diagnosis

TABLE 1-3 Evaluating the Literature

Abstract

What are the objectives, findings, and conclusions of the study?

Introduction

What is the purpose of the diagnostic test?

Materials and Methods

How are the patients selected?

Are they representative of those who are ordinarily tested?

How is the test(s) performed and interpreted?

Are the interpretation criteria well defined and reproducible?

What is the gold standard for diagnosis? Is it appropriate?

Are the sonologists blinded from the final diagnosis and is the final diagnostician (pathologist) blinded from sonographic interpretation?

Is the gold standard applied uniformly?

In a comparison study, are the tests evaluated fairly?

Results

How is the accuracy reported?

Are the spectrum of disease and important covariates, such as comorbidity, age, sex, and body habitus, accounted for in tabular presentation of data?

Is the statistical analysis clearly described and appropriate?

Discussion

Are the deficiencies in the methodology of accuracy assessment acknowledged and discussed?

Are other relevant factors, such as disease prevalence, therapeutic effectiveness, and cost, adequately accounted for in the clinical recommendations?

From Black WC: How to evaluate the radiology literature. *AJR Am J Roentgenol* 154:17, 1990.

and thus reduce the number of these cases. Unfortunately, despite the best medical care, some malpractice suits are brought against physicians.

CONCLUSION

The appeal of the ultrasound examination is that it is a noninvasive, safe procedure that has a high degree of patient acceptance and can yield a wealth of information. It is always a delight to examine the obstetric patient and reassure her about her pregnancy, when appropriate.

When a pathologic process is first identified, the role of the sonologist is that of a detective who attempts to piece together all of the

information to arrive at the correct diagnosis (Fig. 1-6). Although discovering a pathologic process is always disconcerting, the sonologist can be a counselor to the patient and the clinician and can help guide them to appropriate management decisions. However, there are times when an abnormality is strongly suspected but it may be equivocal or may not fit into a specific category. Under these circumstances, the best pathway for the sonologist to follow may be to do a follow-up examination and seek consultation. If time does not allow a follow-up examination, then the sonologist should communicate to the referring physician and the patient that a definitive answer is not possible and that decisions will have to be made with less-than-perfect information.

We are hopeful that this text will serve two purposes: to educate and to excite. If those reading this text maintain the same enthusiasm

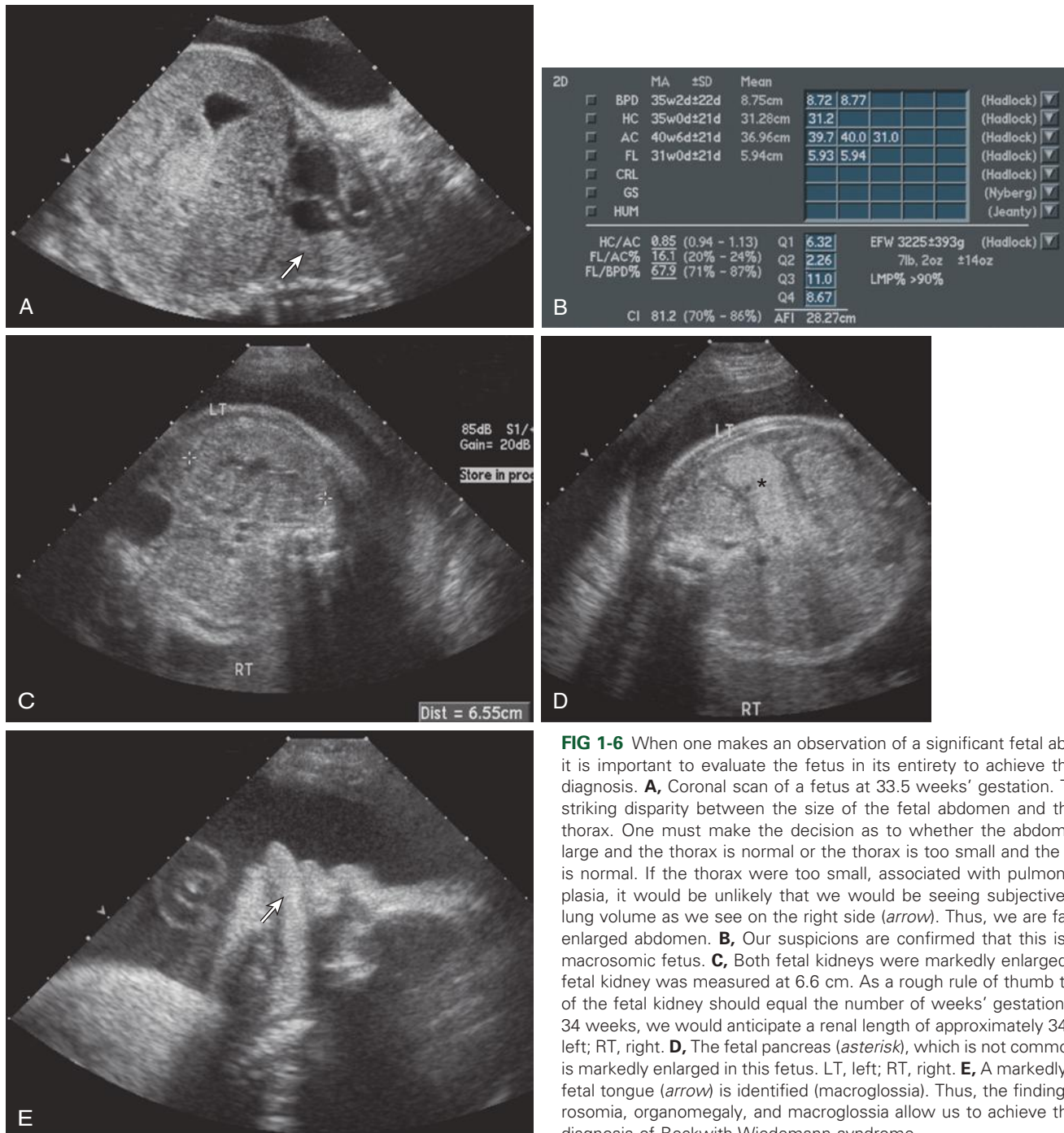


FIG 1-6 When one makes an observation of a significant fetal abnormality it is important to evaluate the fetus in its entirety to achieve the correct diagnosis. **A**, Coronal scan of a fetus at 33.5 weeks' gestation. There is a striking disparity between the size of the fetal abdomen and that of the thorax. One must make the decision as to whether the abdomen is too large and the thorax is normal or the thorax is too small and the abdomen is normal. If the thorax were too small, associated with pulmonary hypoplasia, it would be unlikely that we would be seeing subjectively normal lung volume as we see on the right side (arrow). Thus, we are favoring an enlarged abdomen. **B**, Our suspicions are confirmed that this is in fact a macrosomic fetus. **C**, Both fetal kidneys were markedly enlarged. The left fetal kidney was measured at 6.6 cm. As a rough rule of thumb the length of the fetal kidney should equal the number of weeks' gestation. Thus, at 34 weeks, we would anticipate a renal length of approximately 34 mm. LT, left; RT, right. **D**, The fetal pancreas (asterisk), which is not commonly seen, is markedly enlarged in this fetus. LT, left; RT, right. **E**, A markedly enlarged fetal tongue (arrow) is identified (macroglossia). Thus, the findings of macrosomia, organomegaly, and macroglossia allow us to achieve the correct diagnosis of Beckwith-Wiedemann syndrome.

for obstetric and gynecologic sonography that we have, we will have fulfilled our goal.

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Genetics and Prenatal Genetic Testing

Mary E. Norton, Britton D. Rink

SUMMARY OF KEY POINTS

- In the population, 2% to 3% of newborns have a congenital malformation or genetic disease identified at birth.
- Despite advances in genetics, the cause of more than half of human congenital abnormalities remains unknown.
- Chromosomal abnormalities are present in about 0.9% of newborns and include abnormalities of chromosome number as well as abnormalities of chromosome structure.
- The embryo is most sensitive to teratogenic effects between 3 and 8 weeks of development.
- Current available tools for fetal aneuploidy screening include cell-free DNA screening, various forms of multiple marker screening with first and second trimester maternal serum analytes, and ultrasound measurements, including nuchal translucency.
- The prevalence of many single gene disorders varies with race and ethnicity, and testing is often recommended based on an individual patient's background.
- Expanded carrier screening, including panels to simultaneously test for a large number of genetic conditions, is increasingly utilized for prenatal genetic screening.
- Chorionic villus sampling (CVS) and amniocentesis are both routinely used for prenatal diagnostic testing and can provide tissue for such tests as fluorescence in situ hybridization (FISH), karyotyping, chromosomal microarray analysis (CMA), and DNA-based tests.
- The loss rate attributable to CVS and amniocentesis is estimated to be between 1/500 and 1/1000 and decreases with provider experience.
- Many structural fetal abnormalities are associated with an increased risk of aneuploidy as well as copy number variants detectable with chromosomal microarray.

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Genetic diseases are often perceived to be so rare that the average practitioner will seldom encounter them. However, increasing knowledge and technologic advances in genetic testing have demonstrated that this is far from the case. The availability of prenatal diagnosis for a wide range of disorders continues to increase with advances in genetics. In addition, progress has been made in population screening tests to identify couples who carry a genetic disorder. New techniques, such as cell-free DNA screening, have also changed the field of prenatal diagnosis significantly. These improvements in prenatal screening and diagnosis mean that many more at-risk couples are able to have unaffected children. In addition to reproductive choice, carrier screening and fetal diagnostic testing afford the important opportunity for preparation of the family and the delivery site for the birth of a fetus with a known genetic disorder.

Ultrasound plays a central role in the provision of prenatal screening and diagnosis. Not only is ultrasound key to guiding prenatal diagnostic procedures, but also integration of a genetics-based prenatal diagnosis program has been shown to increase the accuracy of diagnosis when compared to ultrasound alone.¹ This chapter includes a discussion of genetics, with an emphasis on recent advances relevant to prenatal diagnosis and a description of current strategies for genetic testing with a focus on how genetic screening and sonography together contribute to the provision of accurate and precise prenatal diagnosis.

Genetics and Birth Defects

According to most studies, 2% to 3% of living newborns have a congenital malformation.² When considering birth defects noted in the first years of life, this incidence is nearly doubled. With the decline in infant mortality in the United States from infection and malnutrition, congenital malformations are now a leading cause of infant death (>20%) and are responsible for greater than 30% of intensive care nursery admissions.³ Congenital defects range from enzyme deficiencies caused by single gene mutations to complex associations of structural defects. The continuum between purely biochemical abnormalities and structural birth defects includes disorders of structure, function, metabolism, and behavior.

Birth defects result from the interaction between the genetic makeup of the embryo and the environment in which it develops. The basic developmental information is encoded in genes, but the genotype is subjected to environmental influences that can impact the observed phenotype. In some cases, the genetic information is expressed regardless of environment, whereas in others, environmental causes interfere with normal development despite a normal genotype. Although some processes are primarily environmental and others mainly genetic, the distinctions between the two are not perfect.

Despite considerable advances and research over past several decades, the cause of more than half of human congenital abnormalities remains unknown. Of those with a recognized cause, approximately 15% to 20% are autosomal genetic diseases and 20% are cytogenetic in origin. Fewer than 1% of anomalies are thought to result from teratogenic medications. Some of the remaining defects are associated with other environmental exposures during pregnancy,

including infectious agents (3%), maternal disease states (4%), mechanical problems (1% to 2%), irradiation, and unknown environmental causes. The remainder are of unknown or complex causes (multifactorial, polygenic, spontaneous errors of development, and synergistic interactions of teratogens) (Fig. 2-1).^{3,4}

Developmental Disorders: Causes, Mechanisms, and Patterns

Errors in morphogenesis are often classified by dysmorphologists according to the underlying pathogenesis (Fig. 2-2). *Malformations* are defects in the structure of an organ resulting from a specific primary abnormality of development, such as a congenital heart or neural tube defect (NTD). *Deformations* are abnormalities of form, shape, or position caused by mechanical forces such as intrauterine molding or constraint. Factors leading to deformations may be extrinsic (e.g., oligohydramnios owing to ruptured membranes) or intrinsic (e.g., oligohydramnios owing to renal agenesis). Deformations may also occur postnatally; for example, an infant may develop a flat head from sleeping in one position. A *disruption* is a morphologic defect that results from breakdown of previously normal tissue. Disruptions can be due to extrinsic forces, internal interferences with a developmental process, or vascular insults. Examples of disruptions include amputations owing to amniotic bands, and gastroschisis and porencephaly, both thought to result from in utero vascular insults.

A *sequence* is a pattern of multiple abnormalities resulting from a single primary anomaly or mechanical factor; it may be a malformation, deformation, or disruption. An example is Potter sequence, in which oligohydramnios from any cause leads to similar features of fetal compression: characteristic facial features and abnormal positioning of the hands and feet. A *syndrome* is a pattern of multiple abnormalities known to have a common, specific cause. An example is Down

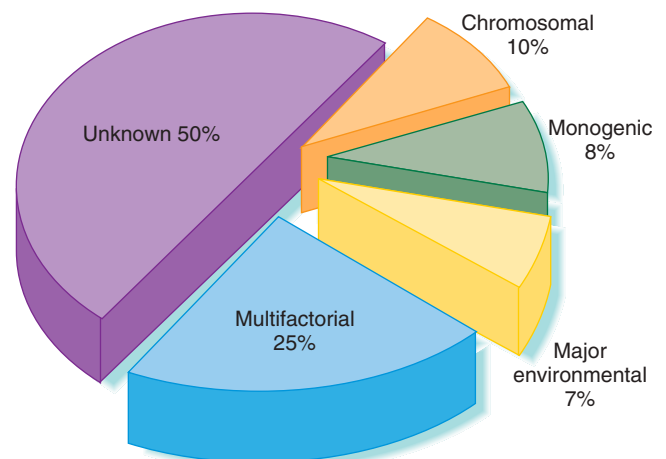


FIG 2-1 Prevalence of genetic diseases in the population. (From Carlson BM: Human Embryology and Developmental Biology, 3rd ed. Philadelphia, Mosby/Elsevier, 2004, used with permission.)