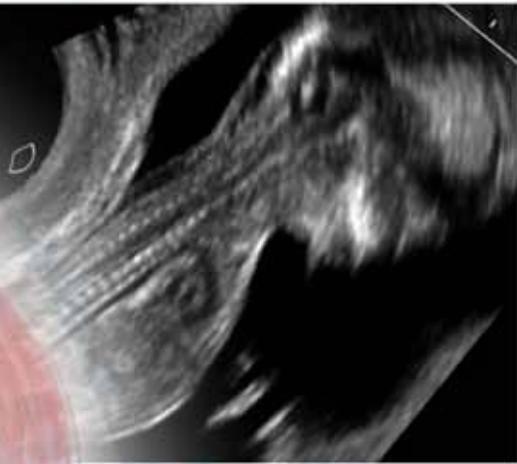


Evolving applications of first-trimester ultrasound

➡ Thanks to advances in technology and increasing understanding of fetal development, it is now possible to assess fetal anatomy in the first trimester in some pregnancies

Ilan E. Timor-Tritsch, MD, and Simi K. Gupta, MD



During the first trimester, ultrasonography can facilitate evaluation of the fetal spine and may be especially important in patients who have a history of spina bifida.

In recent years, prenatal screening and testing have begun to shift from the second trimester to the first. Ultrasonographic evaluation—a large component of fetal testing—is also applied earlier in gestation to provide information to clinicians and patients about the integrity of the pregnancy. The shifting of the classic, “gold standard” anatomy scan to the first trimester was made possible by high-frequency transvaginal transducers and by greater understanding of the early signs of fetal pathology.

First-trimester ultrasound (FTUS) achieves several important goals:

- It can detect many pregnancy complications, such as placental abnormalities.
- It can highlight some chromosomal anomalies.
- It can include a detailed fetal anatomy scan, if desired.

Scanning can be performed transvaginally or transabdominally, although the

transvaginal route is preferred during the first trimester, because of the high-resolution images it yields. Clear images improve reliability and accuracy. However, at times, a combination of transvaginal and transabdominal scanning may be preferred.

In this article, we describe both basic and advanced FTUS applications.

An important application: emergency imaging

FTUS can help determine the source of any first-trimester vaginal bleeding, differentiating between threatened abortion, early fetal demise, and ectopic pregnancy.

It also can help confirm the pregnancy location, revealing whether it is a definitive intrauterine or definitive ectopic gestation, although FTUS can also be inconclusive in this application. An inconclusive scan in the presence of a positive pregnancy test is known as a “pregnancy of unknown location.”¹ Along with FTUS, serial measurement of serum human chorionic gonadotropin (hCG) is used to arrive at a diagnosis.

Two underreported complications in women who have undergone cesarean delivery are cesarean-scar pregnancy and early placenta accreta.² As the rate of cesarean delivery has increased, so have these two serious complications. Successful treatment depends on their identification at the earliest possible time.³ Early diagnosis is feasible—even before the time of first-trimester screening—if the sonographer focuses on the lower anterior uterine segment.^{4,5}

IN THIS ARTICLE

Establishing fetal viability
page 38

Basic elements of a fetal anatomy scan
page 39

Can first-trimester ultrasonography predict the risk of preeclampsia?
page 43

CONTINUED ON PAGE 38



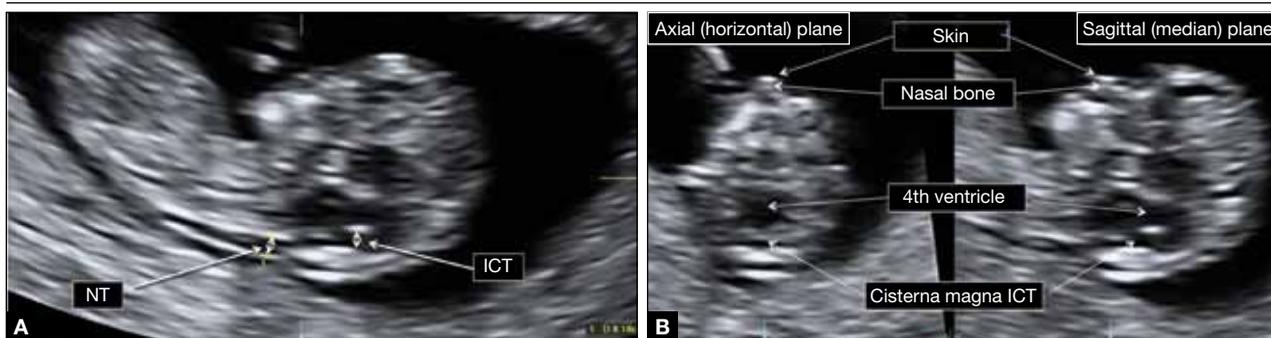
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The authors report no financial relationships relevant to this article.

FIGURE 1 First-trimester cranial anatomy



A. The nuchal translucency (NT) and intracranial translucency (ICT). **B.** Nasal bone and ICT.

Fetal viability can be established

High-frequency transvaginal probes allow earlier and more accurate diagnosis of embryonic and fetal viability, even in the first trimester, through the assessment of cardiac activity.

In addition, early pregnancies can be dated accurately by sequential sonographic visualization of the gestational sac, yolk sac, embryonic pole, cardiac activity, and amniotic sac. Measurement of the fetal crown-rump length (CRL) also provides landmark information that aids in dating the pregnancy and managing different aspects of the gestation.

The first trimester is also the best time to identify, date, and evaluate multifetal gestation in regard to chorionicity and amniocity. These variables, which are important in determining optimal management and potential outcomes, are more difficult to determine later in pregnancy.

Once viability is confirmed, FTUS and biochemical testing can be used for aneuploidy screening between 11 and 13-6/7 postmenstrual weeks. Sonographic measurement of the anechoic space between the skin and the edge of the soft tissue that overlies the cervical spine—otherwise known as nuchal translucency (NT)—is used to determine the likelihood that the fetus is aneuploid (**FIGURE 1**). NT measurement alone has sensitivity of 64% to 70% for the detection of Trisomy 21, as well as a false-positive rate

of 5%. When NT assessment is combined with the measurement of maternal serum pregnancy-associated plasma protein A and free serum hCG, sensitivity increases to 82% to 87%.⁶ When maternal age is factored into the equation, sensitivity may exceed 90%.⁷

Assessment of the nasal bones, ductus venous flow, tricuspid regurgitation, and hepatic artery flow in combination with NT measurement can also improve sensitivity in the detection of Trisomy 21.⁷

Advanced applications include an assessment of fetal anatomy

At the time of NT assessment, FTUS can be broadened to evaluate the age-related anatomy of fetal organs and organ systems.⁸ At 12 postmenstrual weeks, approximately 40% to 50% of common, ultrasonographically discernible anomalies are already present.^{9,10}

The American Institute of Ultrasound in Medicine (AIUM) recommends that “embryonic/fetal anatomy appropriate for the first trimester should be assessed” during the first trimester.¹¹ Although AIUM provides no details about the list of structures to be evaluated, it does provide a guideline for the second-trimester anatomy scan that can be adapted easily for use in the first trimester.

The International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) will soon publish guidelines for FTUS—including its use as an anatomy scan. The ability to



The American Institute of Ultrasound in Medicine recommends first trimester assessment of embryonic/fetal anatomy, with further assessment, as appropriate, in the second trimester

American Institute of Ultrasound in Medicine: Basic elements of a standard second-trimester fetal anatomy scan*

Focus	Structures
Head, face, neck	Cerebellum Choroid plexus Cisterna magna Lateral cerebral ventricles Midline falx Cavum septi pellucidi Upper lip Nuchal fold**
Chest	4-chamber view Outflow tracts, if feasible
Abdomen	Stomach: presence, size, and situs Kidneys Bladder Umbilical cord insertion site (into fetal abdomen) Umbilical cord vessel number
Spine	Cervical, thoracic, lumbar, and sacral
Extremities	Legs and arms: presence or absence
Sex	

* A more detailed fetal anatomic examination may be indicated if an abnormality is found on the standard examination.

** May be helpful during a specific age interval to detect any increased risk of aneuploidy.

assess anatomic structures in the first trimester depends on the quality of the equipment as well as the training of the sonographer or sonologist.

Second-trimester follow-up is recommended

Because some congenital anomalies cannot be identified in the first trimester (or may progress into the second trimester), a follow-up US in the second trimester always should be recommended. AIUM guidelines for a second-trimester anatomy work-up mandate the visualization and documentation of a number of structures and organs (TABLE).

What to look for during first-trimester imaging

Before we describe which anatomic structures and fetal anomalies can be assessed

during the first trimester, we'd like to highlight two articles that provide a comprehensive list of anomalies that have been identified in the first trimester—one by Fong and colleagues and the other by Syngelaki and colleagues.^{10,12} Additional ultrasound images are available in the VIDEO that accompanies this article at obgmanagement.com (see the QR code at right). We'd also like to point out that it is important to evaluate the cervix in the first trimester, keeping the risk of preterm delivery in mind, as well as the maternal uterus and adnexae (looking for pathology) and the placenta (looking for abnormalities) (FIGURE 2, page 40).

Imaging of the head and neck

The cranial bones should be visible at the time of FTUS. Their absence—acrania, which leads to anencephaly—can be diagnosed even before 12 weeks' gestation.

WATCH A VIDEO!

 First-trimester fetal ultrasonography showing anatomic features

by Ilan Timor-Tritsch, MD, and Simi Gupta, MD

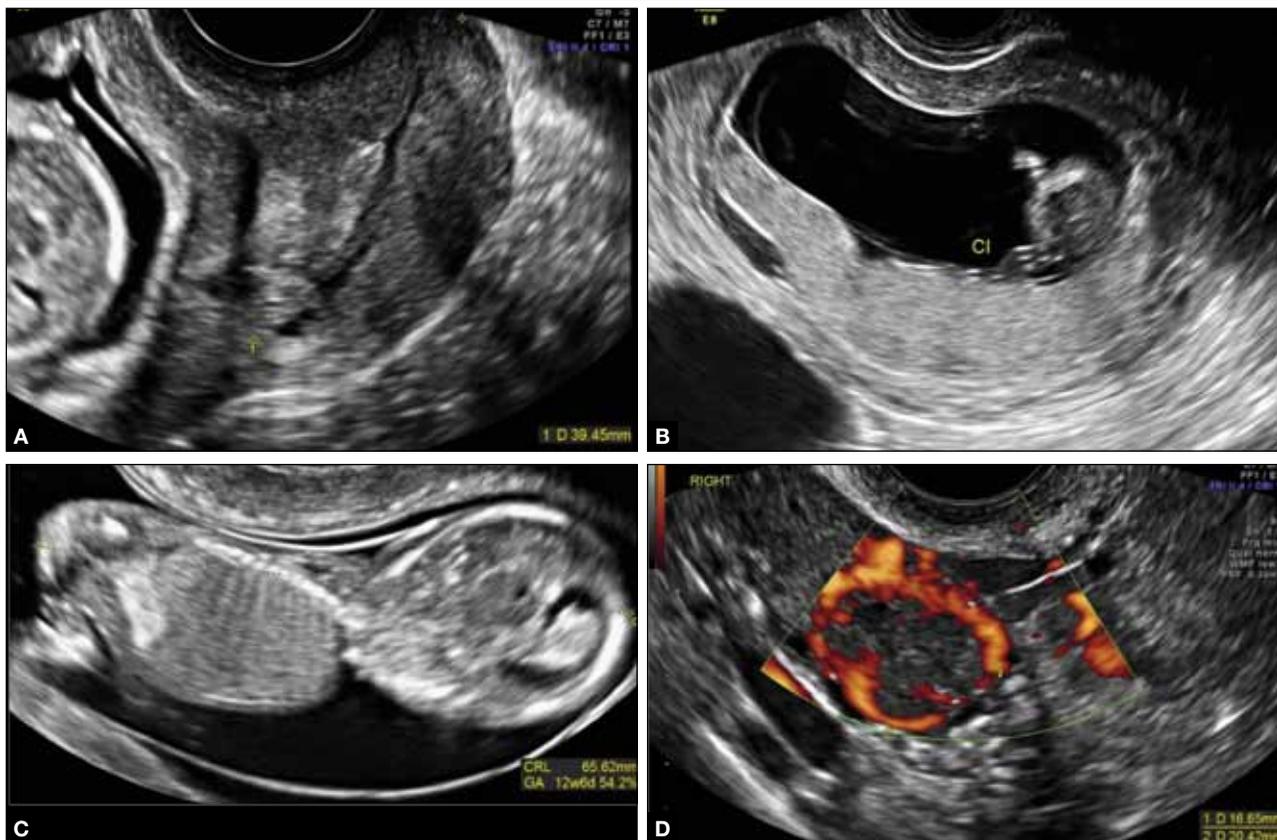


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CONTINUED ON PAGE 40

FIGURE 2 Cervix, placenta, crown-rump length, and maternal adnexae

First-trimester imaging of: **A.** Cervix (transvaginal view). **B.** Placental cord insertion. **C.** Crown-rump length. **D.** Right ovary with the corpus luteum.

At 11 to 14 postmenstrual weeks, the US image of the brain is dominated by the two relatively prominent lateral ventricles, which are filled with the choroid plexuses (**FIGURE 3**, page 41). In this age range, the cortical mantle appears to be relatively thin, which is normal. The two hemispheres of the brain should be symmetrical in size, divided by the important midline falx. Ventriculomegaly can be diagnosed relatively early if the choroid plexuses do not fill the ventricles. Significant ventriculomegaly identified during the first trimester is an ominous sign, usually pointing to a poor outcome, but should be confirmed in later scans.

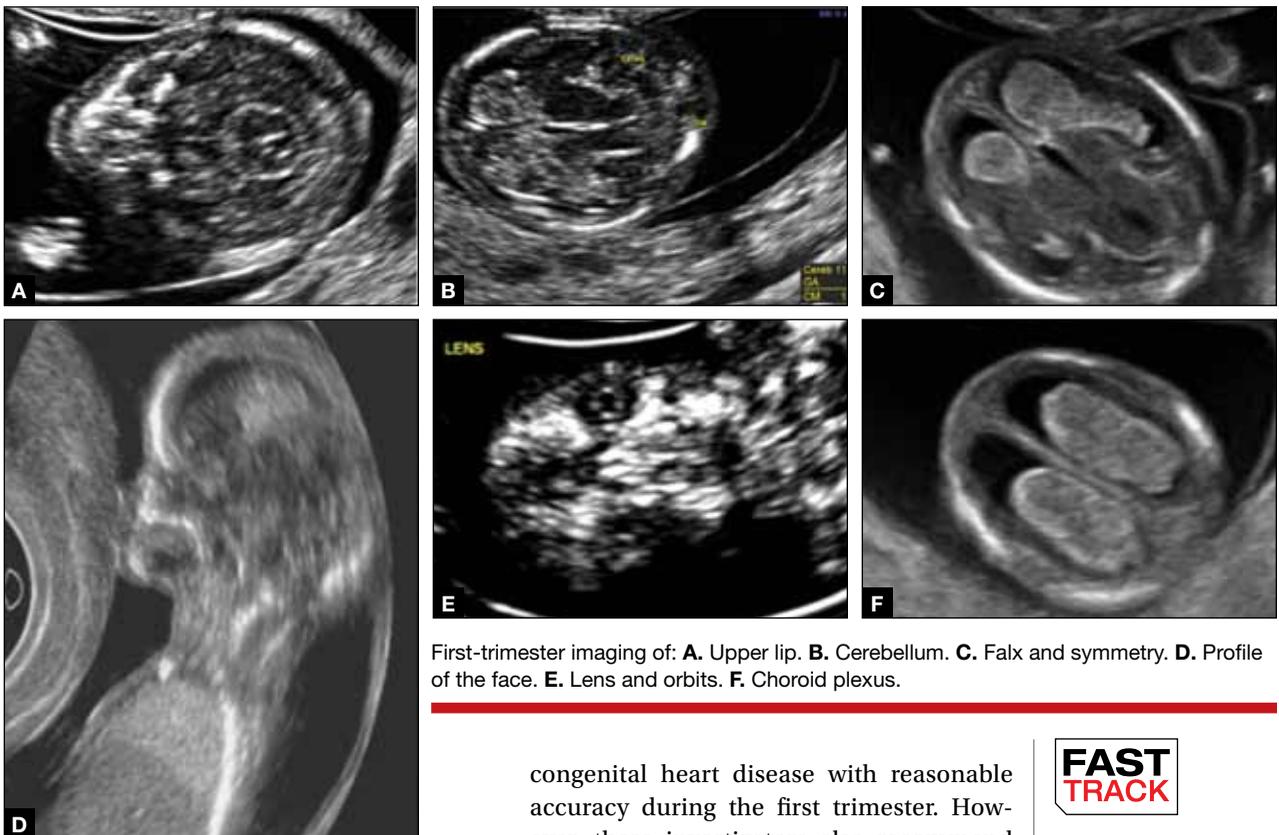
Holoprosencephaly, which presents with clear communication between the frontal horns or the entire lateral ventricles, is without a doubt one of the earliest detectable brain pathologies (along with anencephaly).

In recent publications, researchers have introduced the intracranial translucency (ICT) for early diagnosis of spina bifida.¹³ More large-scale research is needed to determine the sensitivity and specificity of this finding before it can be recommended for routine clinical use.

Other neuroanatomic structures, such as the posterior fossa with the cerebellar hemispheres and cistern magna, the corpus callosum, and the cava (cavum septi pellucidi and Vergae) complete development later; they are better evaluated in the second trimester. However, early observation of a grossly dilated cisterna magna or a large anechoic midline structure should be followed up in a more detailed, later scan.

The neck and nasal bone can be evaluated to help determine the risk for aneuploidy, with special attention focused on the nuchal

FIGURE 3 The head and neck



First-trimester imaging of: **A.** Upper lip. **B.** Cerebellum. **C.** Falx and symmetry. **D.** Profile of the face. **E.** Lens and orbits. **F.** Choroid plexus.

area of the fetus. Measure the NT in the perfect median plane, according to rigorous protocols.^{14,15} During the scan, we also suggest that you attempt to visualize a cross section of the neck. The detection of septations may change the diagnosis from a thick NT to septated cystic hygroma colli. The face, profile, orbits, and lenses are also visible in the first trimester (FIGURE 3), and anomalies such as micrognathia, cleft lip and palate, and anomalies of the profile have been reported, among others.¹⁰

Structures of the chest

The thorax, the integrity of the diaphragm and several basic features of cardiac anatomy can be evaluated during the first trimester, including cardiac position, the four-chamber view, and the outflow tracts (FIGURE 4, page 42). With minimal added effort, the aortic and ductal arches can be added to this list. Multiple investigators have concluded that an experienced sonographer can identify

congenital heart disease with reasonable accuracy during the first trimester. However, these investigators also recommend a second-trimester US to follow up on the anatomy or any abnormalities of the heart identified in the first trimester.¹⁶

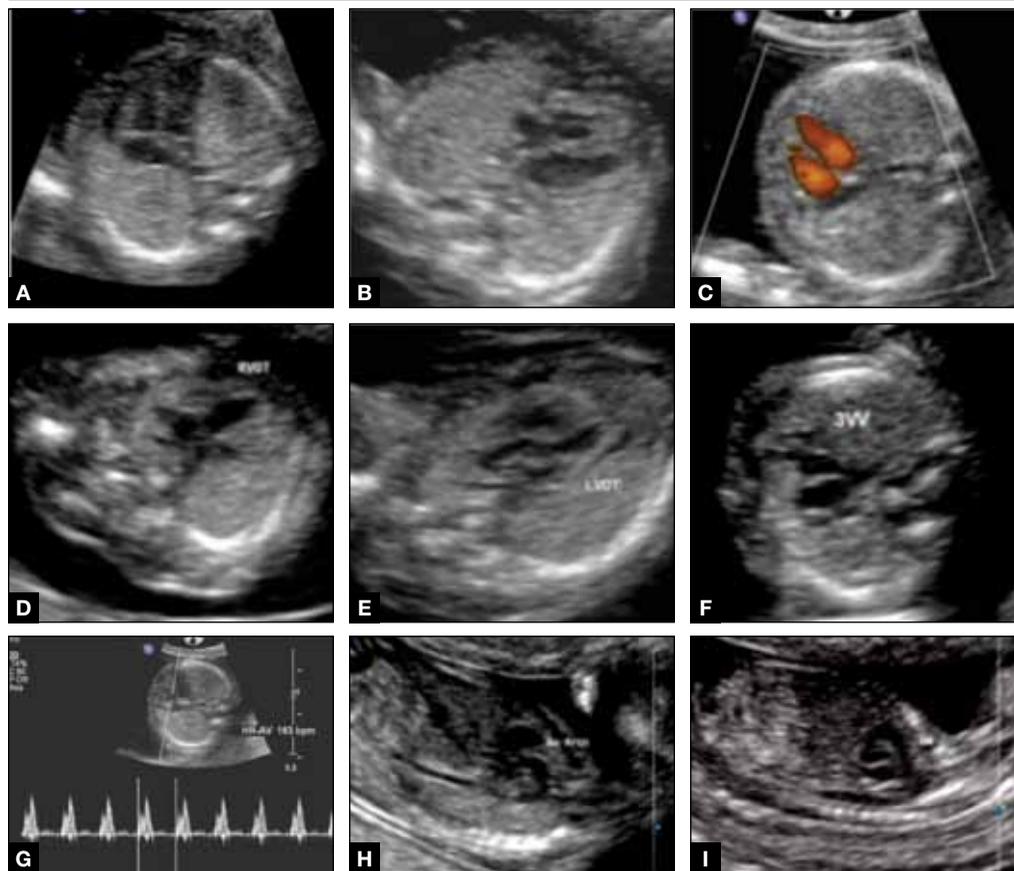
Structures of the abdomen

The presence or absence and location of the anechoic stomach and bladder should be determined in the first trimester (FIGURE 5, page 43). By observing the heart axis and location of the stomach on the left side, normal situs solitus can be confirmed. The bladder is the second hypoechoic structure in the abdomen and should be visualized easily. Its absence or enlargement may be a sign of other congenital anomalies.

The slightly hyperechoic appearance of the normal kidneys should be documented, along with their tiny, anechoic renal pelvises. The renal arteries can be visualized easily via color Doppler (used as briefly as possible). If they cannot be detected, consider possible agenesis of one or both kidneys. Renal dysplasia, renal parenchyma that has

FAST TRACK

The slightly hyperechoic appearance of the normal kidneys should be documented in any first-trimester fetal anatomy scan

FIGURE 4 Thorax

First-trimester imaging of: **A.** Apical four-chamber view. **B.** Septal four-chamber view. **C.** Color Doppler four-chamber view. **D.** Right outflow tract. **E.** Left outflow tract. **F.** Three-vessel view. **G.** Heart rate. **H.** Aortic arch. **I.** Ductal arch.

**FAST
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Look for umbilical cord insertion into the abdomen only after 12 postmenstrual weeks

been replaced by multiple cysts, and ectopic kidneys can also be identified in the first trimester.

Look for umbilical cord insertion into the abdomen only after 12 postmenstrual weeks because the physiologic umbilical hernia is still present before that age. You can observe the coursing of the two umbilical arteries on either side of the bladder to determine the umbilical cord vessel number using color or power Doppler (again, as briefly as possible).

Although fetal gender may be determined in the first trimester, it may not be accurate before 12 weeks, when it can be based on the angle of the genital tubercle.¹⁷

Imaging of the extremities

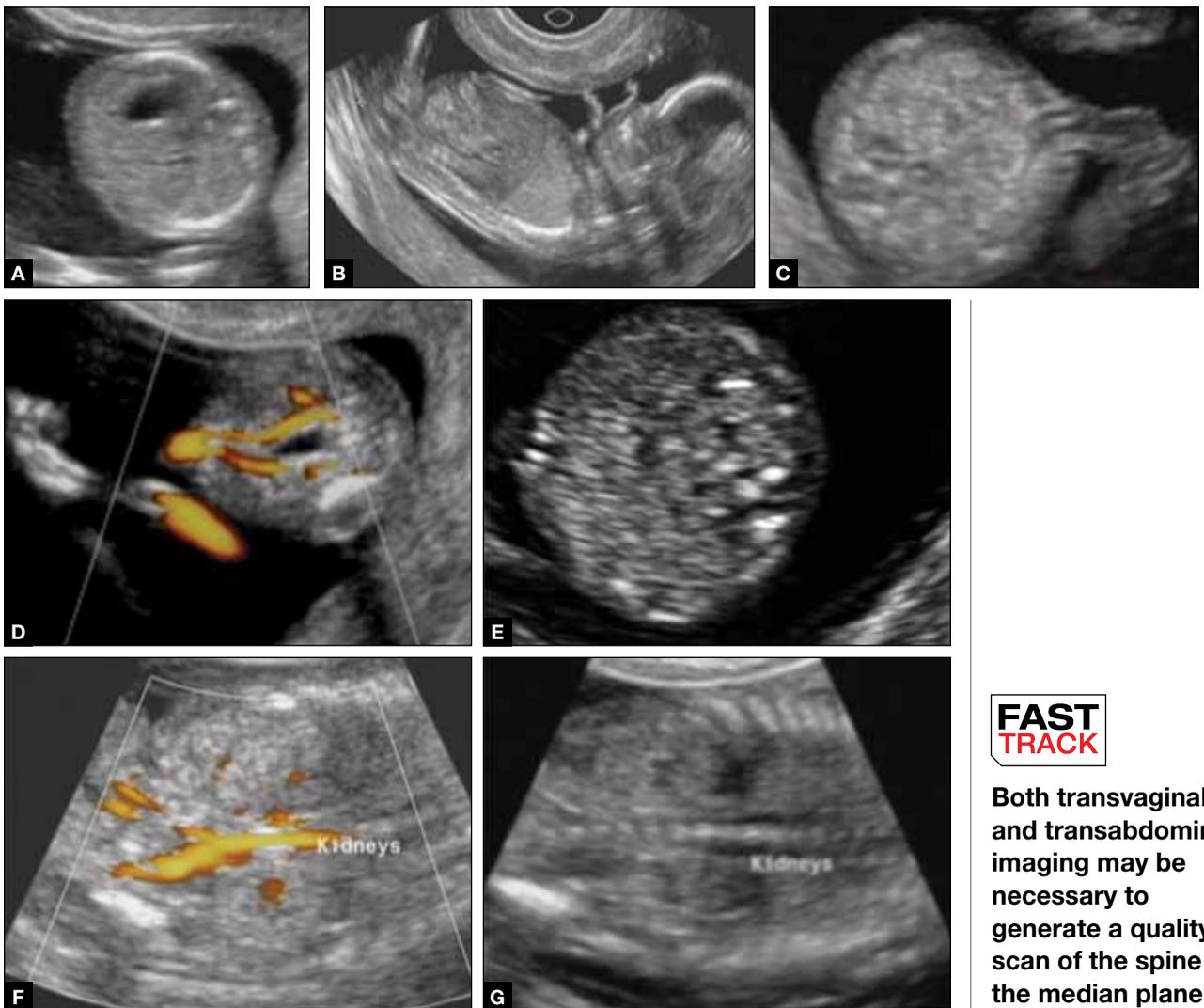
Any FTUS assessment of fetal anatomy

should include visualization of the extremities. Ample amniotic fluid and active fetal movements facilitate this examination. Be sure to document the presence or absence of long bones in the upper and lower extremities, as there have been numerous reports of anomalies affecting these structures in the first trimester (**FIGURE 6**, page 44). Limb abnormalities and isolated deformities (eg, club foot)—not to mention the absence of a limb segment—can be observed and evaluated in the first trimester. Even the relatively small hands and phalanges can be assessed if pathology is suspected.¹²

The fetal spine

Evaluate the spine using a median, longitudinal, and serial transverse section (**FIGURE 6**)

FIGURE 5 Abdomen



First-trimester imaging of: **A.** Stomach. **B.** Diaphragm. **C.** Cord insertion. **D.** Two umbilical arteries and the bladder. **E–G.** Kidneys.

FAST TRACK

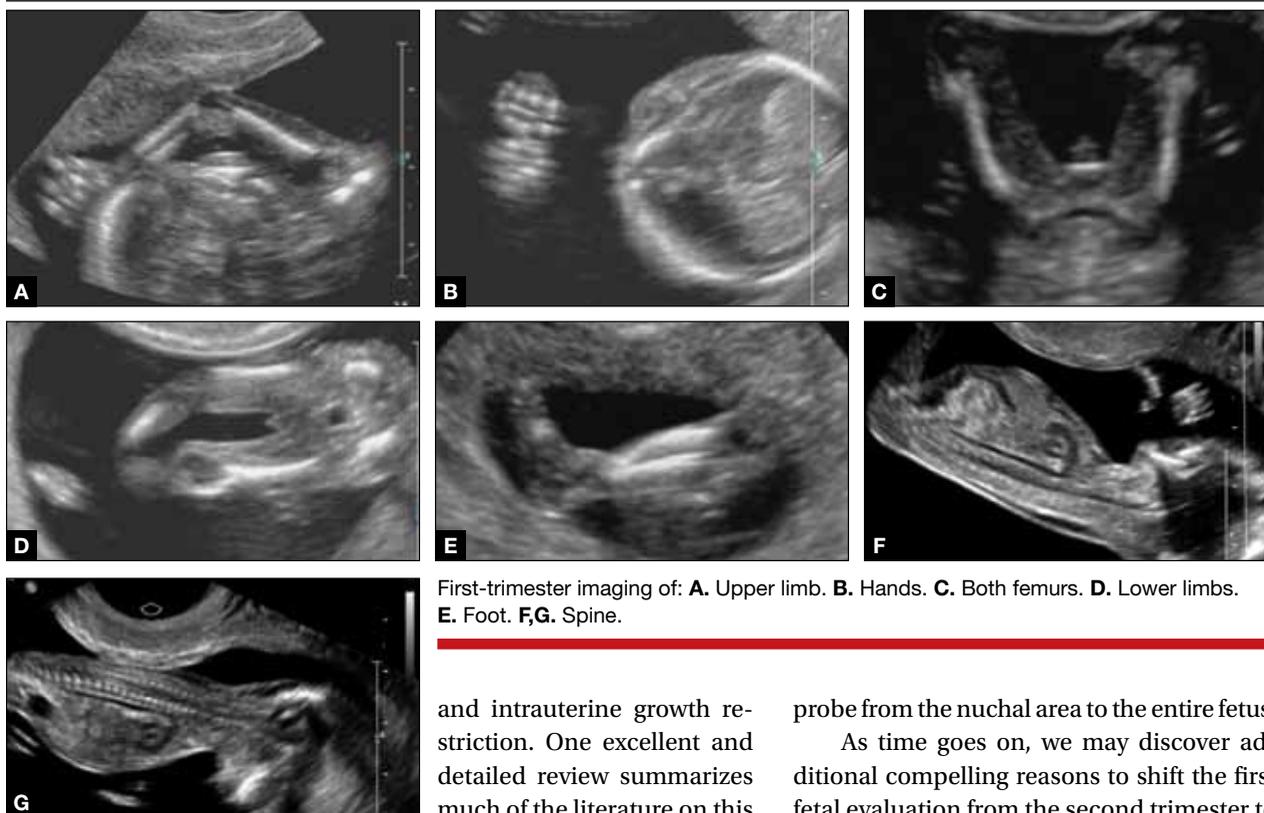
Both transvaginal and transabdominal imaging may be necessary to generate a quality scan of the spine in the median plane

to confirm integrity. Also attempt to assess the overlying skin by visualizing the area when the fetus moves away from the uterine wall. A combination of transvaginal and transabdominal scanning techniques may be necessary to generate a quality image of the spine in the median plane.

As we mentioned earlier, the intracranial translucency may be utilized in the future as a tool to screen for neural tube defects. It is even more important to focus on the spine in patients who have a history of spina bifida.

Can FTUS predict the risk of preeclampsia?

Recent data on an experimental application of FTUS suggest that it may be used one day to evaluate uterine artery Doppler velocity waveforms as a way of predicting preeclampsia and intrauterine growth restriction. Multiple algorithms have been proposed that combine uterine artery Doppler measurements with maternal history or biochemical markers, or both, to determine the risk of early and late-onset preeclampsia

FIGURE 6 Spine and limbs

First-trimester imaging of: **A.** Upper limb. **B.** Hands. **C.** Both femurs. **D.** Lower limbs. **E.** Foot. **F,G.** Spine.

and intrauterine growth restriction. One excellent and detailed review summarizes much of the literature on this subject.¹⁸

Although more prospective studies are needed to elucidate fully which women should be screened, which algorithm should be applied, and which intervention is best, this application of FTUS is highly promising and is likely to increase in coming years in the United States.

The many benefits of FTUS

For at least 20 years, we and others have advocated the use of ultrasound in general and transvaginal ultrasound in particular to identify anomalies in the first and early second trimester. Over that period, advances in transducer technology, faster computers, and deepening understanding of developmental anatomy have facilitated such use. Increasing reliance on first-trimester screening brings more and more women with gestations from 11 to 14 weeks to the obstetrician's office, providing an opportunity to expand the narrow viewing angle of the ultrasound

probe from the nuchal area to the entire fetus.

As time goes on, we may discover additional compelling reasons to shift the first fetal evaluation from the second trimester to the first. In the meantime, we find no reason not to widen the viewing angle of transducer probes to assess fetal anatomy at the first-trimester scan. 📌

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