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### Abbreviations:

MIP = maximum intensity projection  
2D = two-dimensional  
3D = three-dimensional

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# Fetal Skeletal Dysplasia: Three-dimensional US—Initial Experience<sup>1</sup>

**PURPOSE:** To compare the prenatal ultrasonographic (US) features of skeletal dysplasia by using two-dimensional (2D) and three-dimensional (3D) US to determine whether 3D US can reveal additional diagnostic information.

**MATERIALS AND METHODS:** Seven pregnant women suspected of having skeletal dysplasia were examined by using 2D US and 3D US. Data regarding the thorax, spine, face, limbs, hands, and feet were compared. Multiplanar and volume-rendered US images were evaluated.

**RESULTS:** The skeletal dysplasias studied included camptomelic dysplasia ( $n = 2$ ), thanatophoric dysplasia ( $n = 1$ ), osteogenesis imperfecta ( $n = 1$ ), arthrogryposis ( $n = 2$ ), and short-limbed dysplasia ( $n = 1$ ). Three-dimensional US, by allowing review in a standard anatomic orientation, was better than 2D US in depicting abnormal spatial relationships such as short ribs, splayed digits, and absent bones. Three-dimensional US enabled the acquisition of additional information in two fetuses with facial abnormalities and in two fetuses with scapular aplasia or hypoplasia (one fetus had both facial and scapular anomalies); it enabled a specific diagnosis in one fetus. The archiving capabilities of 3D US allow the review and manipulation of data after the patient has left the clinic.

**CONCLUSION:** In three of seven patients, 3D US provided additional information in the evaluation of skeletal dysplasias, as compared with 2D US.

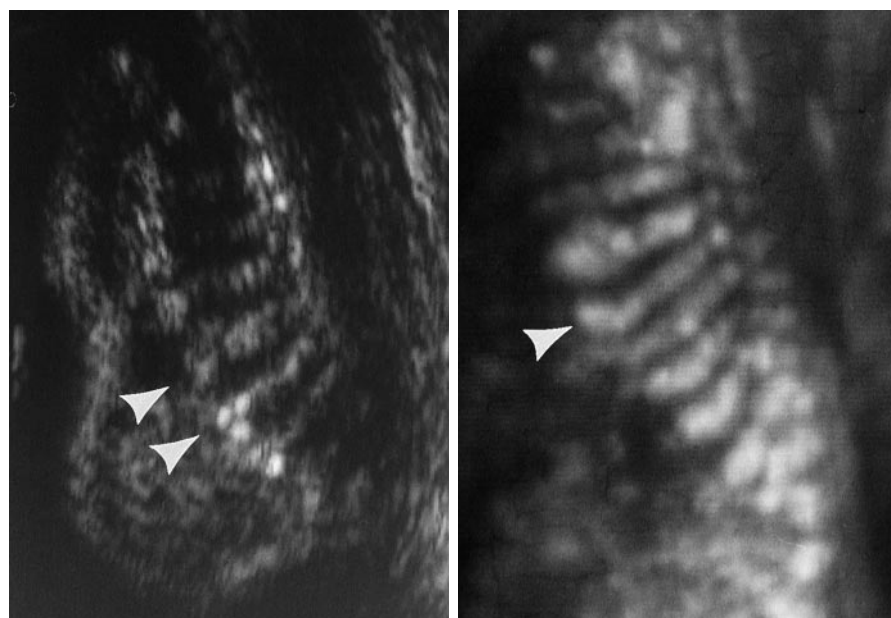
Accurate prenatal diagnosis of skeletal dysplasia allows families to make appropriate decisions for obstetric management and delivery. The specific diagnosis of a skeletal dysplasia in utero requires familiarity with a complex algorithm of ultrasonographic (US) features. Although it is often difficult to determine the specific type of skeletal dysplasia present and whether it is lethal, this information may be extremely valuable in planning obstetric care. Several investigators (1–4) have suggested that recent developments in three-dimensional (3D) US offer advantages in evaluating fetal anomalies. The purpose of this study was to compare the prenatal US features of skeletal dysplasia by using two-dimensional (2D) and 3D US to determine whether 3D US can reveal additional diagnostic information.

## MATERIALS AND METHODS

Seven pregnant women were initially examined by using both 3.5- and 5.0-MHz transducers with models XP/128 (Acuson, Mountain View, Calif) and HDI (Advanced Technology Laboratories, Bothell, Wash) 2D US units at the University of California, San Diego. The majority of these women were referred from outside institutions because of abnormal US findings, and one of them had an abnormally low  $\alpha$ -fetoprotein level. Following the completion of the 2D US examinations in which a skeletal dysplasia was identified, the women were recruited for 3D US studies after investigational review board approval and written informed consent were obtained. The 3D US data were obtained within 3 weeks after the correlative conventional 2D US data were obtained at fetal gestational ages that ranged from 19 to 34 weeks.

The 3D US data were acquired by using a Combison 530 unit (Medison, Pleasanton,

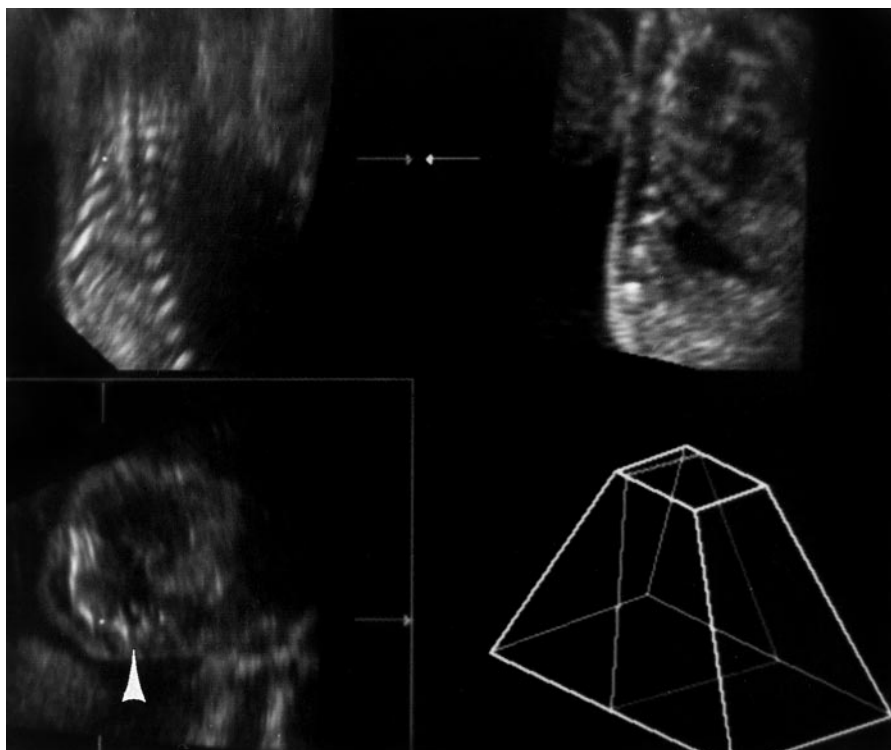
**Figure 1. Case 2.** Thorax of a fetus with thanatophoric dysplasia at 24 weeks gestation. (a) Sagittal 2D US image of the chest shows the ribs (arrowheads), but it is difficult to determine whether they are short. (b) The multiplanar image shows the thorax in the coronal (top left), sagittal (top right), and transverse (bottom left) planes, and a truncated pyramid (bottom right) that demonstrates the plane of section of the highlighted image (bottom left). The arrowhead points to the spine. (c) Sagittal volume-rendered MIP image enables adequate visualization of the shortened ribs. The twelve ribs are demonstrated, and the arrowhead indicates the seventh rib. The shortened ribs are not appreciated at multiplanar imaging. In b, the ribs on the left side of the images are not well depicted due to their position in the lower portion of the volume acquisition.



a.

c.

Calif) with a 5.0-MHz, mechanically swept annular-array volume transducer. Approximately six volume data sets were acquired per patient (range, four to ten), with an acquisition time of approximately 5 seconds per volume. The volume data collected included those in the thorax, spine, face, limbs, hands, and feet. Rendering of volume data required 3–15 minutes per data set. Three types of 3D US volume-rendering display algorithms were used: surface, light, and maximum intensity projection (MIP). The surface- and light-rendered images showed a smoothly contoured view, almost sculpturelike, which was used to evaluate the facial features and extremities. The inner bone structures were highlighted on the MIP images, which enabled visualization of the skeleton—especially the spine, ribs, and scapulae—with higher contrast (Fig 1). Both the surface-rendered and volume-rendered MIP images were displayed and rotated on the graphics display monitor. This allowed a full 360° rotation of the volume-rendered image of the fetal skeleton and thus visualization in the standard anterior, posterior, and lateral projections, as well as at many additional degrees of obliquity. The surface-rendered images of the fetal faces were rotated by using a 180° arc; this provided views of the lateral (ie, profile) to anterior projections. The surface-rendered images of the extremities were viewed by using 360° of rotation.

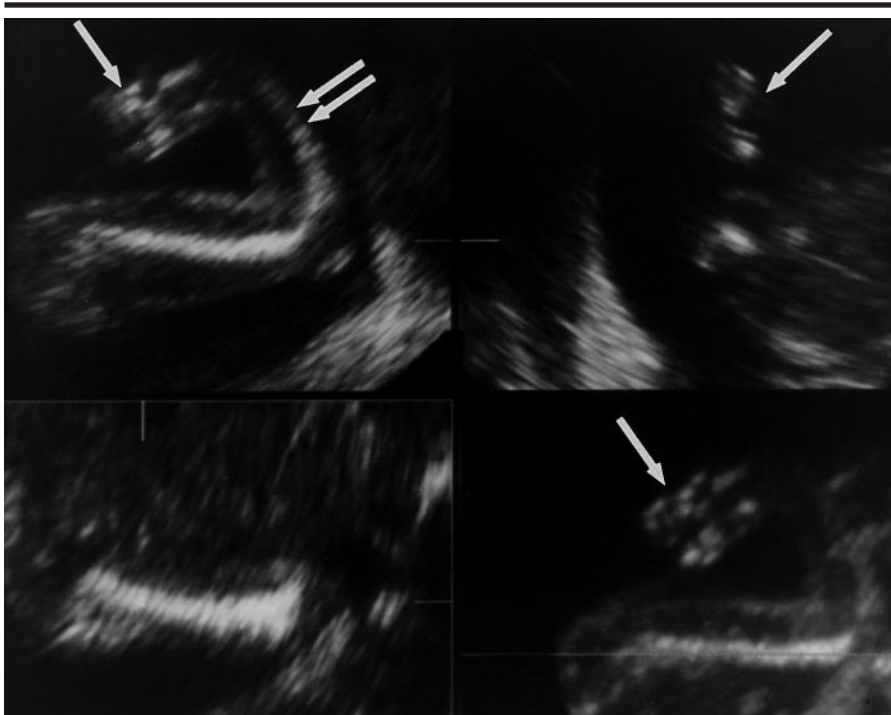


b.

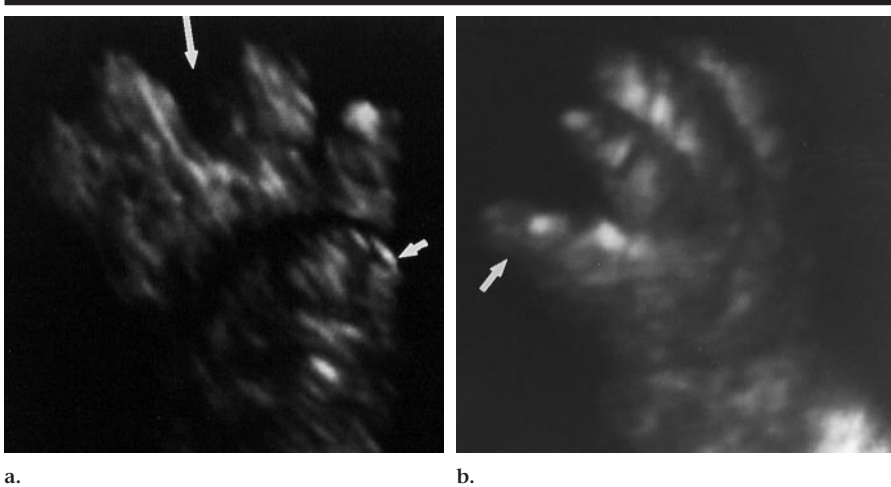
The 3D US volume data were saved to permit further manipulation and review of the data after the patient had left the clinic. These data were reviewed to identify the US features associated with skeletal dysplasia. The 3D US volume data were reviewed by rotating the images into a standard anatomic orientation and were dis-

played as simultaneous tomographic images in three perpendicular planes—transverse, sagittal, and coronal—with a truncated pyramid in the bottom right corner demonstrating the plane of section (Fig 1). The volume was rotated so that the examiner was looking directly at the upright fetus. The limbs and distal extremities were evaluated subjectively

for shortening, bowing, and abnormal positioning by using multiplanar and MIP volume-rendering techniques. The spine and thorax were evaluated for ossification, platyspondyly, and other anomalies by using volume-rendering MIP techniques. The face was evaluated by using multiplanar and surface and light volume-rendering techniques. The 3D US images



**Figure 2. Case 7.** Abnormal hand in a fetus at 19 weeks gestation. The top left and right images and bottom left image are three orthogonal views of the forearm. The hand (single arrows) is contracted, the wrist is abnormally flexed, and the forearm (double arrow) is shortened. The top left image shows the entire upper extremity, the top right image is a transverse view through the hand and humerus, and the bottom left image is a sagittal view through the humerus. The bottom right image is a volume-rendered image of the forearm and hand with soft-tissue and surface weighting.



**Figure 3. Case 2.** Abnormal hand in a fetus with thanatophoric dysplasia at 24 weeks gestation. (a, b) Two volume-rendered US images of the rotating hand, obtained with a combined surface- and light-rendering technique, show brachydactyly and abnormal widening (long arrow) between the third and fourth digits. The thumb (short arrow in a and b) is shown (a) coming straight out of the image and (b) as the image is being rotated.

were compared with the 2D US images to determine whether additional, equivalent, or less information was obtained. The cases were reviewed and compared by two physicians (D.H.P., K.V.G.) by using consensus only.

Information regarding the influence of 3D US imaging on patient treatment was requested from the primary physician and genetic counselor; this was predominantly related to whether the examination findings changed the pa-

tient's mind about termination of pregnancy or the timing and/or mode of delivery.

## RESULTS

The diagnoses for the seven fetuses in this study were, on the basis of neonatal outcome, camptomelic dysplasia in two cases; thanatophoric dysplasia, one case; osteogenesis imperfecta, one case; arthrogryposis, two cases; and unspecified short-limbed dysplasia, one case. Five of seven fetuses had long-bone abnormalities, which are summarized in Table 1. All of the bone abnormalities were identified with both 2D US and 3D US. Coronal, sagittal, and transverse planar images of the long bones permitted a systematic review and depicted the relationship between the long bones and the hands and feet. One fetus (case 4) with arthrogryposis had a dislocated hip at delivery that was not identified by using either 2D US or 3D US; it is not known whether this abnormality was present prior to delivery.

Four fetuses had hand abnormalities, and four had foot abnormalities (Table 1). MIP and surface-rendered images were helpful in conceptualizing the abnormalities, due to the rotation of the 3D US surface-rendered images of the abnormally positioned hands and feet into standard planes for multiplanar viewing, such as in a fetus with a claw hand and reduced number of digits (case 7). In the evaluation of this fetus, the volume-rendered images provided an improved appreciation of the hand deformity (Fig 2). The fetus with thanatophoric dysplasia (case 2) had shortened digits consistent with brachydactyly and splaying between the third and fourth digits (Fig 3). The splaying was more clearly visualized on the volume-rendered images that were rotated on the video monitor than on the conventional 2D US planar images. The volume data allowed the hands of all fetuses to be evaluated after the patient had left the clinic. The number of digits was not specifically counted during the conventional 2D US real-time examination in one case, but it was confirmed to be normal with the volume data. The foot abnormalities in four fetuses were identified with both 2D US and 3D US (Table 1). The 3D US volume-rendered images were more helpful in evaluating positional changes in the hands and feet than the 2D US planar images. The real-time capability of conventional 2D US enabled demonstration of fixed positioning over

**TABLE 1**  
**Summary of US and Pathologic Findings of the Spine, Long Bones, and Hands and Feet**

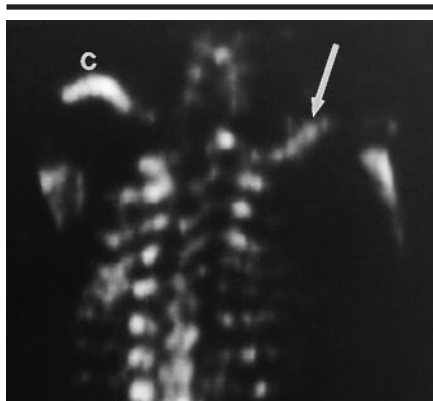
Fetus No./ Gestational Age (wk)	Diagnosis	Spine*			Long Bones			Hands and Feet		
		2D US	3D US	Pathologic	2D US	3D US	Pathologic	2D US	3D US	Pathologic
1/26	Osteogenesis imperfecta	Normal	Normal	NE	Short, bowed, thick	Short, bowed, thick	Short with fractures	NE	NE	NE
2/24	Thanatophoric dysplasia	Normal	Normal	NE	Short	Short	Short	Trident hand	Trident hand	Trident hand, brachydactyly
3/30	Camptomelic dysplasia	NTD, scoliosis	NTD, scoliosis	Scoliosis	Short	Short, bowed	Short, bowed	NE	NE	NE
4/21	Arthrogryposis	Normal	NE	NE	Normal bones and hips	Normal bones and hips	Dislocated hip	Distal foot extension, ulnar deviation	Distal foot extension, ulnar deviation	Pronated limbs, contractures
5/34	Arthrogryposis	Normal	NE	NE	Normal	Normal	NE	Clenched hands, dorsiflexed feet	Clenched hands, dorsiflexed feet	Dorsiflexed (rocker bottom) feet
6/21	Camptomelic dysplasia	NTS	NTS	NTS	Bowed	Bowed	Bowed ulnar deviation at wrist	Normal hand, splayed foot	Normal hand, bifid foot	Bifid foot
7/19	Short-limbed dysplasia	Normal	NE	NE	Short, single bones	Short, single bones	NE	Right club foot, claw	Right club foot, claw	Claw

Note.—NE = not evaluated.  
 \* NTD = neural tube defect, NTS = narrow thoracic spine.

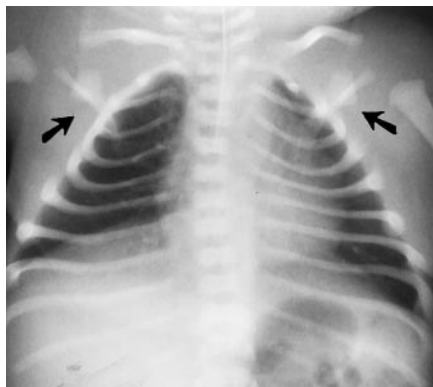
**TABLE 2**  
**Summary of US and Pathologic Findings of the Chest, Ribs, Scapulae, and Head and Face**

Fetus No./ Gestational Age (wk)	Diagnosis	Chest Size			Ribs			Scapulae			Head and Face		
		2D US	3D US	Pathologic	2D US	3D US	Pathologic	2D US	3D US	Pathologic	2D US	3D US	Pathologic
1/26	Osteogenesis imperfecta	Small	Small (bell)	NE	Fracture	Crinkled, short	NE	Present	Present	NE	Normal	Normal (close to uterine wall)	Flat nasal bridge, skull fracture
2/24	Thanatophoric dysplasia	Small	Small	NE	Short	Short	NE	Present	Present	NE	Midfacial hypoplasia	Midfacial hypoplasia	Midfacial hypoplasia
3/30	Camptomelic dysplasia	Normal	Normal	Normal	Normal	Normal	11 Pairs	Absent on rescan	Absent	Absent	Normal	Micrognathia, flat face	Micrognathia, flat face
4/21	Arthrogryposis	NE	NE	NE	NE	NE	NE	NE	NE	NE	Normal	Normal	Mild micrognathia
5/34	Arthrogryposis	NE	NE	NE	NE	NE	NE	NE	NE	NE	Normal	Possible flat face	NE
6/21	Camptomelic dysplasia	Normal	Normal	Normal	Normal	Normal	Normal	NE	Hypoplastic	Hypoplastic	Normal	Flat face	Flat face
7/19	Short-limbed dysplasia	NE	NE	NE	NE	NE	NE	NE	NE	NE	NE	NE	NE

Note.—NE = not evaluated.



a.



b.

**Figure 4.** Case 6. Hypoplastic scapulae in a fetus at 21 weeks gestation. (a) Volume-rendered image of the thorax and spine, obtained by using a MIP technique, shows the upper portion of the scapula (arrow), but the lower portion of the scapula is hypoplastic and not ossified. The scapula on the opposite side is not visible on this image. C = clavicle. The differentiation between the scapulae and clavicles was made on the multiplanar views and rotating volume-rendered view (neither are shown). (b) Anteroposterior radiograph shows the hypoplastic scapulae (arrows).

time and thus proved to be helpful in the evaluation of the limbs of two fetuses with arthrogryposis.

The unsuspected scapular hypoplasia in one case (Fig 4) and aplasia in one case were identified in the fetuses with camptomelic dysplasia (cases 6 and 3, respectively) by using the volume-rendered images and were confirmed on the multiplanar images. These findings were not identified during the initial interpretation of the 2D US images; however, the scapular aplasia in one fetus with camptomelic dysplasia was present on the repeat 2D US scan that was obtained after the interpretation of the 3D US images. The scapular abnormalities of one of these fetuses strongly suggested a specific diagnosis of camptomelic dysplasia that was

not identified on the conventional 2D US images.

Two fetuses had rib abnormalities. Short ribs without acute angulation were identified in the fetus with thanatophoric dysplasia at both 2D US and 3D US. Three-dimensional US depicted the short ribs more clearly than did 2D US when the thorax was rotated on the monitor by using volume rendered MIP imaging (Figs 1, 5). Crinkled, angled ribs that were suggestive of fracture were identified in the fetus with osteogenesis imperfecta at both 2D US and 3D US. Three-dimensional US volume rendering, however, made this finding more obvious (Figs 1, 5). Both of these fetuses had corresponding pulmonary hypoplasia that was diagnosed by using thoracic circumference curves with both 2D US and 3D US. The ribs and chest sizes in the two fetuses with camptomelic dysplasia were normal at both 2D US and 3D US and at the time of delivery (one at autopsy, one at physical examination) (Table 2). The ribs were not evaluated in the other three cases.

Two fetuses had spine abnormalities. One fetus had splaying of the posterior elements, scoliosis, and a neural tube defect, and the other had a narrowed bone thoracic canal. These abnormalities were identified at both 2D US and 3D US. The 3D US time-of-flight display monitor provided a constant anatomic orientation of the middle canal in the fetus with scoliosis and thus improved visualization of the abnormal bone elements on the transverse planar images. In addition, small changes in obliquity on the MIP images, with rotation, enhanced the conspicuity of the narrowed posterior elements.

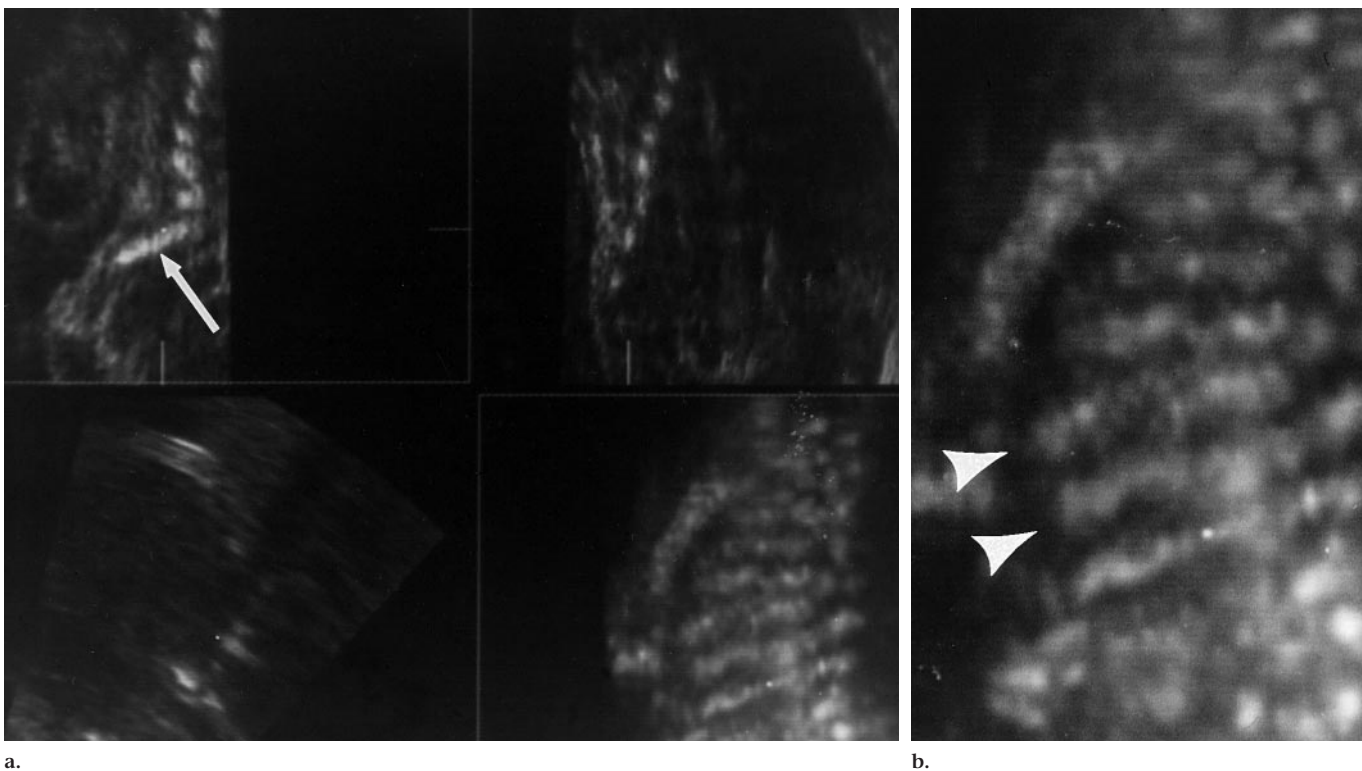
Three fetuses had abnormal facies: micrognathia, midfacial hypoplasia, and flat facies (Table 2). The sagittal plane was important in assessing the profile; the coronal and transverse planar images provided reference information for localizing the true midline sagittal plane. The volume-rendered image of the profile also was helpful. The 3D US-based diagnosis of both the fetuses with camptomelic dysplasia (cases 3 and 6) was flat facies, which was undetected at 2D US (Fig 6). In addition, the micrognathia in one fetus (case 3) was detected only at 3D US. The midfacial hypoplasia in the fetus with thanatophoric dysplasia (Fig 7) was identified at both 2D US and 3D US. A case of mild micrognathia and another of flat nasal bridge were not identified with either 2D US or 3D US. Patient treatment was not influenced by the 3D US findings in any of the cases.

## DISCUSSION

Three-dimensional US offers a way to visualize fetal skeletal dysplasias. The results of our study support the observations of earlier investigators (1,5,6) that 3D US assists in clarifying the spatial relationships of deformed limbs and in demonstrating skeletal abnormalities in a manner that is not possible with conventional 2D US. Both multiplanar and volume-rendered display methods are important in assessing these skeletal abnormalities. In our study, the unique capabilities of 3D US assisted in (a) identifying scapular anomalies that were unsuspected at 2D US, (b) depicting the fetal face on a true midline sagittal image for evaluation of suspected anomalies, (c) appreciating limb abnormalities through rotation, (d) assessing short ribs and chest hypoplasias by enabling the entire fetal skeleton to be viewed on a single image, and (e) archiving data for reassessment, counseling, and teaching.

Three-dimensional US allowed the examiner to rotate volume data into a standard anatomic orientation so that he or she was looking directly at an upright fetus. All data were reviewed by using the same standard format that permitted the scrolling of images parallel to the coronal, sagittal, and transverse planes. These images could be precisely viewed simultaneously with a localizer to reference one image to another within the same volume. The data could also be rotated into oblique planes to optimize viewing, which proved to be helpful in identifying unpaired distal bones. Multiplanar viewing capability allowed precise definition of the imaging plane. It also proved to be helpful in the evaluation of fetal facial profiles and the fetal spine. This ability to manipulate data is an advantage of 3D US compared with 2D real-time US, in which random fetal movement often causes difficulty in evaluating the anatomy.

Volume rendering offers advantages over planar imaging in visualizing skeletal dysplasias. It is important to optimize the rendering display monitor for the information desired, because volume data can be evaluated with different techniques (ie, surface rendering, MIP, and light rendering). The MIP mode enabled the best depiction of the skeletal abnormalities such as limb shortening, scoliosis, and short ribs. The surface mode was excellent for demonstrating complex positional relationships such as club foot, limb contracture, and splayed digits. Surface imaging also provided an almost photographic effect for viewing fetal fa-



**Figure 5. Case 1.** Short ribs in a fetus with osteogenesis imperfecta at 25 weeks gestation. (a) Multiplanar US image shows truncated ribs. The arrow points to the only rib that could be identified in its entirety on a single planar image; this rib would have been difficult to recognize as abnormal without 3D US volume-rendered data. The top left image is a sagittal projection; the top right image, a coronal projection; the bottom left image, a transverse projection; and the bottom right image, a volume-rendered MIP image. (b) On the lateral volume-rendered MIP image, the rib shortening (arrowheads) in this fetus with corresponding chest hypoplasia is highlighted.

cial anomalies such as micrognathia and midfacial hypoplasia (Fig 7), as well as the entire fetus (1,3,7). In our study and in others' (1,7-9), the rotation of volume-rendered data improved the identification of and diagnostic confidence in identifying anomalies of the fetal skeleton. Rib shortening, although diagnosed at both conventional 2D US and 3D US, was easier to appreciate when the fetal images were rotated toward a lateral plane with slight changes in obliquity. The appreciation of positional limb abnormalities also was enhanced by the ability to rotate the image of the limb to an optimal position.

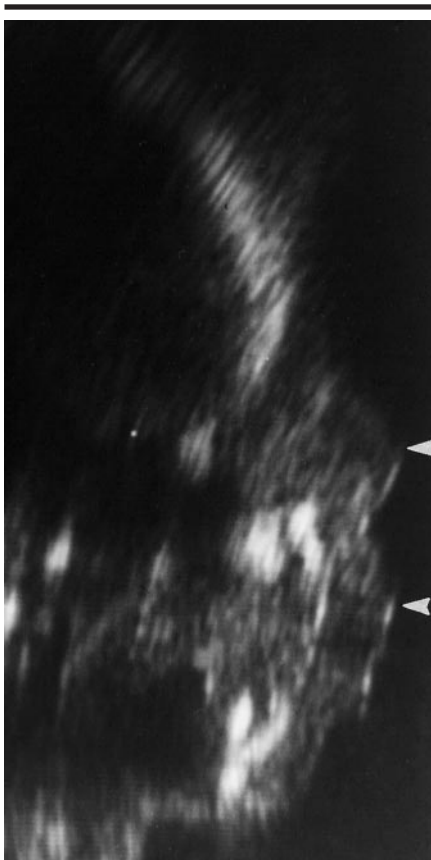
Three-dimensional US enabled the identification of unsuspected scapular anomalies owing to its clearer depiction of the entire volume. The combination of MIP volume rendering and rotation of the volume data allowed the diagnoses of hypoplastic scapulae and absence of scapulae in two fetuses with camptomelic dysplasia. After the 3D US images were interpreted, a repeat 2D US scan was obtained in one case, and at real-time 2D US imaging, the fetus had absent scapulae; this was not noted at the initial interpretation. The hypoplastic scapula of

the second fetus could not be identified in retrospect on the 2D images. In these two cases, the 3D US data allowed a prenatal diagnosis of a specific skeletal dysplasia in utero. The rotation of the volume data enabled viewing in an optimal orientation to identify these abnormalities.

The fetal face is important in the evaluation of skeletal dysplasia. Three-dimensional US provided additional information in the evaluation of facial anomalies. It facilitated diagnoses of mild micrognathia and flat face when the conventional 2D US images were normal. In an article by Merz and colleagues (8), it was noted that a true midline facial profile is present in only 70% of cases of 2D US interpretation. Three-dimensional US imaging has capabilities for multiplanar localization in a true midline sagittal plane and thus improves the identification of these abnormalities and increases interpreter confidence in the evaluation of normal facial profiles because the precise plane of imaging is known (7,8). Identification or exclusion of facial defects is important to both the family and clinician because facial anomalies may be markers of more complex abnormalities.

The archiving of volume data is valuable for enabling further data characterization. Saving 3D US data on storage media provides the opportunity to manipulate the data after the patient has left the clinic and to evaluate anatomic structures that were not initially scrutinized during the examination (1,9,10). The data may also be reviewed by consulting physicians off-site or online, trainees, or family members after the time of examination, with further manipulation of the data as necessary. The volume data can also be rendered by using different techniques or thresholds to highlight the anatomic area of interest or exclude adjacent structures.

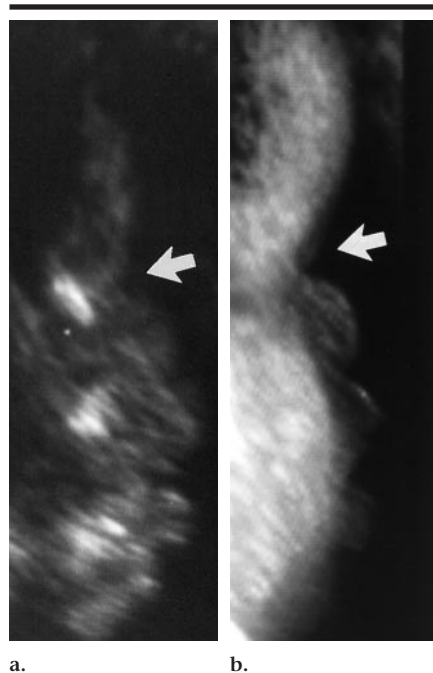
The 3D US technique is limited by the quality of the initial data acquisition for all subsequent manipulations. Any change in fetal position during the course of the examination necessitates the reacquisition of the data set to avoid motion degradation (1,11,12). For satisfactory surface-rendered images, the structure of interest must be surrounded by amniotic fluid. If adjacent placental structures or overlapping limbs cannot be excluded from the rendered volume, they may interfere with surface rendering. This be-



**Figure 6.** Case 6. Sagittal planar image of a fetus with camptomelic dysplasia at 21 weeks gestation shows flat facies (arrowheads). This abnormality was identified at 3D US but not at 2D US.

comes a major limitation of 3D US with oligohydramnios and as the fetus progresses toward term (1,7). It may not be as much of a problem with MIP images that depict the skeleton.

In conclusion, 3D US assisted in the appreciation of spatial orientation by means of rotation, surface-rendering techniques, and multiplanar displays. In three of seven fetuses, this examination provided additional information about the face and scapula in the evaluation of skeletal dysplasias, as compared with 2D US. Three-dimensional US imaging, by



**Figures 7.** Case 2. Midfacial hypoplasia in a fetus with thanatophoric dysplasia at 24 weeks gestation. (a) Multiplanar and (b) volume-rendered images (with combined surface- and light-rendering technique) of the facial profile show a concavity (arrow) in the midface that is consistent with midfacial hypoplasia, which was confirmed at delivery.

providing a global rather than planar view of the anatomy, enabled the identification of abnormal scapulae that were not appreciated with 2D US. As a result, 3D US improved confidence in the prenatal diagnosis of a specific skeletal dysplasia. Three-dimensional US imaging, by means of depth perception cues and rotation, also facilitated increased appreciation of positional limb abnormalities and allowed improved visualization of spine anomalies. Three-dimensional US images could be manipulated to highlight the suspected pathologic entity after the patient had left the clinic. Three-dimensional US, by demonstrating additional findings and increasing interpreter confi-

dence, provided additional information in the evaluation of suspected fetal skeletal dysplasias compared with 2D US, but it did not change management in this small preliminary study.

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