Three-dimensional Ultrasonographic Appearance of the Fetal Akinesia Deformation Sequence

Rodrigo Ruano, MD, Yves Dumez, MD, Marc Dommergues, MD

Objective. To present the appearance of the fetal akinesia deformation sequence by three-dimensional ultrasonography after four-dimensional ultrasonographic scanning. Methods. Three-dimensional surface-rendering images were used to show the fixed postural abnormalities of the fetal extremities and body. Four-dimensional ultrasonography was used to show that the postural abnormalities were fixed and to confirm the absence of fetal movements. These images were compared with the postmortem examination findings. Results. Three-dimensional ultrasonographic images of 3 cases of fetal akinesia deformation sequence are presented. Surface-rendering imaging and imaging in the skeletal mode were performed to show the postural abnormalities and the arthrogryposis. Conclusions. Facilitated by polyhydramnios, three- and four-dimensional ultrasonography provided clear images of the fixed postural abnormalities, which were confirmed by postmortem examination. Although prenatal diagnosis can be easily made by conventional ultrasonography, three-dimensional ultrasonography might be used as a confirmatory approach. In addition, the quality of recent three-dimensional imaging might help prenatal counseling by providing images more readily understandable to the parents than those obtained by two-dimensional ultrasonography. Key words: contractures; fetal akinesia deformation sequence; fetal movements; postural deformities; prenatal diagnosis; three-dimensional ultrasonography; ultrasonography.

Fetal akinesia consists of the abolition of movements as a result of myogenic, neurogenic, or skin disorders. Prenatal diagnosis is generally made by conventional two-dimensional ultrasonography (2DUS) in the second half of pregnancy, chiefly by showing the absence of fetal movements in association with abnormal positions of members. We present images from 3 fetuses with the akinesia sequence at 22, 24, and 25 weeks’ gestation in whom three-dimensional ultrasonography (3DUS) and four-dimensional ultrasonography (4DUS) were performed.
Materials and Methods

Three cases of suspected fetal akinesia were referred to our unit because of absence of fetal movements associated with polyhydramnios at 22, 24, and 20 weeks’ gestation, respectively. At referral, conventional ultrasonographic examinations revealed abolition of movements associated with fixed postural abnormalities of the limbs, confirming the diagnosis of the fetal akinesia deformation sequence. Polyhydramnios was also observed as a consequence of absence of fetal swallowing.

The patients consented to undergo 3DUS and 4DUS (Voluson 730; Kretztechnik AG, GE Medical Systems, Zipf, Austria; 4- and 8-MHz transducer) after being informed that the procedure would be undertaken for research purposes. Four-dimensional ultrasonography, which is 3DUS in real time, was performed to see fetal movements. The images were recorded and processed afterward. The patients elected to undergo termination of pregnancy. Three-dimensional ultrasonographic and 4DUS images were compared with postmortem findings.

Results

The first case is presented in Figures 1–5, the second in Figures 6 and 7, and the third in Figures 8 and 9.

Discussion

Here we report the 3DUS imaging of fetal akinesia. Three-dimensional ultrasonography provided clear images of abnormal positions of the fetal limbs and arms. Real-time 3DUS (4DUS) allowed us to show that these postural abnormalities were fixed, with no fetal movements. On the basis of informal conversations with patients, we think that the 4DUS images helped the couples in understanding these fixed postural abnormalities and thus becoming aware of the severity of the disorder.

The fetal akinesia sequence consists of a spectrum of disorders leading to a common phenotype: multiple joint contractures, limb pterygium, intrauterine growth restriction, pulmonary hypoplasia, and distinct craniofacial anomalies.5 In utero fetal movement is essential for development of normal limb function, normal facial musculature, and normal chest mechanisms necessary for pulmonary development.
So far, prenatal diagnosis has been based on conventional 2DUS, showing the absence of fetal movements associated with postural abnormalities of the extremities and polyhydramnios.\textsuperscript{1,2,4} According to Witters et al,\textsuperscript{6} the etiologic prenatal diagnosis is possible in up to 53\% of cases, mainly on the basis of a postmortem examination.

Although the prenatal diagnosis of the fetal aki-nnesia sequence can be correctly made by 2DUS, we think that 3DUS and 4DUS could help in confirming the diagnosis by providing clear, detailed images of the sequence. The absence of fetal movements and the fixed postural abnormalities are shown more clearly on 4DUS than on conventional 2DUS. These images, which illustrate the ever-increasing quality of 3DUS, may be helpful for parents in understanding the characteristics of the postural abnormalities, thus improving prenatal counseling.

Figure 2. Three-dimensional image of the fetal profile confirming permanent flexion of the left arm. Note that the muscles of the left arm and the thoracic wall appear to be hypoplastic.

Figure 3. Hyperflexion of the muscles of the fetal fore arms and hands. Incidentally, a uterine septa is shown on 3DUS (arrows).

Figure 4. Three-dimensional image showing deformation of the right hand.
Figure 5. A and B, Surface-rendered 3DUS images showing abnormal posturing of the fetal feet. C, Postmortem photograph showing the multiple joint contractures of the inferior extremities, with major postural deformities of both feet.

Figure 6. A, Four-dimensional image showing fixed fetal immobility in the second case. We observed hyperflexion of the arms and hands, with fixed hyperextension of the legs and feet. B, Postmortem photograph of the fetus after termination of pregnancy confirming the fixed abnormal position of the limbs. Note the hyperflexion of the upper extremities and the hyperextension of the lower extremities, which had been diagnosed by 3DUS and 4DUS findings.
Figure 7. A, Surface-rendered 3DUS image of the fetal hands after 4DUS scanning. Note the fixed contracture of the fingers and the hands. B, Anterior aspect of the fetal hands in the surface-rendering mode after 4DUS scanning. C, Skeletal mode 3DUS image showing deformities of the fetal hand bones, probably as consequence of arthrogryposis. D, Photograph of the fetus after termination of pregnancy showing the abnormal posture of the arms and hands.
Figure 8. A, Surface mode 3DUS image from the third case showing fixed abnormal postures of both fetal superior and inferior extremities. Also note the hyperextension of the fetal legs and feet. B, Skeletal mode 3DUS image showing the fixed postural abnormalities of the fetal extremities. C, Photograph of the fetus after termination of pregnancy confirming the abnormal fixed posture of the limbs.
References


Figure 9. A. Surface-rendered 3DUS image showing the rigid, fixed hyperextension of both fetal legs, with vicious positioning of the feet. Fixed posture of the second finger is also shown. Note the shape of the limbs, suggesting major muscular atrophy. B. Postmortem photograph confirming the fixed posture of the second finger.