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REVIEW ARTICLE

Early Detection of Fetal Abnormality

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ABSTRACT

After introduction of high-frequency vaginal transducer, transvaginal two-dimensional ultrasound has established a field of sonoembryology and most of the major fetal abnormalities have been detectable in the first trimester. Three-dimensional ultrasound adds an objective and comprehensive information to two-dimensional sonographic findings.

Keywords: Sonoembryology, High-frequency vaginal transducer, Transvaginal two-dimensional ultrasound, Three-dimensional ultrasound.

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INTRODUCTION

After introduction of high-frequency vaginal transducer, transvaginal two-dimensional (2D) ultrasound had established a field of sonoembryology[1] and most of major fetal abnormalities have been able to be detectable in the first trimester.[2] Three-dimensional (3D) ultrasound adds an objective and comprehensive information to 2D sonographic findings. Recent technology of 3D ultrasound has provided not only fetal surface imaging but also multiplanar image analyses of the embryonal and fetal structures. Rotation of embryo and close scrutiny of the volume allow the systematic review of anatomic structures, such as cord insertion, limb buds, cerebral cavities, stomach and bladder.[3] Appropriate fetal midsagittal section after rotating fetal image improved accuracy of nuchal translucency measurement.[4] Thus, 3D sonoembryology[3,5] has been established. Early diagnosis by 3D ultrasound, of amnionicity in twin pregnancy at 6 weeks,[6] and alobar holoprosencephaly at 9 weeks[7] were recently reported. Organ volume analysis is one of the most interesting topics in 3D technology. There are several articles on volume analysis in early pregnancy by using 3D ultrasound technology. Blaas et al reported the successful volume imaging and volumetry of embryonal brain cavity.[8,9] Kupesic et al published their article of volume assessment of the gestational sac and yolk sac.[10] In this chapter, we introduced sonograms of abnormal embryos/fetuses detected by transvaginal sonography.

ABNORMAL YOLK SAC

In normal pregnancy, yolk sac diameter showed an increase from 5 to 11 weeks of menstrual age, followed by a decrease and its disappearance after 12 weeks. Several reports described that abnormal size of yolk sac was significant in the subsequent spontaneous abortion.[11-14] In early pregnancy, when huge yolk sac is observed, embryonal demise may occur within several days. Figure 1 shows a huge yolk sac beside an embryo with normal heartbeat. Embryonal death was confirmed 1 week later and villous chromosome was trisomy 15. Embryo occasionally grows with huge yolk sac. Figure 2 shows normal appearance of

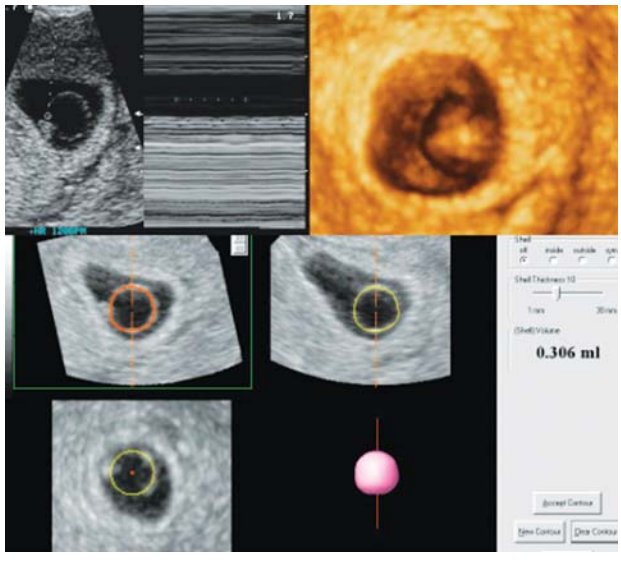

Fig. 1: Huge yolk sac: Upper—2D image with normal regular heartbeat and 3D image. Lower—Three orthogonal views with volume calculation 7 weeks and 3 days of menstrual age. Regular heartbeats of 120 bpm beside large yolk sac. Embryonal death was confirmed one week later. Villous chromosome was trisomy 15

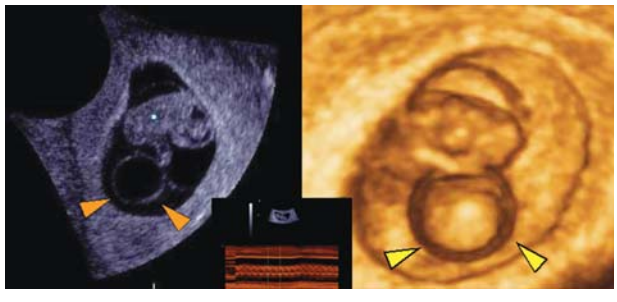

Fig. 2: Huge yolk sac: Left—2D image with normal appearance of early embryo and large yolk sac (arrowheads). Middle—Regular heartbeat of 174 bpm. Right—3D image of fetus and yolk sac (arrowheads). 8 weeks and 3 days of menstrual age. Fetal demise was confirmed on the next day. Villous chromosome was 48, XY, +15,+21, double trisomy

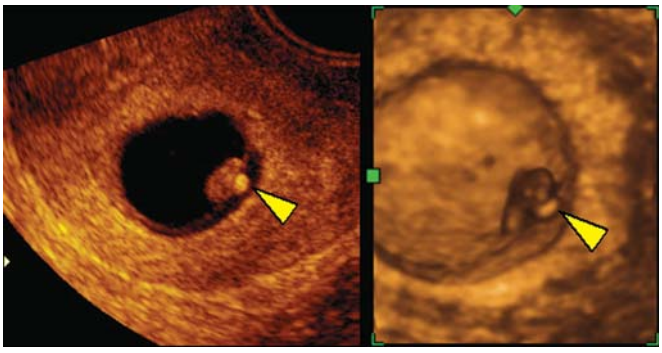

Fig. 3: Hyperechoic yolk sac: Left—2D image, right—3D image. Nine weeks and 1 day of menstrual age. Normal heartbeat was seen. Embryonal death was confirmed 3 days later. Villous chromosome was trisomy 15

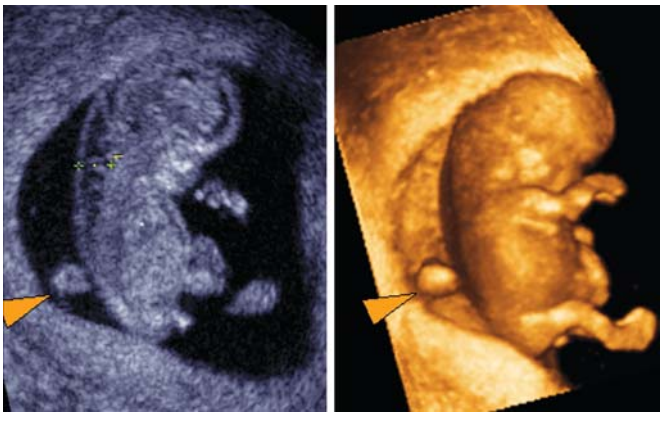

Fig. 4: Hyperechoic yolk sac: Left—2D image, right—3D image. 11 weeks and 3 days of gestation. Referral due to nuchal translucency. Normal heartbeat was seen. Amniocentesis resulted in trisomy 18

early embryo with regular heartbeat of 174 bpm beside a large yolk sac at 8 weeks and 3 days of menstrual age. Fetal demise was confirmed on the next day and villous chromosome was 48, XY, +15,+21, and double trisomy. Hyperechogenic yolk sac is also highly associated with chromosomal aneuploidy. Figures 3 and 4 show an embryo/fetus with normal heartbeat beside a hyperechoic yolk sac. Abnormal karyotypes of trisomy were confirmed in both cases. Thus, careful observation of yolk sac size and appearance is necessary in early pregnancy.

FETAL STRUCTURAL ANOMALY

Recently, most of fetal structural anomalies are detectable in the first and early second trimesters by transvaginal sonography. Transvaginal ultrasound scan between 12 and 15 weeks of gestation provides us many of fetal information *in utero*. Facial abnormalities including lowset ears, micrognathia and exophthalmos can be detected (Figs 5 and 6). Fetal limb abnormalities, difficult to be demonstrated by transabdominal sonography in the first half of pregnancy, can be clearly detected by transvaginal approach. Figure 7 shows severe clubfeet in a case of spina bifida, myelo-

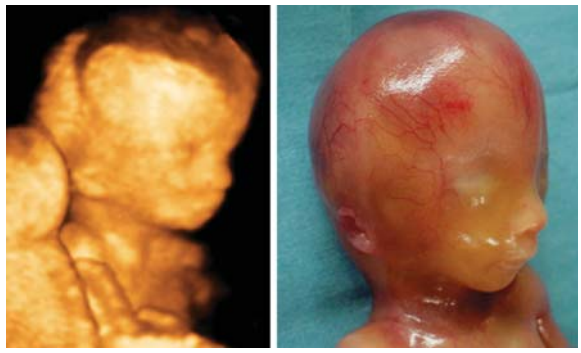

Fig. 5: Lowset ear and micrognathia at 15 weeks of gestation. Left: 3D image. Right: Face of aborted fetus. Growth retardation, single umbilical artery and additional anomalies were observed. Fetal karyotype was trisomy 18

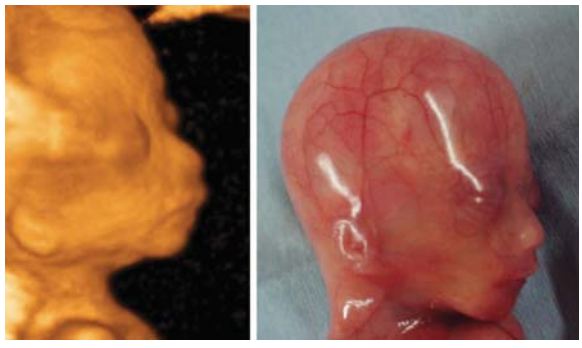

Fig. 6: Facial anomaly observed in a case of holoprosencephaly at 15 weeks of gestation. Left: 3D image. Right: Face of aborted fetus. Lowset ear and exophthalmos are demonstrated. Fetal karyotype was partial delation of chromosome 9

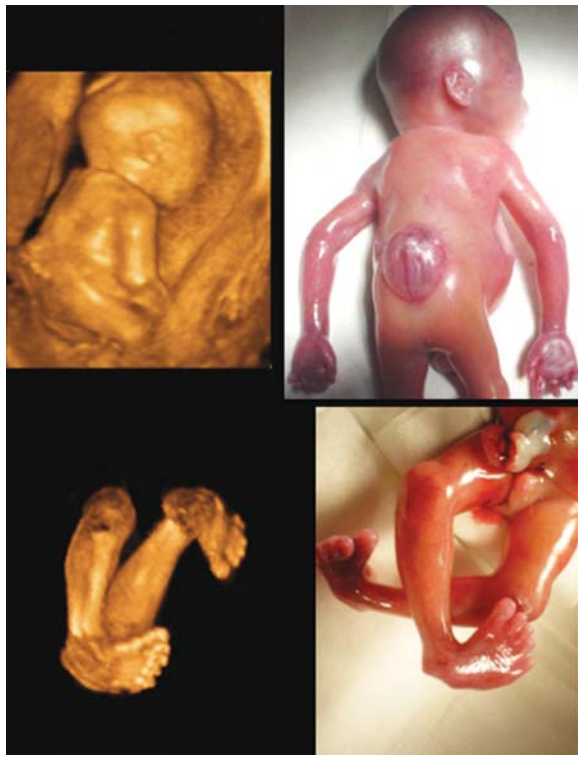

Fig. 7: Fetus with myelomeningocele, severe kyphosis and clubfoot at 20 weeks of gestation. Upper left: 3D image of fetus (transabdominal sonography). Upper right: Aborted fetus. Lower left: 3D image of fetal legs (transvaginal sonography), severe clubfeet are seen. Lower right: Legs of aborted fetus. Fetal karyotype was normal

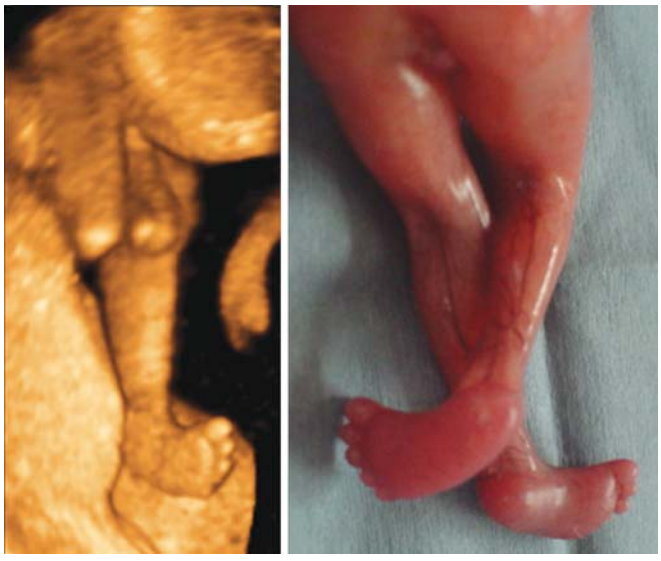

Fig. 8: Fetal clubfoot at 15 weeks of gestation. Left: 3D image of fetal leg. Right: Legs of aborted fetus. This fetus has chromosomal aberration

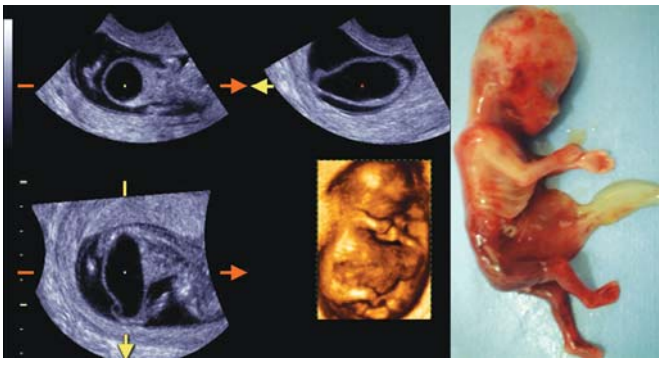

Fig. 9: Prune-belly syndrome at 11 weeks of gestation. Three orthogonal views and 3D surface image. Enlarged Prune-belly-like bladder is detected. Right photo shows aborted fetus at 12 weeks of gestation. Fetal karyotype was normal

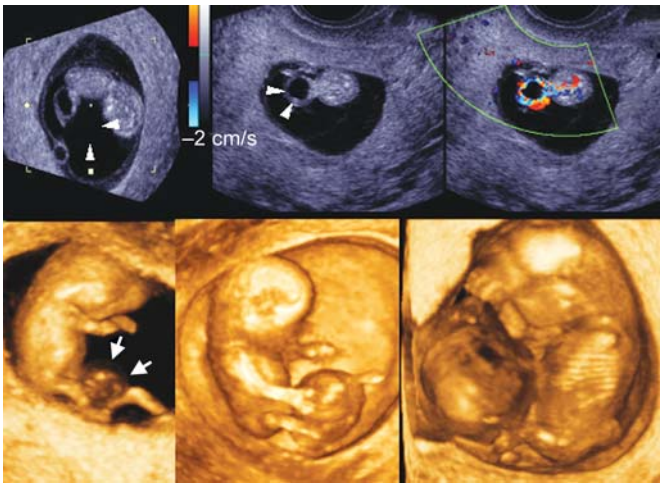

Fig. 10: Exstrophy of the bladder in the first trimester. Upper: 2D sagittal (left), coronal (middle) and coronal color Doppler image (right). Extracorporeal cyst (arrowheads) is seen. Note the umbilical arteries running along the cyst. Lower: 3D images at 10 weeks (left), 11 weeks (middle) and 12 weeks (right) of gestation. Longitudinal images show gradual but rapid increase of the outside cyst. No bladder was seen inside of the fetus

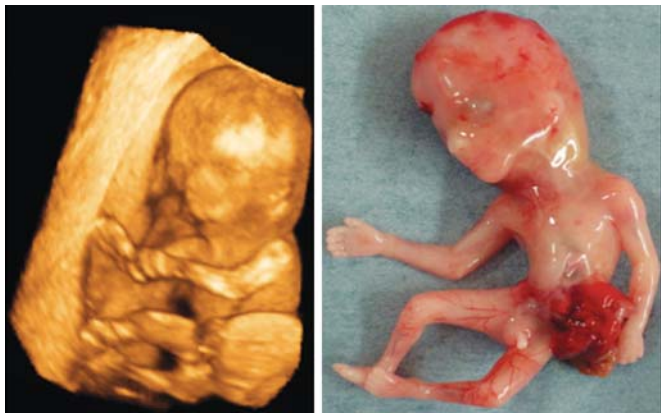

Fig. 11: Omphalocele at 12 weeks of gestation. Left: 3D surface image of the fetus. Omphalocele was seen at the left side of the fetus. Right: Aborted fetus. Sac of omphalocele was ruptured on delivery. Fetal chromosome was normal

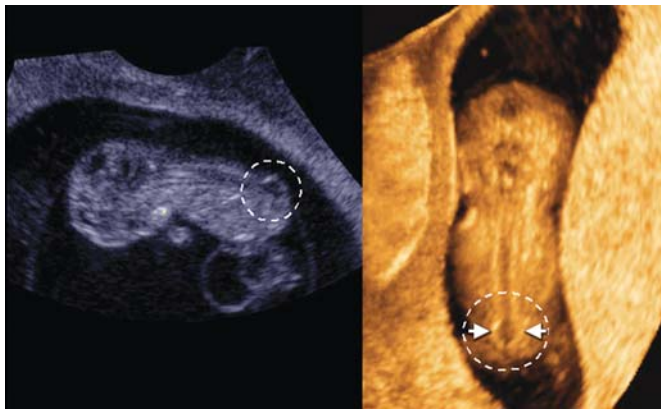

Fig. 12: Spina bifida at 9 weeks of gestation. Left: 2D sagittal image. Cystic formation was seen (white circle) at lumbar part. Right: 3D image of neural tube. Clear dilatation of the neural tube is demonstrated (arrows)

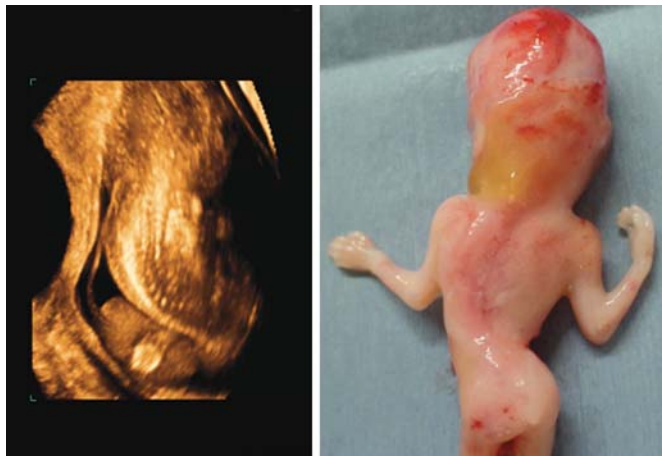

Fig. 13: Severe scoliosis at 12 weeks of gestation. Left: 3D image of fetal back. Severe scoliosis is demonstrated. Right: Back view of aborted fetus. Fetal karyotype was normal

meningocele and severe kyphosis. Figure 8 shows fetal clubfoot at 15 weeks in a case with chromosomal aberration. Fetal abdominal abnormalities such as prune-belly syndrome (Fig. 9), exstrophy of the bladder (Fig. 10) and omphalocele (Fig. 11) are easily detectable in the first trimester. Spina

bifida includes several types of myelomeningocele, myelocystocele, meningocele and myeloschisis.

Although, Blaas et al reported cases of spina bifida before 10 weeks of gestation, spinal level and degree of spina bifida are variable and its early detection is quite difficult.[15] Figure 12 shows early detection of spina bifida at nine gestational weeks. At 9 weeks, vertebral bony structure is not detectable, but neural tube dilatation is clearly demonstrated in the case. Vertebral scoliosis is detectable from early pregnancy (Fig. 13). Pleural effusion is often found in early pregnancy because of congenital heart diseases and/or chromosomal aberration. In most of cases with early detection of pleural effusion, subsequent fetal demise may be confirmed. In our series, over 90% of cases with early pleural effusion was associated with 45, X, Turner's syndrome (Fig. 14). Fetal hydrops in the first trimester is also strongly associated with abnormal karyotype. Figures 15 and 16 show hydropic fetus with pleural effusion or cystic hygroma. In many of cases with cystic hygroma, congenital heart disease, tachycardia and general edema, fetal chromosome test may result in abnormal karyotype, especially 45, X. Figure 17 shows hydropic fetus with cystic hygroma, axillary lymphadenopathy at 15 weeks of gestation. Congenital heart disease of double outlet right ventricle (DORV) was simultaneously detected. Although Turner's syndrome was suspected, but this case had abnormal wrist flexion and contracture shown on the right Figure. Most cases of 45, X have no upper limb abnormalities. Fetal demise occurred 3 days later and chromosome was trisomy 18. Congenital diaphragmatic diseases including diaphragmatic hernia and eventration of diaphragm highly cause subsequent hypoplastic lung. Early detection was quite difficult. We had a case with agenesis

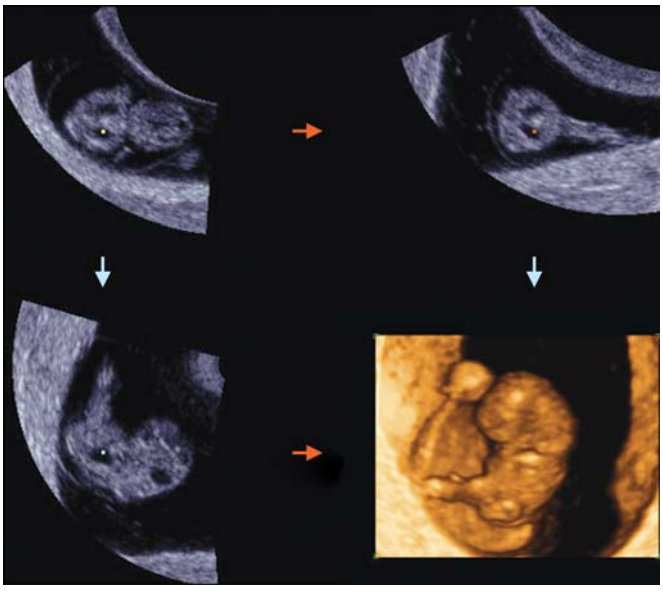

Fig. 14: Pleural effusion at 9 weeks of gestation: Three orthogonal views and 3D surface image. Bilateral pleural effusion is clearly demonstrated. Fetal heartbeat was normal and regular. Fetal demise was confirmed 3 days later and villous karyotype was 45, X, Turner's syndrome

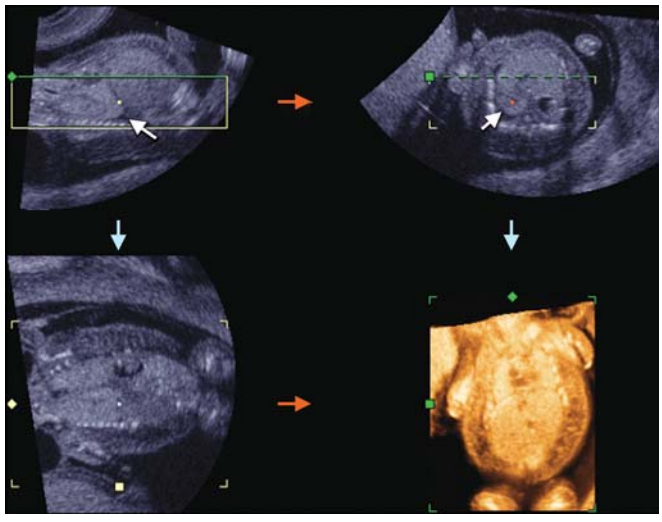

Fig. 15: Hydropic fetus at 14 weeks of gestation: Three orthogonal views and 3D surface image. Severe general edema and unilateral pleural effusion (arrows) are demonstrated. Fetal karyotype was 45, X

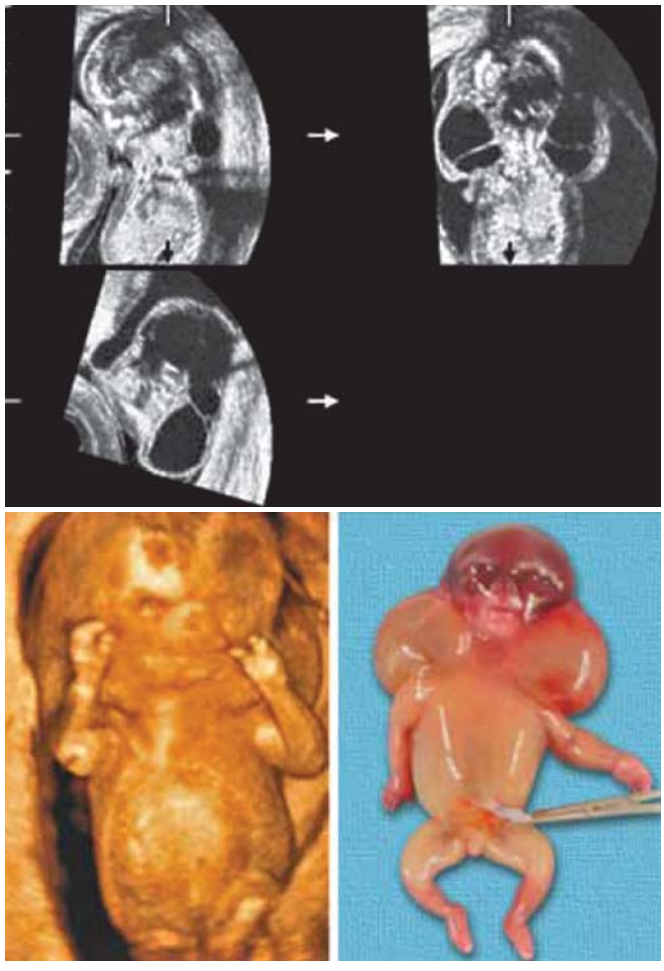

Fig. 16: Cystic hygroma at 14 weeks of gestation: Three orthogonal views (upper), 3D surface image (lower left) and aborted fetus (lower right). Fetal karyotype was 45, X

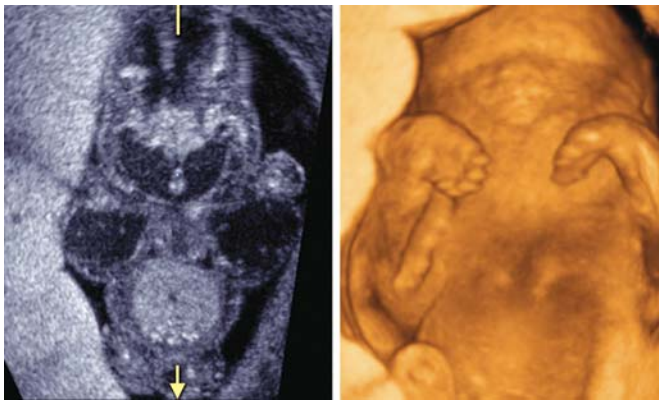

Fig. 17: Hydrops, cystic hygroma, axillary lymphadenopathy and abnormal wrist flexion and contracture at 15 weeks of gestation. 2D coronal image (left) and 3D surface image (right). Bilateral pleural effusion is also demonstrated on the 2D image. In this case, congenital heart disease of DORV was detected. Fetal karyotype was trisomy 18

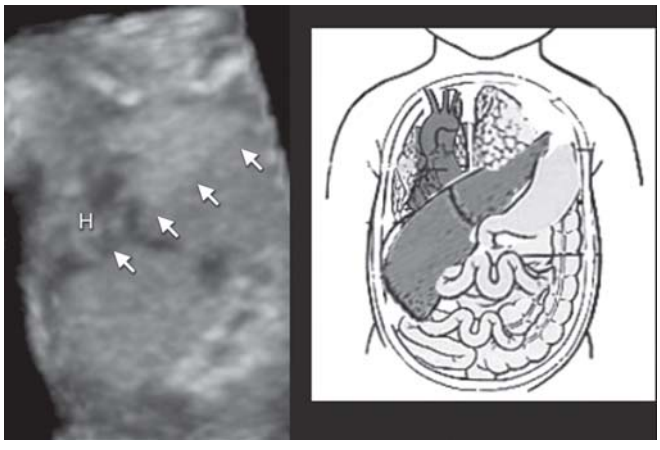

Fig. 18: Congenital diaphragm defect at 13 weeks of gestation: Left: A cutting image of 4D cardiac mode showing fetal thoracoabdominal part. Dextrocardia (H) and abnormal lung-liver border is demonstrated (arrows). Left lung was visible at this stage, but became completely hypoplastic lung in late pregnancy. Right is schematic figure of the left sonographic image. Fetal karyotype was normal

of diaphragm, which was strongly suspected from early pregnancy. Figure 18 shows dextrocardia and abnormal lung-liver border at 13 weeks of gestation. Left lung was visible at this stage, but became completely hypoplastic lung in late pregnancy.

FUTURE ASPECTS

Transvaginal sonography and 3D ultrasound had contributed to reveal pathophysiological natural history of congenital structural abnormalities. Recent advanced technology of 4D ultrasound has been adding *in utero* information of real-time fetal motion and positioning and 4D echocardiography. With technological development of ultrasound, further detailed detection in early pregnancy is expected.

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