Importance of first trimester scan – 4 of 4

Screening for early fetal structural anomalies

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Introduction

Improvement of high resolution ultrasound scan equipment, have enabled obstetricians to examine the fetus during first trimester in more detail than ever before. It is established that first trimester ultrasound scan is superior to second trimester scan in determining gestational age, risk assessment for chromosomal aneuploidy and chorionicity in multiple pregnancy[1-3]. Moreover, first trimester scan can also be used in screening fetal structural anomalies. It is important that the operator needs to be familiar with the ultrasonograpic appearance of fetal structures in first trimester when evaluating fetal anatomy (Table 1). Transverse sweep across the fetal body from the crown to rump enables to visualise all the fetal structures (Figure 1). It has been apparent that most of fetal structural problems can be reliably screened during first trimester[4-9] (Table 2). However, some cardiac anomalies cannot be confirmed during first trimester.

Table 1. Routine first trimester screening for structural anomalies

Structure	Anatomical landmarks
Skull and brain	Transverse section of the head to demonstrate the skull, midline echo and the choroid plexuses filling the large lateral ventricles
Face	Examination of the profile, orbits and upper lip
Spine	Examination longitudinally to demonstrate the vertebral bodies and overlying skin
Heart	Examination of four-chamber view
Thorax	Visualization of the shape of the thorax, lungs and diaphragm
Abdomen	Demonstration of stomach, bladder, normal insertion of the umbilical cord into the abdomen
Limbs	Visualization of all the long bones, hands and feet (including shape and echogenicity of long bones and movement of joints)

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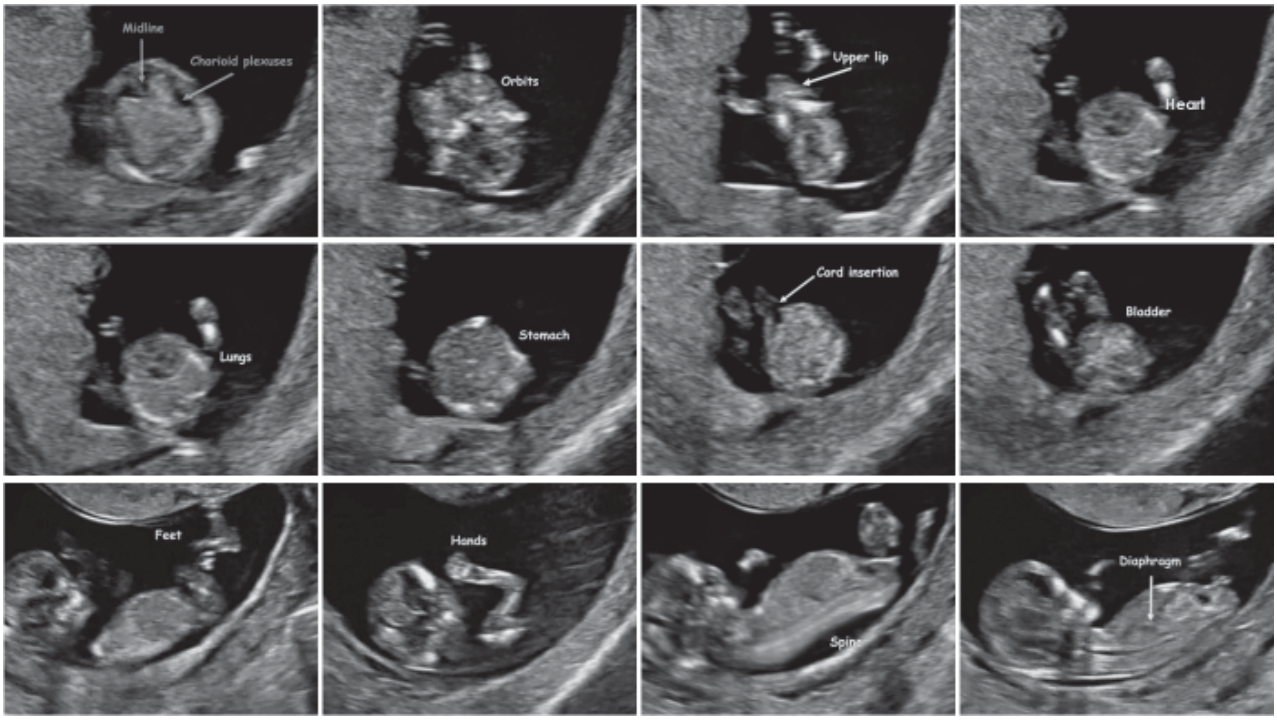

Figure 1. **Normal anatomy during first trimester.**
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Table 2. Summary of studies examining sensitivity of first-trimester ultrasound examination in detection of major fetal structural defects

Reference	*Population*	*Method of scan*	*n*	*Cases with major structural anomalies (n (%))*	*Ultrasound sensitivity (%)*
Economides and Braithwaite[4] (1998)	Low risk	TA/TVS	1632	13 (0.8)	54
Carvalho *et al.*[5] 2002	Low risk	TA/TVS	2853	66 (2.3)	28
Taipale *et al.*[6] 2004	Low risk	TA/TVS	4513	33 (0.7)	18
Chen *et al.*[7] 2004	High risk	TA/TVS	1609	26 (1.6)	54
Souka *et al.*[8] 2006	Low risk	TA/TVS	1148	14 (1.2)	50
Ebrashy *et al.*[9] 2010	Low risk	TA/TVS	2876	31 (1.1)	68

Central nervous system anomaly (CNS):

Acrania, exencephaly and anencephaly sequence

The diagnosis of anencephaly in second trimester is based on the absence of the cranial vault and cerebral hemispheres. However, the brain may appear normal during first trimester in case of cranial vault defect. In normal fetuses, the skull appears to be hyperechogenic compared to underlying tissues by 11 weeks' gestation. In the first trimester the main feature of vault defect is acrania. Without a cranium there is progressive degeneration of the exposed cerebral tissue such that exencephaly eventually progresses to anencephaly (Figure 2).

Many other CNS malformations can also be detected in early pregnancy including encephalocele, Dandy-Walker malformation, ventriculomegaly, and holoprosencephaly. An encephalocele is a cranial defect with protrusion of brain tissue and is often associated with microencephaly, spina bifida, ventriculomegaly, and Meckel-Gruber syndrome[10] (Figure 3 & 4). Dandy-Walker malformation is characterized by cystic dilatation of the fourth ventricle and complete or partial absence of the cerebellar vermis and it may be associated with genetic syndromes including Walker-Warburg syndrome, chromosomal abnormalities. Incomplete cleavage of the primitive prosencephalon or forebrain results in holoprosencephaly. Alobar holoprosencephaly is the most severe form and it is incompatible with life. Holoprosencephaly is associated with trisomy 13[9].

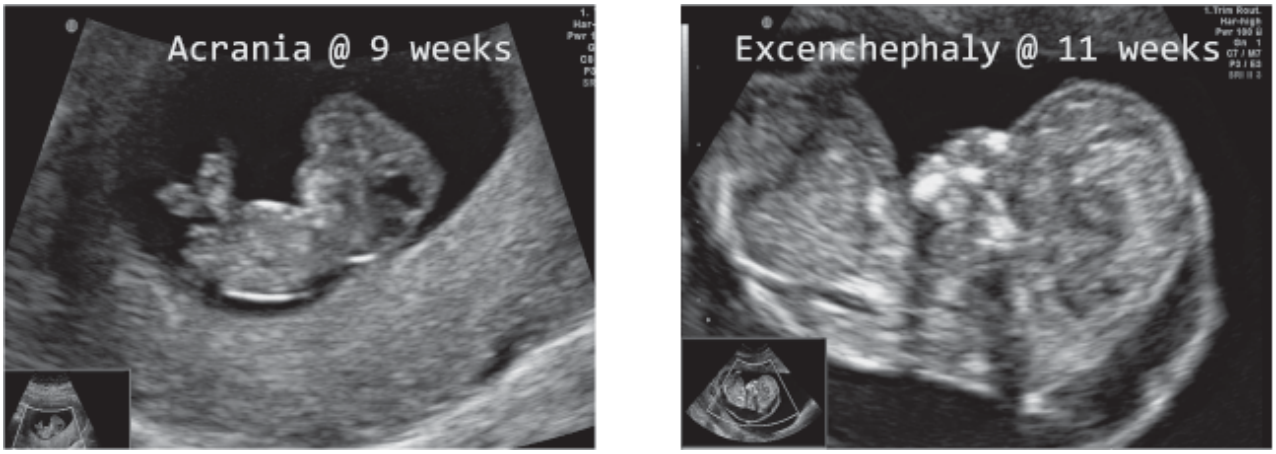

Figure 2. **Ultrasound image of the fetus showing acrania and excenchephaly.**

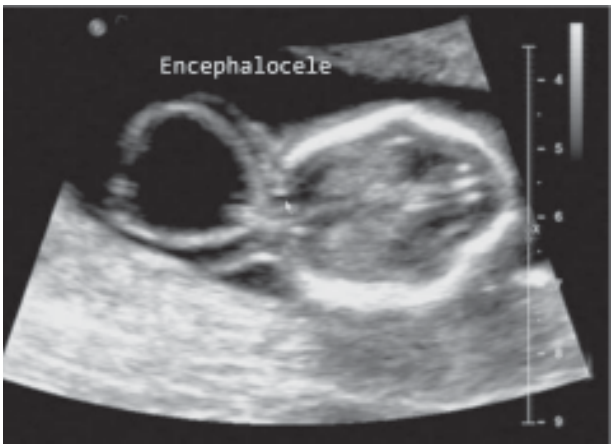

Figure 3. **A 13-week fetus with encephalocele.**

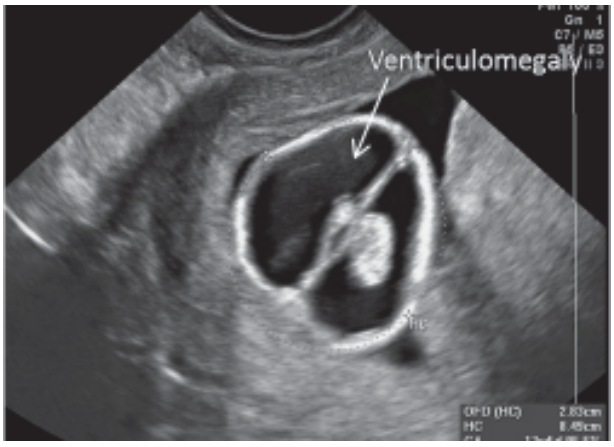

Figure 4. **A 13-week fetus with ventriculomegaly.**

Spina bifida

In almost all cases of open spina bifida there is an associated Arnold-Chiari malformation, which is thought to be the consequence of leakage of cerebrospinal fluid into the amniotic cavity and hypotension in the subarachnoid spaces leading to caudal displacement of the brain and obstructive ventriculomegaly. Manifestations of Arnold-Chiari malformation can be sonographically detectable during second trimester as the lemon and banana signs, which are visible in the transverse section of the brain used for measurement of the head circumference (HC) and assessment of cerebellum and cisterna magna. However, during first trimester the diagnosis of spina bifida is often missed. Sebire et al (1997) reported that none of the 29 fetuses with spina bifida out of 61972 pregnancies was detected during first trimester dating/ nuchal translucency scan[11].

A novel method of first trimester screening for spina bifida has been introduced recently[12]. In the same mid-sagittal view of the fetal face as used for measurement of nuchal translucency (NT) the brain stem and fourth cerebral ventricle are also easily visible. The fourth ventricle presents as an intracranial translucency (IT) parallel to the NT (Figure 5). Intracranial translucency is diminished with the caudal displacement of the brain in the case of open spina bifida.

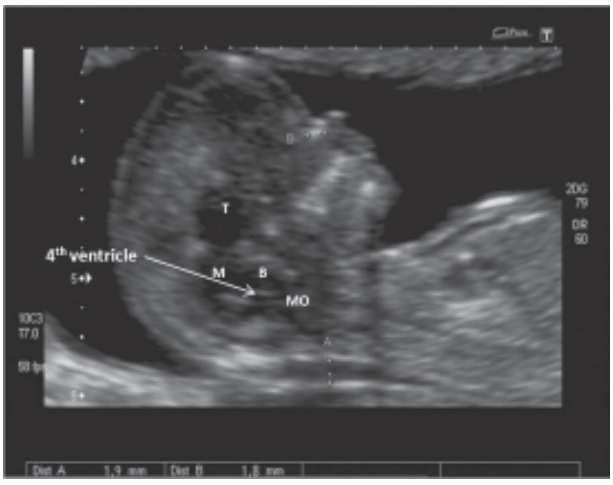

Figure 5. **Ultrasound image in the mid-sagittal plane of the fetal face showing the nasal bone, palate, mandible, nuchal translucency (NT), thalamus (T), midbrain (M), brain stem (B) and medulla oblongata (MO). The fourth ventricle presents as an intracranial translucency (IT) between the brain stem and the choroid plexus.**

Renal tract disorders

The fetal kidneys and adrenal glands can first be visualized by transabdominal ultrasound in all cases by 12 weeks' gestation. A number of pathologic genitourinary conditions have been diagnosed using ultrasound in early pregnancy including bilateral renal agenesis, hydronephrosis, multicystic dysplastic kidney disease, and megacystis.

Fetal megacystis at 10-14 weeks of gestation, defined by a longitudinal bladder diameter of 7 mm or more, is found in about 1 in 1500 pregnancies and there is a risk of about 25% that the fetus will have a chromosomal defect, mainly trisomy 13 or 18 (Figure 6). However, the parents can be reassured that in about 90% of cases with karyotypically normal megacystis <15 mm will resolve without any obvious adverse effects on renal development and function. In contrast, megacystis >15 mm is less associated with chromosomal aneuploidy but severe obstructive uropathy and renal dysplasia will eventually lead to renal failure[13].

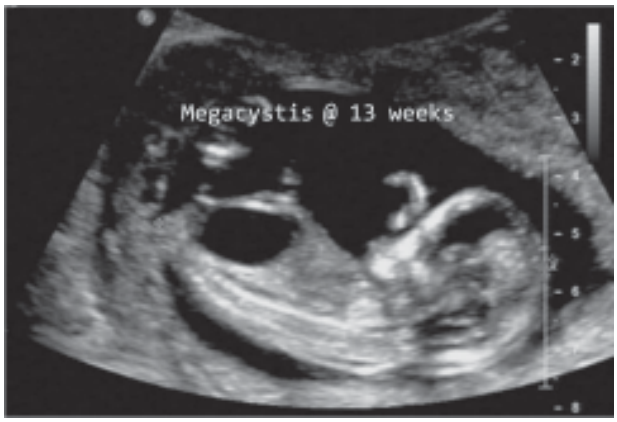

Figure 6: A 13-week fetus with megacystis.

Abdominal wall defects

The physiological herniation and the reduction of the mid-gut hernia completes by 12 weeks' gestation. If the herniation is noted after 12 weeks, an omphalocele should be suspected (Figure 7). Lateral anterior abdominal wall defect with bowel herniation remote from the cord insertion is seen in gastroschisis. Omphalocele at 11-13 weeks of gestation could be a feature in 26% of the fetuses with trisomy 18 and in 28% of the fetuses with trisomy 13[13].

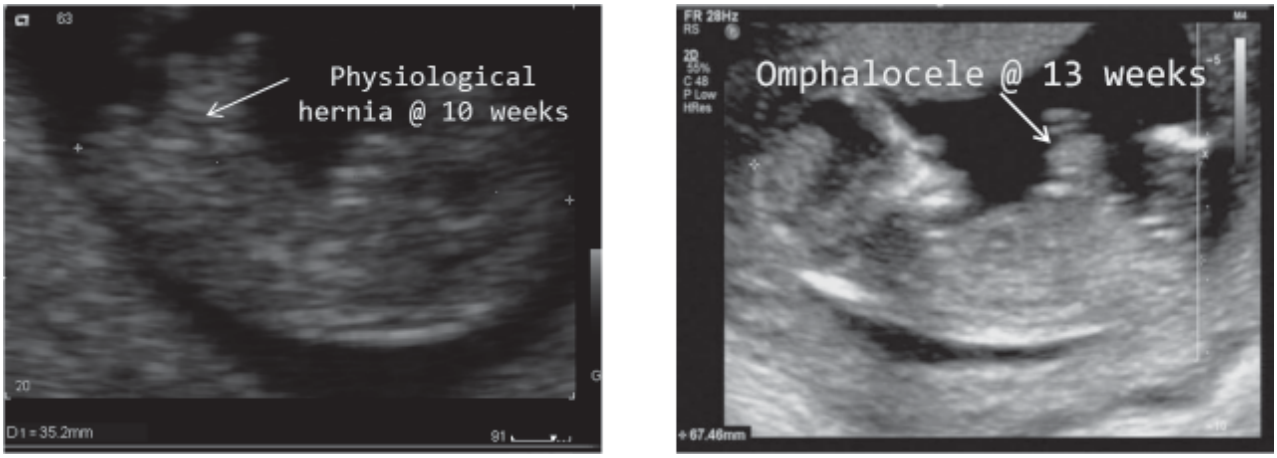

Figure 7. **Ultrasound image in the mid-sagittal plane of the fetus showing physiological herniation of mid-gut at 10 weeks and omphalocele at 13 weeks.**

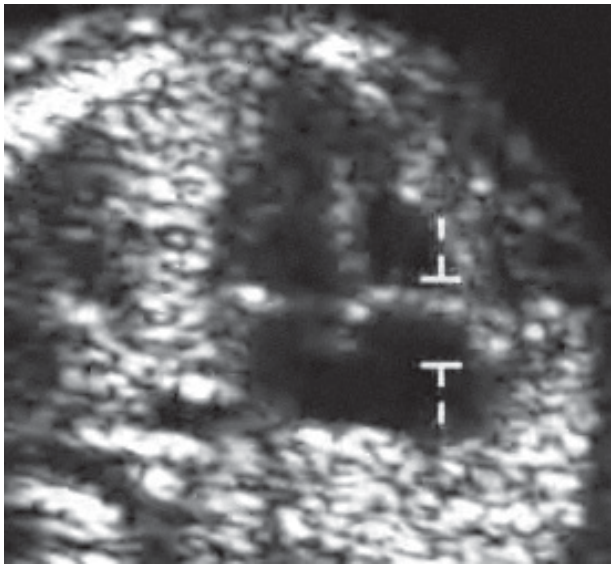

Figure 8. **Four chamber view of the heart during first trimester**
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Cardiac defects

A fetal echocardiography at 18 to 22 weeks for high risk groups is well established as being sensitive and specific for most cardiac abnormalities. The realization of the association between increased nuchal translucency and structural heart abnormalities has led to the identification of an important high-risk group at 11 to 14 weeks. A four-chamber view of the heart can be obtained as early as 11 weeks (Figure 8). However, the heart and associated vessels are so small in the first trimester and alterations in cardiac chamber size may not be apparent until later in gestation (16/40). Most major structural heart defects can accurately be diagnosed from the late first trimester of pregnancy and many families at-risk can be reassured of `normality' of cardiac connections at an early stage[14]. Some lesions may evolve throughout pregnancy and therefore early scans should not replace mid trimester fetal echocardiography.

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