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The value of screening for major fetal abnormalities during the nuchal translucency examination

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Abstract

The nuchal translucency (NT) scan provides an opportunity to examine fetal anatomy. Current opinion on the advantages and limitations of assessing fetal anatomy at this early gestation is divided. Two case studies from our centre will be presented where assessing fetal anatomy was of great benefit – one in finding abnormalities, the other in excluding them.

These cases along with review of the literature support the view that a limited fetal anatomy scan should be performed as part of the NT examination.

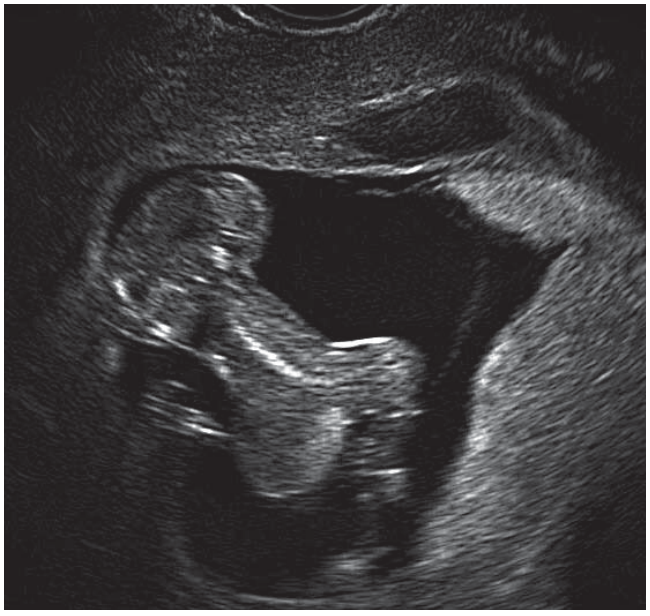

Fig. 1: Acrania and kyphoscoliosis.

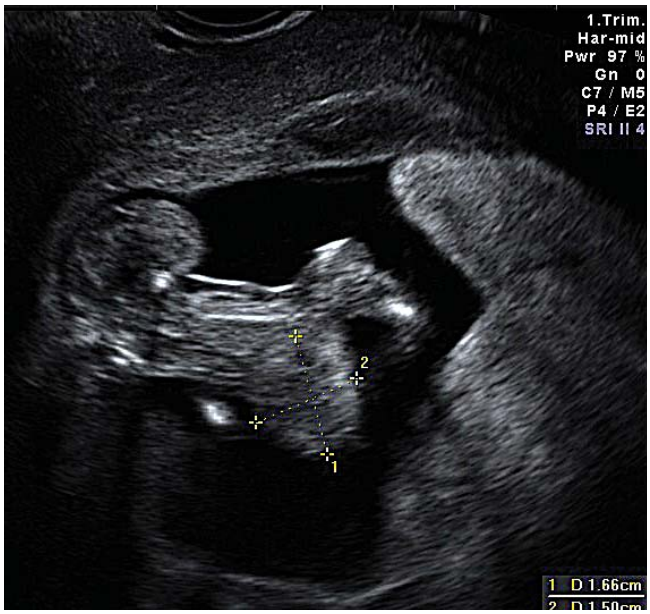

Fig. 2: Omphalocele.

Introduction

The nuchal translucency (NT) ultrasound measurement in combination with maternal age and biochemistry is well established as an effective method of assessing the risk for chromosomal abnormalities at 11–13+6 weeks gestation[1]. Ultrasound also provides the opportunity to screen for major structural abnormalities, however, current opinion is divided on whether fetal anatomy should be routinely assessed as part of this exam.

We present two recent cases from our centre where assessing fetal anatomy at the NT scan was of great benefit – one in finding abnormalities, the other in excluding them.

Review of the literature will discuss:

- whether fetal anatomy can be consistently visualised at this gestation
- the sensitivity of ultrasound in detecting abnormalities at the NT scan
- the advantages of identifying abnormalities in the first trimester
- the limitations involved.

Case 1

A 43-year-old G3 P2 presented for routine NT screening at 12w 6d gestation. Scanning was performed with a GE Voluson 730 Expert (Menio Park, New Jersey, USA). A live fetus was seen with a CRL, which was less than expected at 46.2 mm (11w 2d). The NT measurement was 1.8 mm. Assessment of fetal anatomy by transabdominal (TA) and transvaginal (TV) imaging revealed multiple abnormalities:

Acrania and kyphoscoliosis (Fig. 1); ectopia cordis with the heart being located within a large (1.66 x 1.50 cm) omphalocele (Figs. 2, 3); and abnormally positioned feet – possibly rockerbottom.

The fetal stomach and bladder could not be identified, amniotic fluid volume was normal and the arms and hands appeared normal.

A diagnosis of multisystem abnormalities which were incompatible with life was made.

Real-time visualisation and 3D images (Fig. 4) aided in counselling the patient and her husband and helped them to accept the severity of the findings. The pregnancy was

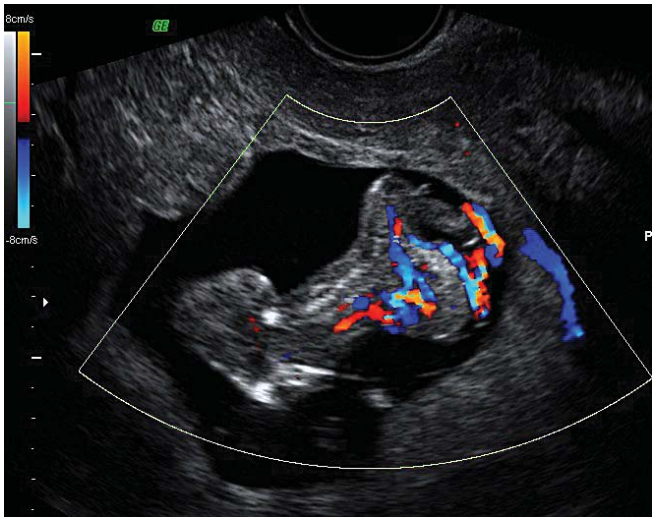

Fig. 3: Ectopia cordis: heart within omphalocele.

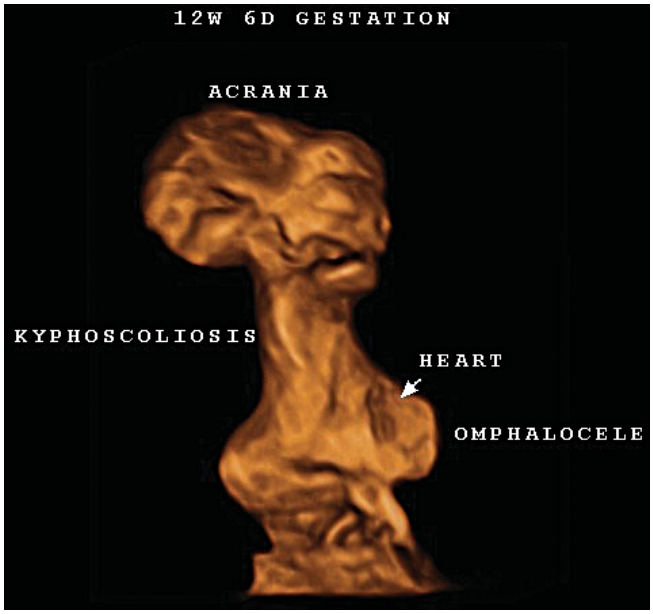

Fig. 4: 3D image of multisystem abnormalities.

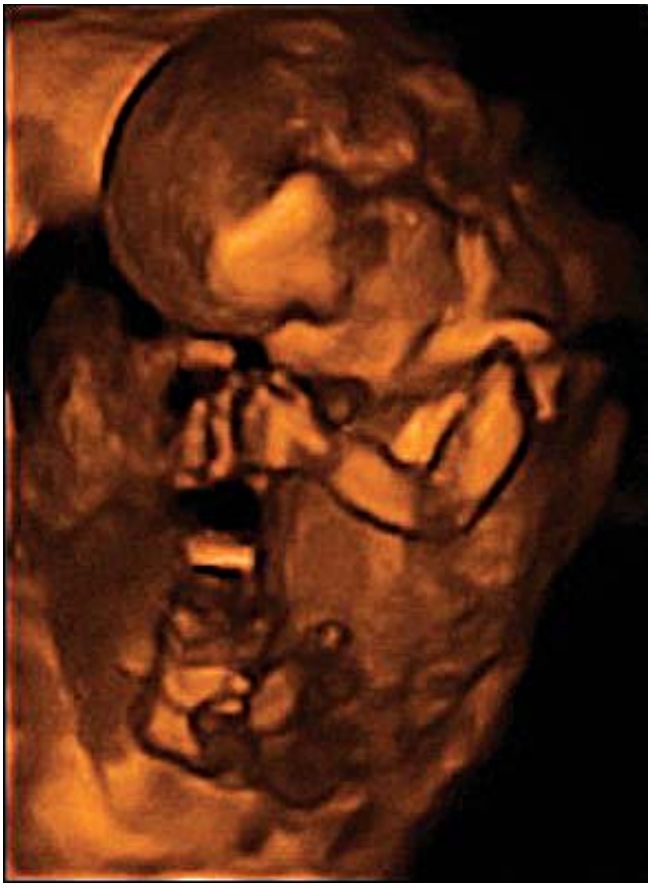

Fig. 5: – 3D image.

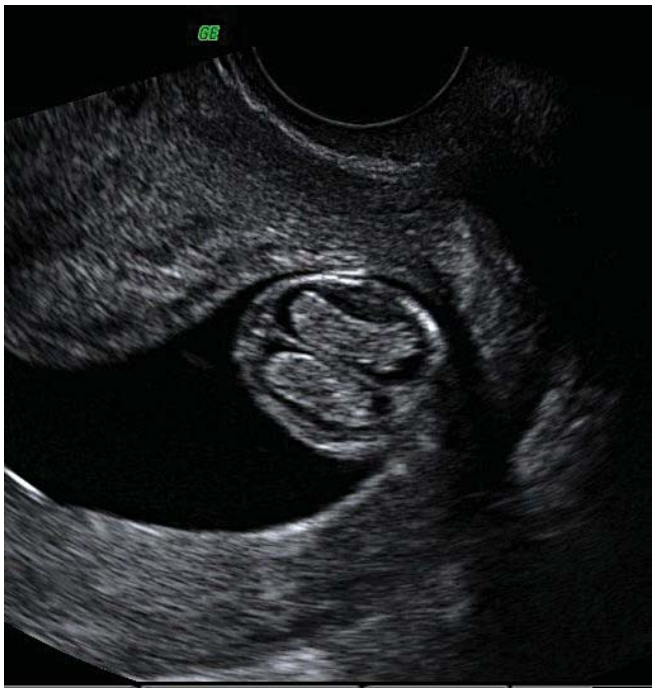

Fig. 6: Skull and brain.

terminated one week later without the need for further invasive prenatal testing.

Postnatal chromosomal karyotype was Trisomy 18 female.

Case 2

A 34-year-old G2 P0 presented for her NT ultrasound at 11w 6d. She was visibly anxious and was under the impression that the scan would tell her if "everything was alright" with the baby. A medical history was obtained which revealed that she suffered from bipolar disorder and had been taking lithium during the first five weeks of the pregnancy. She had been advised by her doctor of the increased risk of the fetus having a heart abnormality.

Lithium exposure has been associated with an increased incidence of congenital defects, particularly Ebstein's anomaly[2]. In this condition, the tricuspid valve has dysplastic leaflets which do not attach normally to the annulus, and the valve orifice is displaced downwards towards the apex of the right ventricle. On ultrasound, this is best demonstrated by the four-chamber view of the fetal heart[3].

The routine NT measurement was obtained (1.4 mm) and biochemical screening performed. In addition evaluation of fetal anatomy based on the Fetal Maternal Foundation (FMF) Nuchal Translucency software program was performed on the GE Voluson 730 Expert. Due to a combination of gestational age and maternal weight (89 kg), both TA and TV scans were required to obtain the necessary views. The scan demonstrated normal fetal anatomy (Figs. 5–13)

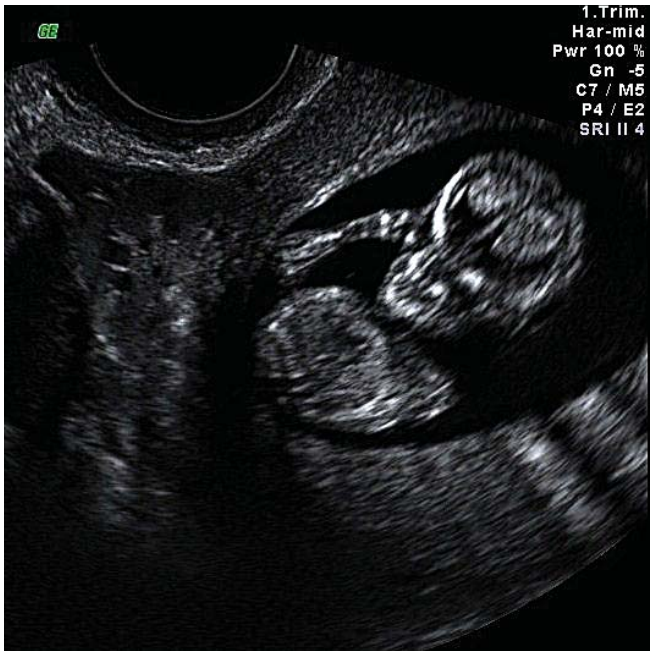

Fig. 7: Face.

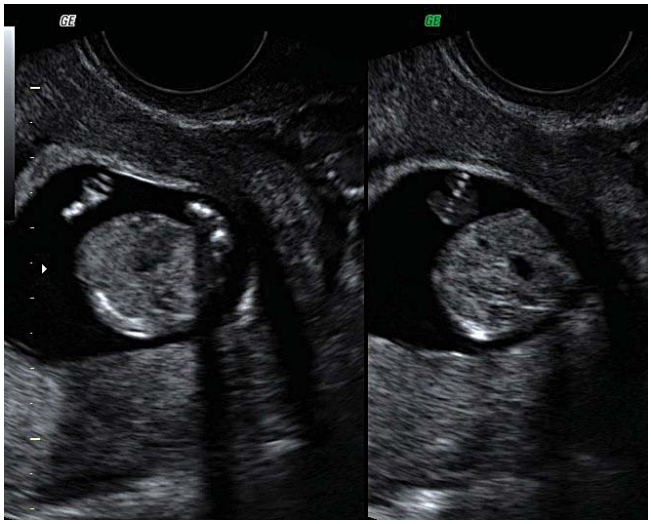

Fig. 8: Heart and stomach.

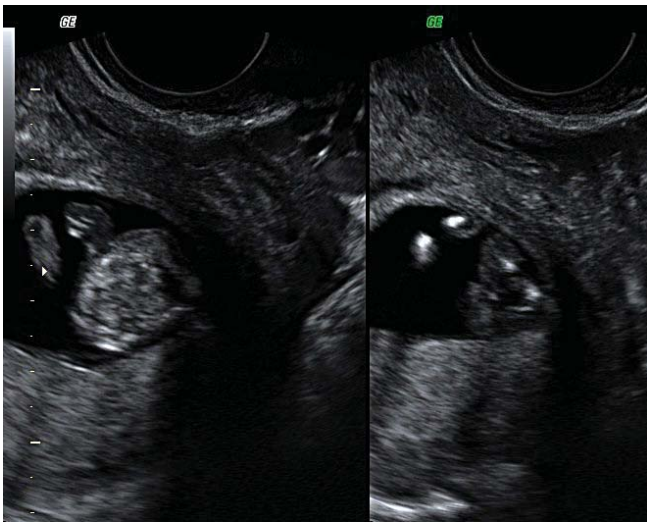

Fig. 9: Cord insertion and bladder.

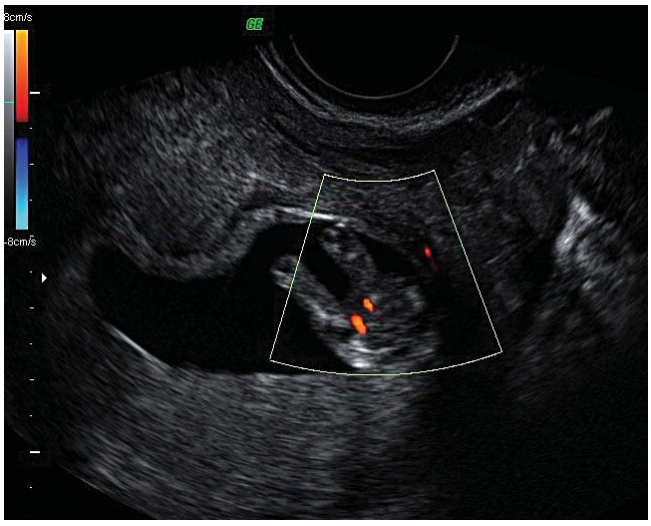

Fig. 10: 3 vessel cord.

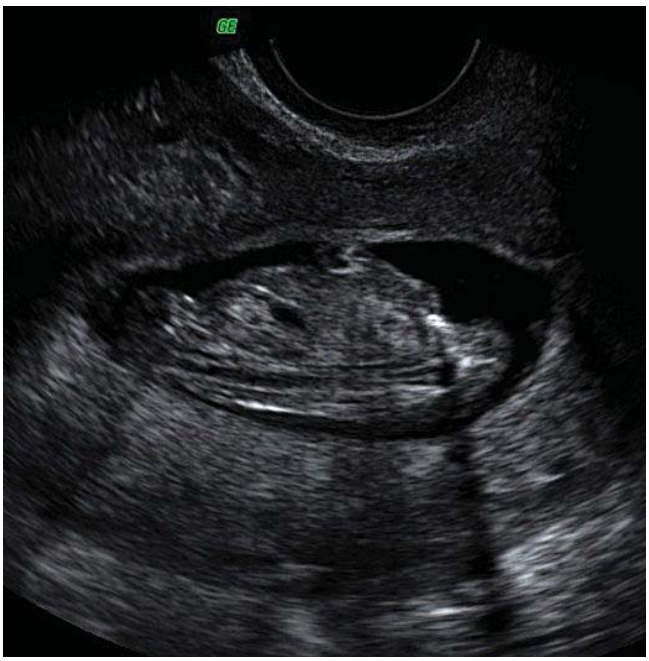

Fig. 11: Spine.

and a normal four-chamber heart (Fig. 14). The NT screening result was low risk for Down syndrome.

Screening of fetal anatomy was of great benefit to this patient. Although it was stressed that further scans would be required during the pregnancy, the normal result significantly reduced her anxiety about the pregnancy and resuming her medication, which she did at 16 weeks gestation. Subsequent scans of fetal anatomy, fetal echocardiography (Fig. 15) and fetal growth demonstrated no abnormalities.

A live female infant was born at 39w 6d by caesarean section. She was discharged with her mother two days later with no neonatal complications.

Discussion

Currently, there is no consensus about whether fetal anatomy should be routinely assessed as part of the NT exam. Although a checklist of fetal anatomy is included in the FMF form, ASUM does not specify to what degree fetal anatomy should be examined and reported in the first trimester[4]. This leaves the decision up to individuals and/or their workplace.

Studies show that examination of non-cardiac anatomy at

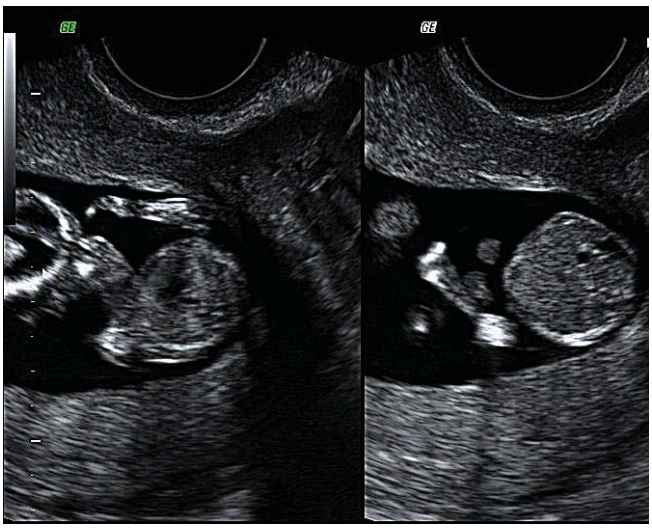

Fig. 12: Upper extremities.

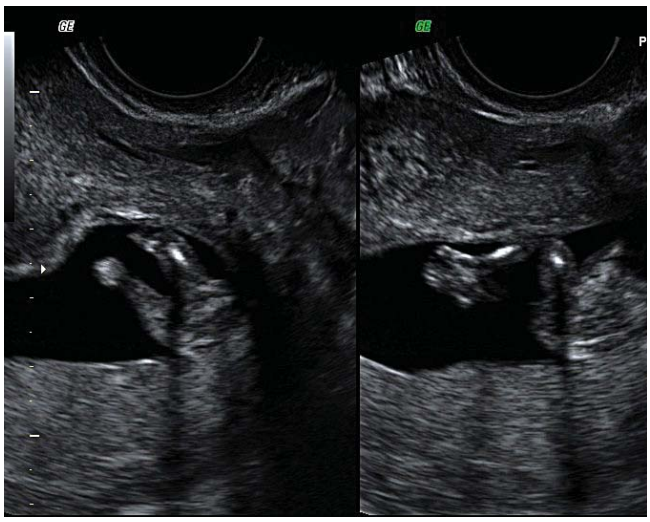

Fig. 13: Lower extremities.

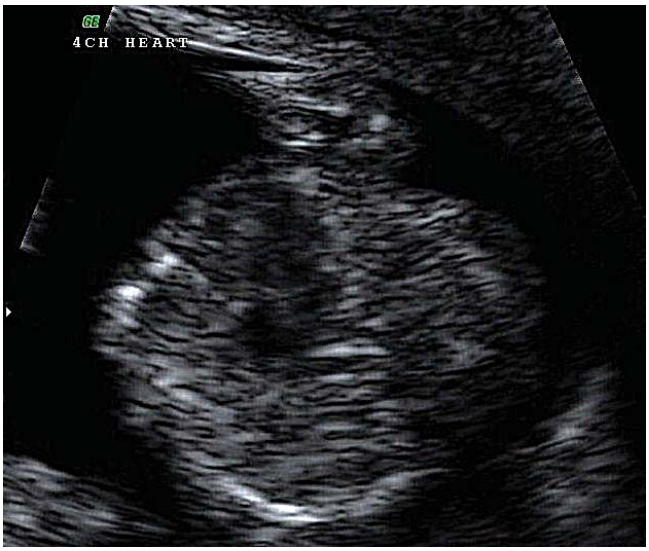

Fig. 14: Normal 4 chamber heart at 11w 6d.

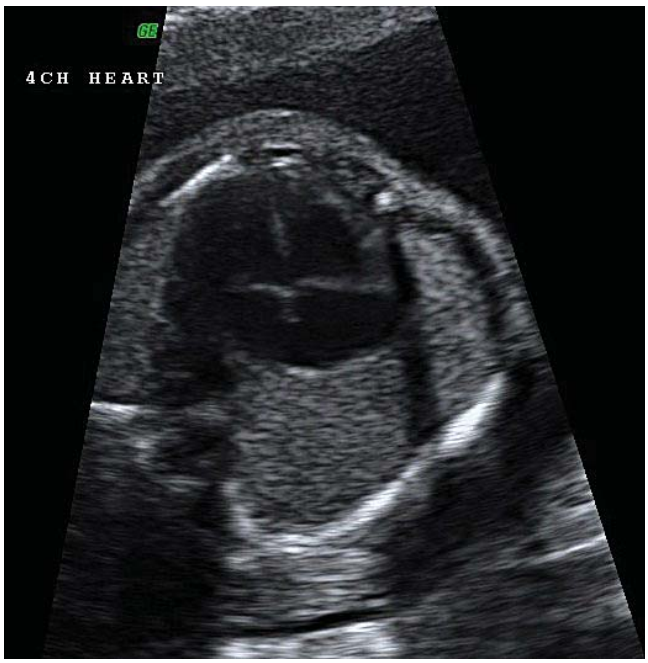

Fig. 15: Normal 4 chamber heart at 25w 6d.

11–14 weeks – skull, brain, face, spine, stomach, abdominal wall, kidneys, bladder and extremities – can be achieved in 75–98% of cases with visualisation improving with increasing gestational age[5,6,7].

Authors agree that a complete examination depends mainly on maternal habitus and gestational age and that a combination of TA and TV scanning achieves the best results. The optimal age for examining anatomy is from 12+0–13+6 weeks[5,6,7] with 13 weeks being the optimal gestation for combining fetal anatomy examination with the NT measurement[7]. The fetal heart is the most difficult organ to visualise consistently[11].

The ability to examine fetal anatomy and NT by a single three-dimensional sweep at 11–13+6 weeks gestation has recently been investigated in our centre[8,9] and further research into this area is continuing.

Table 1 provides a summary of research into the role of ultrasound in the detection of major fetal anomalies at a gestation when the NT measurement is performed. In all these studies a combination of TA and TV scans were performed – Souka and coworkers[11] using both methods routinely while the other authors only performed a TV scan if required to obtain all views of the anatomy or clarify a suspected abnormality. A scanning time limit of 30 minutes was set in all studies except that by Carvalho, *et al*[14].

In all the studies abnormalities were considered major if they were lethal, required medical and/or surgical treatment or if the anomaly caused mental handicap. Isolated increased nuchal translucency was not considered a major anomaly.

In summary, if fetal anatomy is examined by TA and (where required) TV scanning at the time of the NT exam, approximately 50% of major abnormalities can be detected.

There are many advantages to examining fetal anatomy in the first trimester:

- Early detection of an abnormality allows more time to investigate and decide upon appropriate management and treatment options
- If a severe structural abnormality is detected then termination may be offered without the need for further invasive tests such as chorionic villus sampling or amniocentesis
- It allows the option of an early versus late termination of

Table 1: Studies examining the role of ultrasound in detecting major fetal anomalies at 11–14+6 weeks.

Author	Year	Population	Number of patients	Gestational age	Incidence of major abnormalities	Ultrasound Sensitivity
Chen, *et al.*[10]	2008	Unselected	3949	12–14+6	1.6 %	47.6 %
Souka, *et al.*[11]	2006	Low risk	1148	11–14	1.2 %	50.0 %
McAuliffe, *et al.*[12]	2005	Unselected	300	11–13+6	2.0 %	16.7 %
Chen, *et al.*[13]	2004	High risk*	1609	12–14	1.6 %	53.8 %
Carvalho, *et al.*[14]	2002	Low risk	2853	11–14	2.3 %	37.8 %
Whitlow, *et al.*[15]	1999	Unselected	6443	11–14	1.4 %	59.0 %
Economides and Braithwaite[16]	1998	Low risk	1632	12–13+6	1.0 %	64.7 %

*Age 35 or above

pregnancy with a resulting decrease in surgical complications and psychiatric morbidity[7]

- It may help to protect a patient's privacy if they choose to terminate as the pregnancy is generally not physically obvious at this stage and many couples choose to delay announcement until the completion of the first trimester
- It may identify fetuses with abnormalities, which would miscarry spontaneously, providing an opportunity to investigate the cause of the abnormalities, for example by karyotyping[10]
- The reassurance of normality is beneficial, especially in pregnancies that are high risk.

There are limitations in scanning for abnormalities in the first trimester which may influence the results achievable. These include:

- The skill and experience of the sonographer, with it being essential to have an understanding of the normal development of the embryo, the processes involved in the development of fetal anomalies and the chronological order of development of specific malformations[10]
- The resolution of the ultrasound equipment on which the exam is being performed
- A combination of TA and TV examinations may be required
- The smaller the gestational age, the higher the chance of an incomplete assessment
- Unfavourable fetal position
- Maternal factors such as increased habitus or fibroids
- Longer scanning time than if only performing measurements for the NT scan, however it has been demonstrated that views are achievable by an experienced sonographer in 30 minutes or less for the majority of patients
- Many fetal anomalies have a late onset, variable onset or result from an evolving pathophysical process and therefore may not be detected in the first trimester
- The fetal heart is the most difficult organ to consistently visualise and detect abnormalities due to its small size, complex anatomy and the late manifestation of many cardiac structural and functional defects
- Early termination of pregnancy is usually surgical so post mortem confirmation of a suspected abnormality can generally not be obtained

Conclusion

Regardless of examining anatomy in the first trimester a fetal anatomy scan should be still be performed in the second trimester. However, as the NT exam is becoming more widely utilised in both high and low risk pregnancies it is obvious that parents will enquire about the well-being of their baby and expect an honest, well-informed answer.

Studies show that approximately 50% of major fetal abnormalities can be detected at the time of the NT exam and that complete visualisation of fetal anatomy is achievable in the majority of cases. As with other ultrasound examinations, patients and/or the referring doctor should be made aware of any limitations in performing a complete exam. The two cases presented highlight the benefits of both finding and excluding abnormalities.

In order to maintain high quality ultrasounds amongst different centres it is important to ensure there is a minimum standard and we suggest it is time to establish this in terms of examining fetal anatomy at the NT exam. Perhaps ASUM and other national ultrasound organisations should consider formulating new policies and guidelines in this area.

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