Ultrasound in antenatal diagnosis of structural abnormalities

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Abstract

Prenatal diagnosis commenced in the 1980s as part of routine antenatal care. Since then, the technical improvements and standardization of routine antenatal screening around the country have led to reliable diagnosis for fetal abnormalities, allowing for appropriate management strategies to be offered in a timely fashion. The National Screening Committee Fetal Anomaly Screening Program (FASP) recommends screening for eleven fetal conditions with a detection rate of more than 50%. To achieve uniformity in prenatal diagnosis, this committee has established a series of routine views, measurements and images that should be obtained and stored during the anomaly scan. In this article we will present some of the most common structural abnormalities through a series of case presentations.

Keywords antenatal diagnosis; cleft lip/palate; exomphalos; nuchal translucency; talipes; ventriculomegaly

Introduction

Ultrasound was introduced in the 1980s in the UK as part of routine antenatal care. However, uniformity on the timing and requirements of routine ultrasound scanning has only been standardized recently across the country. Obstetric ultrasound has advanced rapidly since its inception, which has aided earlier and better diagnosis of fetal abnormalities.

Currently, there are two routine ultrasound examinations offered to all pregnant women, each with different aims. The first trimester ultrasound is performed between 11 and 14 weeks of gestational age; its primary objectives are to confirm viability, date the pregnancy accurately according to the crown-rump length (CRL) and offer screening for common chromosomal abnormalities. The technological advances of ultrasound have improved resolution and capabilities to such an extent that has changed our understanding and ability to visualize early fetal anatomy. In doing so, structural abnormalities are being detected at an earlier stage, allowing for early counselling and intervention when required. The second ultrasound scan is performed between 18 and 21 weeks. The purpose of this scan is to identify major fetal structural abnormalities. The National Screening Committee Fetal Anomaly Screening Program (FASP) have identified 11 structural abnormalities with anticipated detection rates of >50%, that should be routinely screened for. These include: anencephaly, open spina bifida, cleft lip, diaphragmatic hernia, gastroschisis, exomphalos, serious cardiac abnormalities and four lethal conditions including bilateral renal agenesis, trisomies 13 and 18 and lethal skeletal dysplasias. Although some other conditions can be detected during the anomaly scan, there is insufficient published data on detection rate to establish a standard. If any of these or any other abnormality are suspected, a referral to a Fetal Medicine Unit should be made to confirm diagnosis and offer appropriate management. Identification of any malformation at this stage allows the parents to make decisions regarding continuation of pregnancy, and if the pregnancy continues, appropriate management with intervention or supportive care if required (antenatal or in early postnatal period). Table 1 shows the anticipated prenatal detection rates of these 11 structural abnormalities.

The same committee has established a series of routine views, measurements and images that should be obtained and stored during the anomaly scan. It is important to have a systematic approach when examining the fetus. Before starting the fetal biometry, it is advisable to examine the uterus and its contents, the fetal position and orientation within the uterine cavity, confirming viability, number of fetuses and chorionicity in multiple pregnancies. Once this has been established, the sonographer can proceed with fetal biometry. The standard measurements that should be obtained are: head circumference (HC), abdominal circumference (AC) and femur length (FL). These measurements need to be plotted on appropriate charts according to gestational age. If the pregnancy has not been previously dated (according to CRL on the first trimester), this needs to be done according to the HC and documented in the report. A systematic examination of fetal anatomy needs to include exam of the fetal skull (shape, ossification), brain (views and measures of the posterior horn of the lateral ventricles (atrium), cerebellum, cisterna magna and nuchal fold and evidence of cavum septum pellucidum), spine (sagittal, coronal and transverse view), face (orbits, nose, lips, alveolar ridge and profile), thorax (lungs and diaphragm) heart (establish situs, four-chamber view, left outflow and three vessel view), abdomen (stomach bubble, abdominal wall/insertion of umbilical cord), urinary tract (kidneys, renal pelvises and urinary bladder), limbs (angle of the foot, hands/palms and feet/footprints) and genitalia. In addition, the number of vessels in the umbilical cord needs to be reported, assessment of amniotic fluid, placental site and fetal movements.

We have presented five cases with conditions seen commonly in the practice of clinical fetal medicine. The discussion should help the reader gain a better understanding of the use of ultrasound in the prenatal diagnosis of structural abnormalities.

Case 1: increased nuchal translucency at the first trimester scan

A 38 year old woman in her first pregnancy attended her routine first trimester scan. She booked at 7 weeks with unremarkable past medical history, and normal booking bloods. The pregnancy was dated according to the crown-rump length (CRL) as per national guidelines, which dated her to be 12 + 4 weeks'

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Screening for fetal structural abnormalities. Detection rates

Abnormality	Detection rate
Anencephaly	98%
Open spina bifida	90%
Cleft lip	75%
Diaphragmatic hernia	60%
Gastroschisis	98%
Exomphalos	80%
Major cardiac abnormalities	50%
Bilateral renal agenesis	84%
Lethal skeletal dysplasia	60%
Edward's syndrome (T18)	95%
Patau's syndrome (T13)	95%

Table 1

gestation. The measurement of the Nuchal Translucency (NT) was 3.8 mm (>95th centile). No other major structural abnormalities were seen on the scan. The combined screening test showed a high risk for chromosomal abnormalities (1 in 32). The patient was referred to a Fetal Medicine unit for further counselling and management.

The combined screening test for chromosomal abnormalities has been used since the 1990s. It allows detection of those pregnancies at high risk of trisomy 21. The exponential correlation between maternal age and risk of having a fetus with Trisomy 21 (Down's syndrome) is now well established, and was the primary driving force behind the initial screening programme that offered amniocentesis to pregnant women above a certain age (typically 40 years). However, maternal age alone detects only 30% of all cases, and this approach to screening missed women under 40, who represented the vast majority of the pregnant population. The combination of maternal age, nuchal translucency and serum biochemistry (β-hCG and PAPP-A) improves detection to 85–90% of fetuses with trisomy 21. Whilst early studies used fixed cut-off values for nuchal translucency, it is now understood that nuchal translucency is a dynamic measurement that increases as gestational age advances. The 95th centile is therefore dependent on the gestational age (and therefore, CRL). Nuchal translucency is increased (>95th centile) in approximately 70% of fetuses with Trisomy 21. An increased nuchal translucency can also be associated with other chromosomal abnormalities (Trisomy 13, 18, Turner Syndrome and triploidy amongst others), genetic conditions and fetal structural abnormalities (most commonly cardiac defects). In the second and third trimesters, the presence of nuchal oedema is related to chromosomal abnormalities in one third of the cases (75% of them Trisomies 21 and 18). Oedema is also associated with cardiovascular and pulmonary defects, congenital infections, metabolic and haematological disorders and skeletal dysplasias.

After counselling regarding the possible causes of increased nuchal translucency, our patient decided to have an invasive test. Chorionic villus sampling was performed which showed a normal karyotype, and array-CGH (Comparative Genomic Hybridisation using microarrays) was also normal. The couple decided to continue the pregnancy and they had an early anomaly and fetal cardiac scan at 16 weeks. The cardiac scan showed a complete transposition of the great arteries (TGA) with a ventricular septal defect (VSD). No extra-cardiac structural abnormalities were diagnosed.

TGA represents 5–7% of all cardiac abnormalities with an incidence of 0.3 per 1000 live births. It is more common in males. The aorta arises from the right ventricle and the pulmonary artery arises from the left ventricle. It can be associated with ventricular septal defects (VSD) and/or pulmonary stenosis. Associated extra-cardiac abnormalities are rare and generally, there is no association with chromosomal defects. TGA is well tolerated in utero. Making the diagnosis before birth is important as it enables delivery in a tertiary unit. At birth, a prostaglandin infusion is required to maintain the patency of the ductus arteriosus. In some cases, an emergency balloon septostomy of the inter-atrial septum may also be needed. Cardiac surgery is required, involving an arterial switch of the aorta and the pulmonary artery above the valves. The prognosis is excellent provided the neonate is born at a tertiary unit and adequate treatment is implemented immediately after birth.

Case 2: ventriculomegaly (VM)

A 33 year-old woman attended her routine anomaly scan at 21 + 3 days of gestational age. The patient was booked as a low risk pregnancy at 8 + 3 weeks. The combined first trimester screening test was reported as low risk (1 in 2500 risk for Trisomy 21). She had been taking folic acid for 2 months prior to conceiving, and had stopped after the first trimester scan.

During the scan, the posterior horn of the ventricular atrium was measured as 12 mm on the left and 13 mm on the right side. The posterior fossa (cerebellum and cisterna magna) appeared normal. The cavum septum pellucidum and corpus callosum were visualized. No other extra-cranial abnormalities were noted on the scan and examination of the genitalia revealed a male fetus. Fetal biometry was within the normal ranges for gestational age (Figure 1).

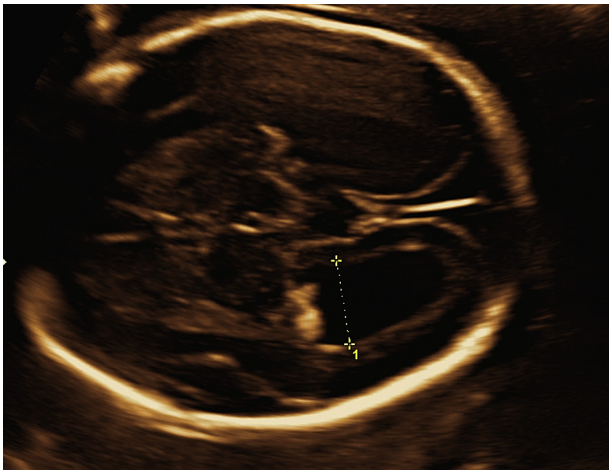

Figure 1 Ventriculomegaly. Transverse view of the head showing mild ventriculomegaly of 12 mm.

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