The use of ultrasound in the antenatal diagnosis of structural abnormalities

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Abstract

Approximately 2% of all fetuses are affected with major congenital abnormalities. Ultrasound is currently the main tool for their prenatal detection. Since its introduction in the 1970s, major advances have taken place in prenatal detection of structural abnormalities. The focus is no longer search and destroy, but to allow wider options for the parents and clinicians. Special care at delivery may also be required in some conditions. Approximately half of all major structural abnormalities can now be detected in the first trimester, but there is wide variation depending on the system involved. Specific post-natal investigations and interventions are indicated, depending on the condition diagnosed. Newer advances in molecular genetics have led to an explosion of additional information that can be obtained before birth. This has also led to difficult ethical questions.

Keywords imaging; prenatal diagnosis; structural abnormalities; ultrasound

Introduction

Obstetric ultrasound was first mentioned in 1958 in a *Lancet* paper by Ian McDonald, John McVicar and Tom Brown, entitled "The investigation of abdominal masses by pulsed ultrasound". In this landmark paper the authors described the appearance of a fetus on ultrasound for the first time, along with other abdominal masses. Nowadays obstetric ultrasound is offered to every pregnant woman in the UK as part of routine antenatal care and its role in diagnosing congenital abnormalities in the fetus is well established. Antenatal ultrasound is the most common technique used to detect structural abnormalities in the fetus. Greater quality of ultrasound scans and improved competence of operators has helped improve the detection rate of fetal abnormalities at earlier gestations. This review discusses the role of ultrasound in the diagnosis of fetal structural abnormalities.

Major congenital abnormalities occur in up to 2% of pregnancies in the UK. These abnormalities are known to increase the rate of stillbirth, as well as mortality and morbidity, not only in the perinatal period, but also throughout infancy and childhood. Diagnosing congenital abnormalities during fetal life allows wider options for the parents and clinicians. If a congenital abnormality is detected in the fetus, the couple is referred to a fetal medicine centre for proper counselling, investigation and further management. This often implies a multidisciplinary approach with fetal medicine specialists, midwives, neonatologists, geneticists and others. Special care at delivery may also be required in some patients, as well as specific post-natal investigations and interventions, depending on the condition diagnosed. In certain cases, termination of pregnancy may be appropriate, if in accord with parental wishes. According to NICE guideline 'Antenatal Care for Uncomplicated Pregnancies': "Women's decisions should be respected, even when this is contrary to the views of the healthcare professional."

Fetal anomaly screening

What is a screening test

Screening is a method of applying a test to the whole population with a view to identify a sub-group with an increased risk for a certain condition. Screening is used whenever there is a treatment to reduce the risk or complications related to that specific condition. In the UK, the Fetal Anomaly Screening Programme (FASP) offers screening of fetal anomalies to all pregnant women during pregnancy. During the first trimester scan, the risk of the baby to have Down's (Trisomy 21), Edward's (Trisomy 18) or Patau's (Trisomy 13) syndromes can be ascertained. The result of this screening is obtained combining maternal age, biochemistry and the measurement of fetal nuchal translucency using ultrasound. Individualized risk for the unborn baby to have these syndromes can be provided to each woman who accepts the offer of screening. Even though all women in the UK are offered the combined screening, women can opt:

- not to have screening
- to have screening for T21 and T18/T13
- to have screening for T21 only
- to have screening for T18/T13 only.

First trimester scan

According to UK NICE guidelines, pregnant women should be offered an early ultrasound scan between 10 weeks 0 days and 13 weeks 6 days to determine gestational age and to detect multiple pregnancies. At this scan the parents are also given the risk of the baby to have common trisomies (Trisomy 21, 18 & 13). According to the European Surveillance of Congenital Anomalies (EUROCAT), a network of population-based congenital anomaly registers in Europe, screening policies have a significant impact on prenatal detection rates for Down's syndrome. Countries that offer a first trimester screening have significantly higher detection rates than countries using a mixed first or second trimester screening policy. The Royal College of Obstetricians and Gynaecologists (RCOG, 2000) advises that an early pregnancy scan before 15 weeks be offered, in order to establish:

- gestational age accurately
- viability
- early detection of multiple pregnancies with chorionicity and amnionicity
- detection of major fetal abnormalities.

Early pregnancy is the most reliable timing to characterize chorionicity and amnionicity. Furthermore, establishment of

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Please cite this article in press as: Curado J, Bhide A, The use of ultrasound in the antenatal diagnosis of structural abnormalities, Obstetrics, Gynaecology and Reproductive Medicine (2018), https://doi.org/10.1016/j.ogrm.2018.09.005

chorionicity and amnionicity is important for the care, testing and management of multifetal pregnancies. It will also inform decisions regarding the number of visits and scans throughout the pregnancy.

With improvement of ultrasound technique, approximately half of antenatally detectable fetal structural problems can be diagnosed in the first trimester scan. In the systematic review performed by Rossi and Prefumo, almost all cases of cystic hygroma were identified, whilst anomalies of the face were less likely to be determined (34% of these anomalies were diagnosed). Congenital heart defects were the most common anomalies and only 45% of affected fetuses were identified. Nevertheless, some anomalies like ventriculomegaly manifest later in pregnancy. Also, indirect signs of spina bifida such as the "lemon sign" (discussed below) are not visible in the first trimester. Hence, the diagnosis of spina bifida is most likely to be made in the second trimester.

Mid-trimester scan

According to the NHS screening programme, this scan "is designed to identify abnormalities which mean that the baby may die shortly after birth, conditions that may benefit from treatment before birth, to plan delivery in an appropriate hospital/centre and/or to optimize treatment after the baby is born". Clearly, some women may opt not to get screened, and this option should be respected. Despite the fact that detection fetal structural abnormalities by the first trimester scan is improving, the second trimester scan remains "the standard of care for fetal anatomical evaluation in both low and high-risk pregnancies" as stated in "ISUOG Practice Guidelines: performance of first-trimester fetal ultrasound scan".

The second trimester scan can also be used to determine gestational age (although with less accuracy than the first trimester scan) and to perform measurements that allow the identification of growth abnormalities later in pregnancy. According to ISUOG Practice guidelines, this scan is an opportunity to screen for the following:

- cardiac activity
- fetal number (and chorionicity if multiple pregnancy)
- fetal age/size
- basic fetal anatomy
- placental appearance and location

Expected prenatal detection rates for fetal abnormalities are as detailed in Table 1.

It is recommended that the second trimester scan is performed between 18 + 0 and 20 + 6 weeks. The timing is crucial because it allows further diagnostic tests that may be needed and ensures the couples have adequate time to consider their options in case an abnormality is detected. The legal limit in the UK for termination of pregnancy is generally 24 weeks' gestation. Legally, pregnancies of over 24 weeks' gestation can only be terminated in exceptional circumstances.

Screening for structural abnormalities in the fetus

The most common method to detect fetal anomalies is a transabdominal scan between 18 + 0 and 20 + weeks' gestation. However, the use of transvaginal ultrasound has improved the rate of detection of fetal anomalies as early as in the first trimester scan. 80% of the most common fetal malformations develop before 12 weeks' gestation, thus the first trimester scan is a crucial. As the first trimester scan has limitations, it is also necessary to offer a second trimester scan to improve the detection rates of fetal abnormalities. Table 2 shows the proportion of examinations (n,%) with inadequate visualization of fetal organs as perceived by an examiner in early (13–14 weeks) transvaginal and late (18–22 weeks) transabdominal scans (Taipale et al. 2004).

Expected detection rate for fetal abnormalities according to FASP (Fetal Anomaly Screening Programme)

Abnormality	Expected detection rate (%)
Anencephaly	98
Open spina bífida	90
Cleft lip	75
Diaphragmatic hérnia	60
Gastroschisis	98
Exomphalos	80
Serious cardiac abnormalities	50
Bilateral renal agenesis	84
Lethal skeletal dysplasia	60
Edward's syndrome (trisomy 18)	95
Patau's syndrome (trisomy 13)	95

Table 1

Effectiveness of ultrasound in pregnancy

Despite the technological improvement in ultrasound scanning, it is still far from being perfect in detection of fetal abnormalities. Table 3 shows the sensitivity of first trimester ultrasound scans (both transabdominal and transvaginal probes were used, depending on technical issues) in detecting fetal structural abnormalities according to different studies. The best sensitivity achieved was 68%. The study with the best sensitivity is also the most recent study by Ebrashy et al. This finding is probably related to the use of better scan machines and improved image definition.

Taipale et al. evaluated the sensitivity of a two-stage screening using first trimester and mid-trimester ultrasound in detecting major fetal structural defects. They concluded that of a total of 4789 fetuses, only 18% of fetuses with major structural anomalies were found in an early scan, and an additional 30% of such fetuses were identified at the mid-pregnancy scan, adding up to 48% for the two-stage screening.

In a report by the National congenital anomaly and rare disease registration service in the UK, 71% of congenital abnormalities were diagnosed prenatally in 2015. 41.8% of these cases underwent a termination of pregnancy. Abdominal wall defects, nervous system anomalies and urinary anomalies were the conditions most frequently diagnosed prenatally. The Eurofetus study, a project involving 61 obstetric ultrasound units from 14 European countries, found that the routine mid-trimester ultrasound scan allows the detection of 55% of major anomalies before 24 weeks of gestation.

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