FETAL ABNORMALITY – ANTENATAL DETECTION

INTRODUCTION
- Prenatal screening for fetal abnormalities using second trimester ultrasound scan and maternal serum screening is offered routinely in the UK
- Routine second trimester ultrasound scans increase the detection rate for fetal abnormalities compared to scans offered on a selective basis only
- Abnormalities may be detected on an ultrasound scan at any stage of pregnancy
- Sensitivity of detection is determined by severity and type of abnormality. More severe and abnormalities developing earlier have a higher detection rate
- False positive rates from ultrasound scanning are <1%

SECOND TRIMESTER ANOMALY SCANNING
Definition
- To identify fetal conditions associated with high morbidity and long-term disability
- Performed between 18–23 weeks’ gestation
- Feticide is recommended for termination of pregnancy after 22 weeks’ gestation, which is associated with increased difficulty in managing a woman who elects termination later than this stage

Ultrasound imaging must only be performed by person fully trained in its use and experienced in detection of fetal abnormality using this technique

Before scan
- Ensure ultrasound equipment is of appropriate standard and in working order
- Check woman’s identity
- Inform woman of nature and purpose of the screening proposed and discuss the limitations of ultrasound scanning in detecting fetal abnormality i.e. sensitivity of detection is only 76% even for life-threatening abnormalities
- Treat woman sympathetically and address anxieties or concerns
- If woman does not wish to be informed of any fetal abnormalities, give her the opportunity to decline anomaly scanning but have a scan to determine placental site and fetal growth, if wanted
- Ensure early process for referral to fetal medicine specialist is in place should an abnormality be detected

ABNORMALITY DETECTED
- Sonographer performing ultrasound examination must report findings to woman personally
- Inform woman and her partner in descriptive but not diagnostic terms

If there is doubt about a diagnosis or a scan feature, refer woman to appropriate expert, giving reason for referral

Referral
- Within one working day, refer to a consultant obstetrician with fetal medicine expertise
- Fetal medicine consultant will re-scan within 5 days and explain findings to woman and her partner
- It may be necessary to repeat information. Written information and diagrams can be helpful
- When major fetal abnormalities are identified, give parents the Antenatal Results and Choices (ARC) booklet (if used locally)
- It may be appropriate for consultant with expertise in fetal medicine to offer fetal karyotyping by amniocentesis or chorionic villus sampling
- Refer confirmed fetal abnormalities in ongoing pregnancies to neonatologist
Fetal abnormalities 2013–15

- Offer antenatal counselling by the neonatologist
- More complex cases may benefit from referral to a tertiary centre e.g. to obtain access for magnetic resonance (MR) imaging or to receive antenatal counselling from neonatal surgeon
- Complete notification to the regional congenital anomaly register

Normal variants
- It is no longer recommended to screen for ‘soft markers for Down’s syndrome’. However, the following appearances should be reported and the woman referred for further assessment:
  - nuchal fold >6 mm
  - ventriculomegaly (atrium >10 mm)
  - echogenic bowel (with density equivalent to bone)
  - renal pelvic dilatation (AP measurement >7 mm)
  - all measurements compare to dating scan (significantly <5th centile on national charts)

Documentation
- A printed formal report must be produced and a copy placed in maternal healthcare record
- Record positive and relevant negative findings that are important to that particular clinical situation
- Store relevant images
- Ensure all reports are dated and signed

SCREENING FOR DOWN’S SYNDROME
- Offer Down’s syndrome screening
  - combined test (11+2–14+1 weeks) with nuchal scan or quad test (14+2–20 weeks)
  - Women with a multiple pregnancy who wish to have Down’s screening:
    - offer combined test (11+2–14+1 weeks), having had the opportunity to discuss implications of screening in twin pregnancy with either an antenatal screening midwife or fetal medicine midwife – see Multiple pregnancy guideline

INVASIVE TESTING FOR FETAL ABNORMALITY
- Performed for fetal karyotyping or other genetic testing

Amniocentesis
- Performed after 15 weeks, by an appropriately trained operator
- rate of miscarriage associated with amniocentesis is approximately 1%

Chorionic villus sampling (CVS)
- Performed after 11 weeks, by an appropriately trained operator
- rate of miscarriage following CVS is approximately 1–2%