Down’s Syndrome Screening - OSCE Station

History Taking

- A basic Obstetric history is a good place to start
- Introduction: name, status and consent!
- Present Pregnancy
  - Ascertain the first day of the last menstrual period as well as any methods of contraception and how regular their cycle is
  - Calculate the estimated date of delivery (subtracting 3 months from the LMP and adding a year and 10 days) and gestational age
  - Ask about any pre-conception folic acid
  - Ask about any previous scans/investigations/screening done
  - Smoking/drinking alcohol?
- Previous Pregnancies
  - How many previous pregnancies, their gestation and their outcomes including any miscarriages/terminations etc
  - Any previous antenatal complications, induction of labour, duration of labour, presentations and method of deliveries (and reasons why this mode was used) as well as weight and sex of babies and how they are now
  - Any postpartum complications?
- Past Medical History
  - Any chronic conditions?
  - Infectious disease (TB, HIV, Hep A/B etc)
- Family History
  - Any inherited conditions
  - Any pre-eclampsia or diabetes in the family which may develop during pregnancy?

Examination

- Explain what you’re going to do and gain consent
- Wash hands
- Ask about any pain and if there are any fetal movements
- Position patient with their head slightly elevated and cover above and below the abdomen
- Inspection:
  - Distention - normal for pregnancy?
  - Visible fetal movements?
  - Linea Nigra
  - Striae gravidarum
  - Previous scars, eg from previous C-sections
- Measure the symphysis fundal height and check it’s consistent with gestation
- Palpate
  - Use flats of hands
  - Palpate for the lie (longitudinal/oblique/transverse)
  - Presentation (cephalic/breech - if twins four poles will be palpable)
  - Palpate for engagement if cephalic presentation. If only 2/5 of the head is palpable then the head is considered engaged
- Auscultation
  - Use pinard stethoscope or a Doppler ultrasound monitor to listen for the fetal heart rate over the anterior shoulder
  - Normal rate is 110 to 160 beats per minute
- Cover patient, help them up and wash hands

**Down’s Screening - OSCE Mark Scheme**
- Down’s syndrome is found in 1 in 1000 pregnancies but increases to 1 in 300 pregnancies in mothers over 30
- Caused by three copies of the chromosome 21
- Characterised by a floppy baby at birth, protruding tongue, heart defects, and intestinal defects. Learning difficulties, cataracts and dementia also occurring at an increased rate in later life. All babies affected to different degrees which is the unpredictable part.
- Can do a number of screening tests through ultrasound and biochemical tests
- 1st Trimester Screening is performed 11 to 14+2 weeks with an adjusted risk score. The nuchal translucency is measured via ultrasound (layer of fluid at back of the head on ultrasound indicates a greater risk of down’s), human chorionic gonadotrophin (HCG) is raised and pregnancy associated plasma protein (PAPP) is lowered on blood tests. Maternal age at delivery (donor’s age if donated eggs), weight, smoking status, gestation and ethnicity are also taken into account. Greater than a 1 in 150 risk is considered high risk for this test and should proceed to further tests
- 15 to 20+6 weeks is Quadruple Screening on bloods: HCG (raised), inhibin (raised), oestridol (decreased), alpha-fetoprotein (AFP - decreased). Maternal age, weight, smoking status, gestation and ethnicity are also taken into account here. Greater than a 1 in 200 risk is considered high risk for this test and should proceed to further tests
- Exceptions to these screening programmes include multiple pregnancies (only nuchal translucency should be used here) or if a woman has had a previously chromosomally abnormal baby or her or her partner have a known chromosomal abnormality. If this is the case, she should proceed straight to diagnostic testing
- If any of these indicate an increased risk, Chorionic Villus Sampling (CVS) can be done between 11+0 and 13+6 weeks and Amniocentesis can be performed between 15 and 17 weeks. Counselling should also be offered
- CVS involves a needle through the abdomen wall to take a sample of the placental cells through ultrasound guidance. 2% risk of miscarriage. 1% risk of Confined Placental Mosaicism
- Amniocentesis is a similar concept but a sample of amniotic fluid is taken instead. Not as many chromosomal/ genetic abnormalities can be detected using this method. 1% risk of miscarriage and safer then CVS beyond 15 weeks
- Location of placenta may influence decision between amniocentesis and CVS as well as gestational age
- Preliminary results are normally available within 3 working days
- These screening processes identify 70% of Down’s cases, categorising 20% of women over the age of 35 in the high risk categories
- If these tests do indicate Down’s, the option to terminate the pregnancy will be offered but more information regarding that will be discussed in such as event