







Prenatal screening

during the first 20 weeks of pregnancy

Things to keep in mind before you decide to have prenatal screening

Ultrasound and prenatal screening is offered to all women at 11-14 weeks and again at 20 weeks. The vast majority of babies are normal at birth, however all women, whatever their age, have a small risk of delivering a baby with physical and/or mental handicap. It is normal for parents to be concerned regarding their babies health. Some parents choose to find out as much as possible about their childs health, while others prefer not to look for problems.

If a fetus has a major structural or chromosomal abnormality parents may have to take a difficult decision concerning the future of the pregnancy. If you don't want to have this information, you may consider not to have prenatal screening.

Is it possible to detect all malformations by ultrasound ?

No, normal results from prenatal screening cannot rule out all malformations.

What is the procedure if a malformation is detected?

Our task is to assess what type of anomaly may be present and advice accordingly. Sometimes further investigations are required such as taking a sample from the placenta (chorion villous sampling) or amniotic fluid (amniocentesis) to find out if the baby's chromosomes are normal. In certain circumstances a special ultrasound examination of the baby's heart is required (fetal echo) which is performed by a pediatric cardiologist. Depending on the nature of the problem a consultation may be recommended from various specialists, for instance cardiologists, plastic surgeons, renal specialists or as is appropiate in each case.

LANDSPITALI - UNIVERSITY HOSPITAL	TEL:	543 1000
ULTRASOUND UNIT open from 8:00 - 16:00	TEL:	543 3256
ANTE-NATAL WARD	TEL:	543 3070

What is ultrasound?

Ultrasound is based on the use of high-frequency sound waves, which make it possible to look into the tissues of the body. A black and white picture is seen on a televison screen. Dense tissues like bone are white while fluids are black. Other tissues appear as various shades of grey.

Ultrasound makes it possible to look into the body of the fetus, look at it's organs and follow it's movements. Ultrasound is considered safe for the mother and fetus, but we nevertheless consider it a medical investigation which should only be done when indicated.



Midwives and doctors who have specialised in prenatal diagnosis and counselling perform the ultrasound examinations at the Prenatal Diagnosis Unit at Landspitali University Hospital (LSH).

Ultrasound at 11-14 weeks

Ultrasound at 11-14 weeks

- to confirm gestational age
- to check that the fetus is developing normally
- to diagnose multiple pregnancies
- to measure nuchal translucency and the nasal bone. By adding measurements of two hormones in the mothers blood a combined risk assessment is performed for trisomy 21 and trisomy 13 and 18
- unfortunately at this time 3% of women will be diagnosed with miscarriage.



Nuchal translucency

This is a collection of fluid under the skin behind the neck of the fetus at 11-14 weeks that can be measured by ultrasound examination. In fetuses with chromasomal abnormalities, cardiac defects and some genetic syndromes nuchal translucency is increased.

Nasal bone

Recent studies show that the presence of nasal bone at 11-14 weeks lowers the risk for trisomy 21. Nasal bone is absent in 2/3 of fetuses with trisomy 21.

Biochemical markers

By measuring the levels of two hormones in the mother's blood (free betahCG and PAPP-A) and combining with ultrasound information the sensitivity for trisomy 21 and trisomy 13-18 is increased.

Combined screening test

Each woman will be given an estimate of her individual risk for trisomy 21 and trisomy 13-18. This is calculated by taking into account the age of the mother, measurements of two hormones in the mothers blood and the findings of nuchal translucency thickness and the presence or absence of nasal

bone. If the results of the combined screening test puts you at risk, you will receive full counselling concerning the significance of these risks and the options for further testing discussed such as fetal chromosomal testing.

Women with a nuchal translucency measurment greater than 3,5 mm will be offered fetal echo at 20 weeks, irrespective of chromosomal results.

Chorion villous sampling (CVS) and Amniocentesis

The main indication for CVS/amniocentesis is to examine the chromosomes of the fetus. Normally there are 46 chromosomes in every cell. The chromosomes are arranged in 22 pairs, a total of 44 and in additon there are two sex-chromosomes which determine gender. The chromosomes carry the genetic code of the body. In the placenta and in the amniotic fluid there are live cells from the fetus. These cells are cultured and investigated for chromosomal abnormalities. The most common is trisomy 21 (Down's syndrome). In trisomy 21 all cells carry an extra chromosome number 21. Other chromosome defects are less common.

Who is offered fetal chromosomal testing?

Women:

- with previous history of fetal chromosomal abnormalities
- with family history of inherited chromosome abnormalities
- if ultrasound examination at 11-14 weeks or 20 weeks reveals risk factors for chromosomal abnormalities.
- older than 35 years who do not want risk assessment with ultrasound and/or biochemstry.

How is the chorion villus sampling and amniocentesis performed?

Chorion villus sampling (CVS)

Local anesthetic is given. A fine needle is then passed through the mother's abdomen into the placenta and a sample of the placenta is obtained. During the procedure the needle is carefully observed with ultrasound.

This is most often done at 11-12 weeks but sometimes later in the pregnancy. The result is available 2 days later for chromosomal aberration but can take longer for inherited diseases. The medical staff at the prenatal diagnosis unit of the University Hospital (LSH) will counsel you regarding when this test is appropriate. The risk of miscarriage following CVS is approximatly 1%.

Amniocentesis

A fine needle is introduced into the amniotic cavity and 10 ml of the fluid surrounding the fetus is removed . During the procedure the needle is carefully observed with ultrasound.

This is most often done at 15 weeks. Results are available 2-3 weeks later. The risk of misscarriage is 1%.

After these procedures it is recommended to stay in bed for the first 24 hours followed with light activity for the next two days. It is normal to feel slight uterine contractions and pressure. You may find it helpful to take medication such as paracetamol (2 tablets 500 mg. each every 6-8 h's).

If you experience strong contractions, bleeding or leakage of fluid, feel free to call us at the ultrasound unit tel: 5433 256 from 8:00-16:00 after four o'clock please call the ante-natal ward at Landspitali tel: 543 3070.

Ultrasound at 20 weeks

- to confirm gestational age
- diagnose multiple pregnancy (if there is no prior ultrasound)
- localize the placenta in the uterus
- to assess fetal health and look for structural malformations

We assess gestational length for all babies at this time and predict expected date of delivery. Most babies are of similar size during the first 20 weeks, therefore it is a good time to calculate the due date. We do this by measuring the diameter of the fetal head and the length of the long bones, such as the femur and humerus. At 20 weeks the baby is about 25 cm long and weighs about 300 grams.



Profile at 20 weeks

The baby is examined for fetal anomalies, and sometimes further examinations are needed such as fetal echo or invasive testing like chorion villus sampling or amniocentesis.

The placenta is localized in the uterus. In about 4% cases the placenta is low lying i.e. close to the opening of the womb. This calls for re-assessment usually at 34 weeks.

Kindly observe the following

- Please do not bring children younger than 12 years along for an ultrasound examination. Children under this age have difficulties in relating the ultrasound picture to a baby and they will often be unruly. This may affect the concentration of the ultrasonographer and disturb the parents who may not enjoy the experience.
- If something abnormal is found during the ultrasound examination, the presence of a child is not advisable.
- The examination takes about 20-30 min. Sometimes a repeat exam is needed if the baby has been in a difficult position. Please remember to bring the referral form with you.
- You can purchase pictures of the baby. Please state this at the beginning of the examination.
- If you wish to know the gender of the baby please let us know at the beginning of the examination. It is not always possible to identfy the gender it depends on the position of the baby.





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