About the foetal diagnosis

All foetal diagnoses are voluntary. Pregnant women always decide for themselves whether they want to have a test done or not.

Undergoing early foetal diagnosis

Early foetal diagnosis means a medical examination of the pregnant woman and foetus before the end of the 22nd week of pregnancy for the purpose of obtaining more information about the foetus.

If you undergo a foetal diagnosis, the examination may sometimes show that the foetus is sick or has some sort of deformity or abnormality. In rare cases the examination may also show that there is an increased likelihood that the foetus has a chromosomal abnormality. You may have your own view on whether you want to undergo further examinations.

If anything abnormal about the foetus is found, you will be given information about the medical and social consequences of the abnormality, and about the care that is available. In some cases, the foetus may have a serious condition. In such case, it may be appropriate to discuss whether you want to continue or terminate your pregnancy.

It is important to remember that not all illnesses a foetus may have can be detected through foetal diagnostics. Even if everything seems to be perfectly normal during the examination, there may be conditions that can affect the health of the foetus.

The foetus may also become ill during pregnancy, just like you.

If you or your partner have a predisposition to hereditary diseases or chromosomal abnormality, you should inform your obstetrician of this.

Routine ultrasound screening

All pregnant women are offered ultrasound screening, which is usually carried out during weeks 18-20 of the pregnancy. The purpose of the screening is to

- determine the length of the pregnancy and estimate the date of expected delivery
- see how many fetuses there are in the womb
- examine the foetus so that serious deformities or abnormalities can be detected
- ascertain the position of the placenta

The examination does not include the determination of the sex.

Some abnormalities in foetuses are easier to detect with ultrasound, while others are harder to detect. Not all abnormalities can be detected with ultrasound. There may even be conditions where it is difficult to assess the direct significance of the abnormality.

Support and further information are always available if anything unusual about the foetus is discovered.

CUB

CUB stands for ‘combined ultrasound and biochemical screening’. Biochemical screening involves a blood test and analysis for determining the likelihood of the three most common chromosomal abnormalities, trisomy13, trisomy 18 and trisomy 21.

Trisomy is a form of chromosomal abnormality whereby a person has three copies of a chromosome instead of the usual two.

Trisomy 21, which is another name for Downs syndrome, is the most common form of trisomy.

It is important to understand that CUB is not a secure way of identifying chromosomal abnormality, but a tool for assessing the degree of likelihood of trisomy 13, 18 or 21.

CUB is carried out in week 11-13+6 of the pregnancy. The foetus has to be sufficiently large and the test is usually carried out after 12 or 13 complete weeks of pregnancy, when it is most appropriate for the foetus’s development. An ultrasound examination is carried out to measure the size and heart rate of the foetus

and to inspect its organs. A nuchal translucency (NT) scan of the foetus is also carried out.

A blood sample is taken from the pregnant woman approximately one week before the ultrasound screening.

The results of the ultrasound screening and blood sample are weighed against the woman's age and weight, and fed into a computer program that calculates the likelihood of trisomy in the foetus.

In the Scania Region CUB is provided free of charge to all pregnant women over the age of 33 at the time of last menstruation.

CUB can also be carried out if you are expecting twins, but it involves a number of special considerations.

If the CUB screening shows an increased likelihood of trisomy 13, 18 or 21, continued screening is provided. If

- the CUB screening shows a likelihood of 1/51- 1/1000 for trisomy 13, 18 or 21, a NIPT is provided.
- the CUB screening shows a likelihood of 1/2 - 1/50 for trisomy 13, 18 or 21, an amniotic fluid test or placenta test is provided.

NIPT (Non-invasive prenatal test)

In the Scania Region, the NIPT is always preceded by CUB screening.

The NIPT is a method that enables the foetus's DNA to be checked by taking a blood sample from the woman. Like CUB, the NIPT is a test that indicates the likelihood of chromosomal abnormality, but with a higher degree of accuracy. Just as with CUB, the likelihood of trisomy 13, 18 and 21 is assessed. The NIPT is not carried out on women expecting twins, nor in the case of egg donation.

The NIPT can be carried out from week 10 of the pregnancy, but it is most common to take the test around week 12. If

- the NITP shows a high likelihood of trisomy 13, 18 or 21, an amniotic fluid test or placenta test is provided.

Amniotic fluid test/placenta test

The test involves inserting a thin needle through the abdominal wall in order to retrieve a small amount of amniotic fluid or cells from the placenta.

- The earliest the amniotic fluid test can be done is in week 15+0 of the pregnancy
- The earliest the placenta test can be done is in week 11+0 of the pregnancy
- The test involves an increase in the risk of miscarriage of <0.5 %

The amniotic fluid test/placenta test is analysed primarily by using a method that provides results for trisomy 13, 18 and 21 within one week.

Further analyses of the sample may need to be done, in which case the results period is extended.

What happens then - after the tests?

In the vast majority of cases, the tests are fine and the child is born completely healthy. If the ultrasound screening indicates any form of abnormality, a further assessment by a doctor is always made. Further ultrasound screening may be appropriate, and you may be asked whether you want to undergo an amniotic fluid test or placenta test.

If your foetus is shown to have a chromosomal abnormality and/or deformity, you will be given detailed information and the opportunity to discuss what this might imply. No one has the right to interfere with or attempt to influence your decision if it does not concern them. It may sometimes help, however, to talk with several experts, for example, a genetics advisor, children's doctor, psychologist or counsellor, to enable you to reach the decision that is best for you. It may also be worth while using the links given below to learn more about what it means to be a parent of a child with a disability.

Useful links:

www.1177.se/Skane/Fakta-och-rad/Undersokningar/Fosterdiagnostik/

www.fub.se

www.svenskadownforeningen.se

www.agrenska.se

www.medscinet.se/gensvar

www.socialstyrelsen.se/funktionshinder