Information about prenatal testing

Most children are born healthy

Most children are born healthy but one to two per cent of all newborn children have some kind of serious inherent abnormality, for example malformation or chromosomal abnormalities. Prenatal testing is the common name for examinations (including ultrasound), which can be carried out in order to find out if the fetus has any sort of malformation or chromosomal abnormality. It is important to know that screening cannot give definite answers to everything and that there is no guarantee the child will be healthy.

All screenings are voluntary.

All pregnant women in Stockholm are offered to do a Combined ultrasound and biochemical screening (CUB) or regular ultrasound between pregnancy week 12-14 and a ultrasound between pregnancy week 18-20

35 years or older at the time for delivery

Increased age in pregnant women raises the probability of certain chromosomal abnormalities. The most common abnormality is Down´s syndrome. Information about the chromosomes of an unborn child can only be obtained by way of an amniocenteses or placenta test. As the amniocentesis and placenta tests are associated with an increased risk of miscarriage, a screening is initially offered to find out if there is a high probability that the fetus has trisomy 21, 13 and 18. This is known as CUB screening.

Ultrasound week 12-14

A.) CUB-test or B.) Regular ultrasound

A.) CUB (Combined ultrasound and biochemical screening)

The purpose of CUB is to estimate the probability of the fetus having Down´s syndrome (trisomy 21) or two more serious chromosomal abnormalities (trisomy 13 and 18).

What is the procedure?

The biochemical test can be carried out from the 9th full week of pregnancy, but no later than 5 days before the ultrasound screening. The ultrasound part of the screening can be carried out between 12-14 weeks of pregnancy. The ultrasound screening takes a measurement of the fluid, which gathers at the nape of the fetal neck (nuchal translucency). By combining the age of the mother, the measurement of fluid at the nape of the fetal neck and the results of the biochemical test and some other parameters it is possible to determine the probability of the fetus having one of the above mentioned chromosome abnormalities.

How do I get the results?

In connection with the ultrasound screening, the women is told whether or not there is a high or low probability of her unborn child having chromosomal abnormalities. Most women are told that the probability is low.

- If the probability is assessed as low, no further screening is offered.

- If the probability is assessed as high, the women is offered a NIPT, amniocentesis or placenta test.

B.) Regular ultrasound (pregnancy week 12-14)

Ultrasound is performed via the abdomen and the anatomy of the fetus is checked.

Ultrasound week 18-20

The ultrasound screening is offered to all pregnant women. It is carried out between weeks 18-20. It is not painful and does not carry any known risks. The aim is:

- To find out expected delivery date.
- To see if there´s more than one fetus.
- To detect if there is any serious fetal abnormalities.
- To check the location of your placenta in the uterus.

The ultrasound examination shows full image of the child. The width of the head and femur is measured to get an estimated date of delivery. The examination also enables detection of the other more serious malformations such as myelocele and abdominal wall rupture as well as a number of abnormalities in the skeleton or internal organs. In the event that a serious abnormality is detected the woman will be offered amniocentesis or placenta test (see below).

NIPT – Non Invasive Prenatal Testing

NIPT is a screening method where you through a blood sample from the mother analyze DNA from the fetus. With an accuracy of just over 99 percent, the method provides answers to whether the fetus carries a chromosomal abnormality in chromosomes 13, 18, 21 and the sex chromosomes X and Y. If the result is positive, an invasive test is recommended to confirm the result. The test does not give a diagnosis but only a probability calculation. It is important to have an informed choice, so that you understand the purpose and know what you are accepting

Amniocentesis

This test cannot be carried out before 14-15 weeks of pregnancy. First an ultrasound screening is carried out to see the fetus and determine how far along into the pregnancy the woman is. A needle is then inserted into the uterus and a small quantity of fluid extracted. The sample is sent for chromosomal analysis. It is important to know the amniocentesis procedure is associated with an increased risk of miscarriage. Statistics show one miscarriage per 100-200 samples.

Placenta test

The placenta test can be carried out no earlier than the 11th week of pregnancy.

This test also begins with an ultrasound screening. Once fetus has been seen and it has been assessed how far the pregnancy has progressed, a needle is inserted into the placenta and a small sample collected. The placenta test is also associated with an increased risk of miscarriage. Statistics show one miscarriage per 100-200 samples.

Results and answers

Women whom have undergone an amniocentesis or placenta test due to their age or concern over Down's syndrome are initially offered what is known as a fast analysis test. This means that you can get a definite answer about the probability of Down's syndrome and other common chromosomal abnormalities after just one week. You may also find out gender of your child.

In the event of hereditary chromosomal abnormalities or if an amniocentesis test has been carried out because of an abnormality being detected during the ultrasound screening, a full chromosome mapping is recommended. The result of the chromosome mapping is advised after three weeks.

Take time to contemplate before making your decision about prenatal tests.

Support and help

If it appears that the fetus has some sort of illness or malformation, you will receive all necessary support and help.

Information from Karolinska University Hospital and Vårdguiden.