Further testing after prenatal screening

EXAMINING A SUSPECTED CHROMOSOMAL OR STRUCTURAL ABNORMALITY
Ultrasound examinations

- **Early Pregnancy General Ultrasound** at 10–14 weeks of pregnancy may find structural abnormalities in the foetus. Structural abnormalities that are found so early in pregnancy are often significant. Further testing is needed in many cases to assess what the results (such as an increased amount of fluid in the foetus) mean. Such further tests are mostly done at maternal hospital clinics. A suspicion may turn out to be false and the child comes into the world healthy.

- A **Nuchal Translucency (NT)**: The results of the NT scan are combined with the results of a blood test taken at 8–11 weeks of pregnancy to calculate a risk rate. If the NT scan indicates that the amount of fluid behind the neck of the foetus is unusually high or if the risk rate is high, the family will be offered a foetal chromosome test and often also a new ultrasound examination. If both the chromosome test and the later structural ultrasound examination give normal results, no abnormalities will usually be found in the child after birth.

- **Ultrasound Screening for Structural Abnormalities at 18–21 weeks of pregnancy or after 24 weeks of pregnancy**: This examination is able to find most significant structural abnormalities in the foetus. Many of the abnormalities can be treated surgically after birth. Sometimes it cannot be assessed on the basis of a single examination how severe or significant a detected abnormality is. The family will then be offered a new examination at a prenatal examination unit or various further testing. These tests aim to determine more accurately what the abnormality is like and what has caused it. The examinations also help estimate the further development of the pregnancy and the outlook for the child to be born.
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What does an abnormal result mean?

- The family will be offered an opportunity to discuss an abnormal result and what it means with an experienced obstetrician or geneticist. Where necessary, other specialists can be consulted, such as a paediatrician or paediatric surgeon. Other support persons may also be needed.

Structural abnormalities found in ultrasound examinations vary widely in severity and many of them can be treated after birth for example by surgery.

It is the family and ultimately the pregnant woman herself who decide how the test results will affect the course of the pregnancy. After consideration, some decide to continue the pregnancy while some others choose to terminate it. It is advisable that the family discusses what an abnormal result would mean to them already before deciding to participate in further prenatal testing.

If the foetus is found to have a severe structural abnormality in the ultrasound examination, the pregnancy can be terminated on that basis up to the end of week 23 (up to 24+0 weeks of pregnancy) by permission from the National Authority for Medicolegal Affairs (Section 5 a of Act 239/1970). It is the family and ultimately the pregnant woman herself who decide whether to continue or terminate the pregnancy.

If the foetus is found to have a structural abnormality at or after 24 weeks of pregnancy, the family will be offered an opportunity to discuss the test result and what it means with a doctor. Where necessary, other practitioners and support persons can be consulted as well. The Finnish legislation does not allow pregnancy to be terminated on the grounds of a disease of the foetus.

The family also has the right to change their opinion at any stage during screening or further testing.
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At this stage. Where necessary, further testing can be done, which is also voluntary. These tests aim to determine more accurately what the abnormality is like and what has caused it. They also help estimate how the pregnancy will develop and the outlook for the child to be born. At the same time, information can be obtained that helps plan the delivery and the care of the newborn child.

Chromosomal abnormalities

What does it mean that you belong to the risk group?

About five in a hundred pregnant women receive an abnormal result from Early Pregnancy Combined Screening. The result means that the risk for the child to be born with Down syndrome is higher than 1:250. In other words, a screening result that indicates an increased risk does not yet mean that the foetus would have a chromosomal abnormality. You will be offered further chromosome testing where a sample is taken of the placenta or amniotic fluid. These further tests are fully voluntary. Often there are no chromosomal abnormalities and the child is born healthy.

Further testing

- Chorionic Villus Sampling (CVS): A small amount of cells is taken from the placenta so as to examine the chromosomes of the foetus. The sample is taken at 11–13 weeks of pregnancy during an ultrasound scan. It is taken using a needle through the abdomen if the placenta is located such that the sample can be taken. The sampling feels about the same as taking a blood test. After the procedure you can move normally and
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The examination and counselling take 1 to 3 hours.

There is a risk of miscarriage associated with the CVS test: About one in one to two hundred pregnancies will terminate following the test. Miscarriages are also otherwise rather common in early pregnancy: four to five in a hundred women at 10 weeks of pregnancy will have a miscarriage during the next weeks. As natural miscarriages are so common, the cause of an individual miscarriage may remain unclear.

The results of the chromosome test are available within 1 to 4 weeks of the sampling. Sometimes it is also necessary to take a sample of amniotic fluid (amniocentesis) to be able to interpret the sample taken of the placenta.

**Amniocentesis:** The chromosomes of the foetus can also be examined by analysing cells contained in amniotic fluid. An amniocentesis is usually done at 15–16 weeks of pregnancy, that is, a sample is taken of amniotic fluid using a needle through the abdomen during an ultrasound scan. The sampling feels about the same as taking a blood test. After the procedure you can move normally and go to work. The examination and counselling take 1 to 3 hours. About one in one to two hundred pregnancies will terminate following the test. Even without the sampling, 1 to 2 in a hundred pregnancies will terminate after 15 weeks of pregnancy.

The results of the amniocentesis are available within 2 to 4 weeks. The result of the chromosome test is very reliable. Unclear results that require further testing are very rare.

Before the sample is taken, a specially trained midwife or doctor will give you more detailed information about the risks, sampling procedure and results. You may then also ask any further questions.
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**WHAT IF THE FOETUS IS FOUND TO HAVE A CHROMOSOMAL ABNORMALITY?**

The CVS or amniocentesis tests may reveal a Down syndrome or some other chromosomal abnormality. You will then be offered an appointment with a geneticist or other specialist and a support person. Some chromosomal abnormalities cause no symptoms in the child to be born. After the test results are available, you will have only a little time for consideration in order to decide whether to continue or terminate the pregnancy. After consideration, some families decide to continue the pregnancy while some others choose to terminate it. Alongside the chromosomal abnormality, the decision may be influenced by possible severe structural abnormalities found in the child. The family also has the right to change their opinion at any stage during screening or further testing.

If the foetus is found to have a severe chromosomal abnormality in the ultrasound examination, the pregnancy can be terminated in Finland on that basis up to the end of week 23 (up to 24+0 weeks of pregnancy) by permission from the National Authority for Medicolegal Affairs (Section 5 a of Act 239/1970).

**WHAT IF THE RESULT OF THE CHROMOSOMAL TEST IS NORMAL?**

Normal chromosome test result means that the foetus has a normal number of chromosomes. However, the test is not always able to identify minor chromosomal abnormalities.

Many diseases and defects cannot be found by prenatal tests. In other words, the chromosomes can be found to be normal even if the child had a disease or defect. No screening method can determine with certainty whether the foetus is absolutely healthy.

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*When the foetus is found to have a severe developmental disorder, it is always the pregnant woman herself who decides whether to continue or terminate the pregnancy.*
Further information and peer support is available

The severity of the child’s possible disability is not revealed in the chromosome test but will only to be seen over time. Although no curative treatment is available for many disabilities, the development of disabled children and adults can be supported by individually planned rehabilitation. There are also various other support services available to them and their families. Families whose child has an illness or disability often cope better than what they expected in advance. A child always adds both joy and challenges to life.

Additional information and support is offered during and after pregnancy by health and social care professionals and a great number of different disability and parent associations. Information and open discussion will relieve concern and help make the decisions. The hospital staff will also help the family in the beginning, for example by providing information about support families. Families also receive help from various associations that provide a wide range of training, publications, recreational activities and adaptation training courses.

# Glossary

| **CHORIONIC VILLUS SAMPLING (CVS)** | Using ultrasound as a guide, a sample is taken of the placenta through the mother’s abdomen so as to examine, for example, the chromosomes of the foetus |
| **CHROMOSOME** | Each human cell normally contains 46 chromosomes or 23 pairs of chromosomes (chromosome pairs 1–22 and sex chromosomes X and Y) |
| **AMNIOCENTESIS** | Using ultrasound as a guide, a sample is taken of amniotic fluid through the mother’s abdomen so as to examine, for example, the chromosomes of the foetus |
| **NUCHAL TRANSLUCENCY (NT)** | Measuring the amount of fluid behind the neck of the foetus by an ultrasound scan at 10–12 weeks of pregnancy |
| **WEEKS OF PREGNANCY** | Time from the first day of the last menstruation to the time of the examination, given in full weeks + days (for example 12+3) |
| **RISK RATE** | An estimate calculated by a computer program concerning the risk of the child to be born with Down syndrome |

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