

# VARSINAIS-SUOMEN SAIRAANHOITOPIIRI EGENTLIGA FINLANDS SJUKVÅRDSDISTRIKT

## Foetal screening

Information about the screening for foetal chromosomal and structural anomalies



This pamphlet is intended for all parents who are expecting a child. It is desirable that the parents familiarize themselves with the pamphlet together. It is voluntary to participate in a foetal screening.

### Why is screening of foetal anomalies carried out?

The aim with the examinations carried out at the maternity clinic is to find out if the pregnancy has progressed normally and to get information about possible risk factors.

**All screening is voluntary.**

**The family and ultimately the pregnant woman decide about participation in these screenings.**

Each pregnant mother wishes that the foetus will develop normally and that the child born is healthy. This wish doesn't always come true. Some kind of structural or chromosomal anomaly is discovered in about three out of a hundred newborns. The anomaly is severe in one child out of a hundred. A part of the anomalies can already be discovered in the foetal examinations that are carried out during the pregnancy.

Such rare foetal illnesses and structural anomalies, which cannot in any way be discovered during the pregnancy, also exist. Those screening methods which are described in this pamphlet have been chosen and scheduled so that they would be as accurate and reliable as possible.

Many parents wish to find out about the foetal anomalies, since it can help the planning of the follow-up of the pregnancy, as well as of the delivery. Discovering a foetal illness already before the delivery can ease planning the care of the newborn. If the foetus is found to suffer from a severe illness or disability or have a higher risk to get them, the family has the possibility to consider terminating the pregnancy.

Before making the decision about participating in the foetal screenings, it is good for the family to also consider possible consequences. Luckily the examination results are usually normal. The results can also indicate higher risk for foetal disability or illness. When the examination results are ready there is often very little time to decide about further examinations and whether the pregnancy is to be continued or terminated.

### **What are the screenings for and what can be found through them?**

The risk for anomalies grows with the increasing maternal age. A part of the chromosomal anomalies can be screened already in examinations done during the pregnancy. The most common chromosomal anomaly is Down's syndrome, i.e. trisomy 21. A higher risk for other chromosomal anomalies can also be found in the screening. With an ultrasound scan different structural anomalies can be discovered even if the chromosomes would be normal.

**The maternity clinic makes sure that the examinations are carried out at the right time.**

### **A general ultrasound scan during the early pregnancy**

A general ultrasound scan during the early pregnancy is available for all pregnant women and it is carried out during pregnancy weeks 0+0–13+6 (or thereafter). The main aim of this examination is to ensure that the foetus is alive, to confirm the duration of the pregnancy and to determine the number of foetuses. The aim of this examination is not to look for foetal anomalies, but considerable and severe structural anomalies can already be seen in this examination. If an anomaly is suspected, the mother is informed about it.

However, many structural anomalies cannot as yet be seen at this stage of pregnancy. The majority of the foetuses are normal.

## Screening for foetal anomalies

### Combined screening during early pregnancy

The first and foremost screening method of chromosomal anomalies is the so called **first-trimester combined screening: An ultrasound scan, where the foetal nuchal translucency is measured is carried out during pregnancy weeks 10+0 – 12+6. Blood samples are also taken from the pregnant woman during the same visit.**

By combining the blood sample with the result from the measuring of the nuchal translucency, those foetuses with an increased risk for chromosomal anomalies can be detected. Information about the results of the chromosome screening can be obtained either from the own maternity clinic or the unit which is responsible for screening in the area.

**If you end up in the group at risk in the first stage of the screening, it still does not mean that the foetus would have an illness or a disability.**

If the combined screening shows a greater risk for a chromosomal anomaly, the mother is offered a possibility for a foetal chromosome examination either through a placental or amniotic fluid sample. This further examination confirms if the number of foetal chromosomes are normal. The puncture during the sample taking may cause miscarriage. The likelihood that this happens is about one out of a hundred.

Screening methods which are based on blood sample from the mother cannot be used in twin pregnancies or other pregnancies with multiple foetuses. In such situations the foetal nuchal translucencies can still be measured and the risk for chromosomal anomalies can thus be estimated.

The maternal age increases the risk for chromosomal anomalies. In certain municipalities a mother who has turned 40 has, if she so wishes, the possibility to participate in a chromosomal examination of the placental or amniotic fluid sample directly based on age without preceding screening. You can inquire about this possibility at your own maternity clinic.

As a secondary chromosomal screening method based on blood samples, serum-screening can be used during the second trimester of the pregnancy in such cases where it is too late to do a combined screening. This examination is carried out during pregnancy weeks 15+0 – 16+6. More information about this examination can be obtained from the maternity clinic.

### Structural ultrasound scan

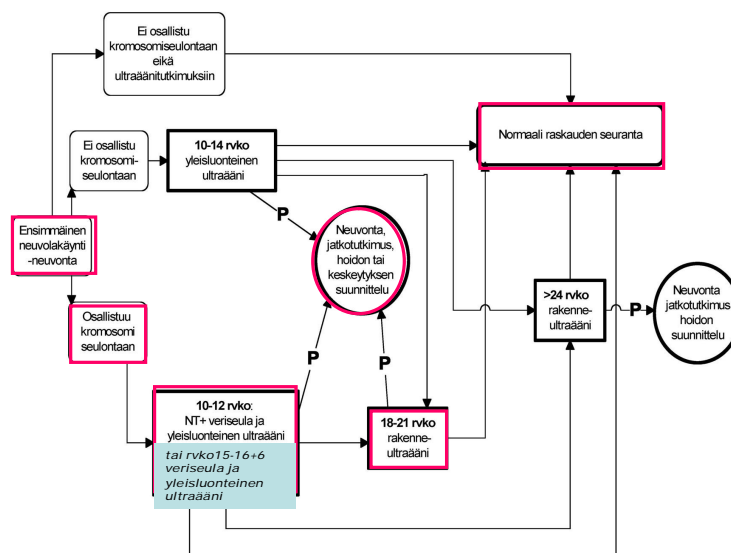
A **structural ultrasound scan during pregnancy weeks 18-21** is available for all pregnant women. In this examination the foetal structures are examined systematically with ultrasound. The examination is painless and harmless for the foetus.

In the ultrasound scan the pregnant woman gets to know about the findings already during the examination. Necessary further examinations are planned according to each discovered anomaly.

### Structural ultrasound scan after pregnancy week 24

From pregnancy week 24 the pregnancy can no longer be terminated based on the foetal anomaly. However, the discovery of structural anomalies can help when planning the care of the child. It is possible to participate in this ultrasound scan instead of the structural ultrasound scan which is carried out during pregnancy weeks 18-21.

### The screening path



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The offered screening programs are according to the Government Decree on Screenings nr 1339/2006.

Further information can be found at the website

<http://www.stm.fi/Resource.phx/publishing/store/2007/05/pr1178537602112/passthru.pdf>

There has to be a referral on all mothers (nurse or physician)

Contact information to the own maternity clinic:

The screenings are voluntary and you are therefore **not summoned** to them despite the referral! The pregnant mother can book an appointment for the screening

**examinations (Turku University Hospital) working days at 13-15 from the number 02 – 3131320.**

**other ultrasound units:**

phone:

time for booking an appointment:

**If the pregnant person does not want screening examinations (or the pregnancy weeks which are suitable for screening have been passed) she is to be offered ultrasound follow-up:**

**1. Ultrasound scan during early pregnancy can be done during pregnancy weeks 10 -  $\geq$ 14**

The number of fetuses and their heart rates, check of the duration of the pregnancy and so on.

**2. Structural ultrasound scan after pregnancy week 24**

The location of the placenta is checked as well as the content of the amniotic fluid, the foetal growth and structure considering the delivery.

**All pregnant women need to have a referral for these examinations as well.**

- **The examinations are voluntary, thus you are not summoned to them despite the referral**
- **The pregnant mother can book an appointment for ultrasound scans (Turku University Hospital) working days at 13-15 from the number 02- 3131320.**