

## **Anomalous result in foetal screening – further examinations**

This pamphlet is meant for families where a foetal anomaly is suspected, based on screening during the time of pregnancy.

**Participation in all further examinations is voluntary.**

## Ultrasound scan

- **Measuring the foetal nuchal translucency:** The foetal nuchal translucency is measured during pregnancy weeks 10+0 -12+6, and the result is combined with the results from the blood sample taken during the same pregnancy weeks after which a risk factor is calculated. If there is more nuchal translucency than normally or the risk factor is high, there is a small risk that the foetus has Down's syndrome = a chromosome disorder. The family is offered foetal chromosome examination, either through a placental sample or a sample of the amniotic fluid.
- **Ultrasound scan during early pregnancy:** during pregnancy weeks 10-14 an ultrasound scan is to be carried out, when the family does not wish a measurement of the nuchal translucency + blood sample examination. In this general ultrasound scan the duration of the pregnancy is ensured as well as the number of foetuses and whether they are all alive. Certain severe foetal anomalies can be seen in the examination. The family is informed about these, as well as about possible further examinations of which the family decides itself.
- **Ultrasound scan concerning structural anomalies either during pregnancy weeks 18–21 or after pregnancy weeks 24+0:** most of the significant anomalies in the foetal structure can be discovered in the examination: the most common anomalies are anomalies in the urinary tract or a heart defect. From time to time such anomalies are discovered, where it is not possible to determine their severity and significance based on an examination. In such cases the family is offered a new examination at the unit which is responsible for foetal studies and/or various further examinations. The point with these examinations is to find out the type and cause of the structural anomaly as well as to determine how the pregnancy will progress and what the prognosis is for the child.

### What does the abnormal finding in the ultrasound scan mean?

A possibility for the family to discuss the abnormal finding and its meaning with an experienced obstetrician and/or a geneticist is arranged, if needed also with a paediatrician or a paediatric surgeon.

Various structural anomalies can be found in the ultrasound scans, a part of these anomalies can be treated after the birth with for example surgery.

**The family, and ultimately the pregnant woman, decides how the examination results affect the course of the pregnancy.** Some will, after guidance and reflection, come to the conclusion that they will continue the pregnancy, and others will choose to terminate the pregnancy.

Already before the decision about further examinations is taken, it would be good for the family to think about what an abnormal result would mean for them. **The family also has a right to change their opinion at any stage of the screening and the further examinations.**

If a severe structural anomaly is discovered in the ultrasound scan, it is possible to terminate the pregnancy based on the finding with permission from The National Supervisory Authority

for Welfare and Health (Valvira) until the end of the 23 pregnancy week (24+0) (Act 239/1970 5 a §).

If a structural anomaly in the foetus is discovered during the 24<sup>th</sup> pregnancy week or thereafter, a possibility for the family to discuss the finding and its meaning with a physician, also with other experts if needed, is organized. **According to Finnish law, the pregnancy cannot be terminated at this point due to foetal illness.** Further examinations will be carried out if needed and also these are voluntary. The point with the further examinations is to find out about the type and cause of the structural anomaly, to find out about the progress of the pregnancy and the prognosis of the child. At the same time it is possible to get information that helps in the planning of the delivery and the care of the newborn.

## Chromosomal anomalies

### What does it mean to end up in the group at risk?

About five women out of a hundred (5%) get an abnormal result in the combined screening (measurement of the nuchal translucency + blood screening). This means that the probability that the foetus has Down's syndrome is greater than 1:250.

**Ending up in the group at risk during the screening does therefore not yet mean that the foetus has a chromosomal anomaly.** The chromosomal examination of the foetus through either the placenta or the amniotic fluid is offered as a supplemental examination. **The further examinations are altogether voluntary.**

### The further examinations

**Before the sample taking a well versed nurse or a physician will give you closer information about the risks, the sample taking, the results and the need for a day's sick leave. You can also ask further questions then.**

- **Examination of the placental sample:** A small amount of cells is taken from the placenta for the chromosome examination of the foetus. The sample is taken during pregnancy weeks 11–13 under ultrasound guidance with a needle puncture through the abdominal wall. The sample taking feels sort of like having a blood sample taken. 1-3 hours are to be reserved for the counselling and sample taking.

A risk for miscarriage is connected to the placental examination: about one of a hundred (1%) pregnancies is terminated due to the placental examination.

The chromosomal examination is ready 1-2 weeks after the sample taking depending on the method. The interpretation of the finding from the placental sample can sometimes call for a sample of the amniotic fluid as a further examination.

- **Examination of the amniotic fluid:** The foetal chromosomes can be examined also in the cells of the amniotic fluid. The examination of the amniotic fluid is the most often carried out during pregnancy weeks 14-16. The sample of the amniotic fluid is taken under ultrasound guidance with a needle puncture through the abdominal wall. The sample taking feels sort of like having a blood sample taken. 1-3 hours are to be reserved for the counselling and sample taking.

About one out of two hundred pregnancies are terminated due to the taking of an amniotic fluid sample (0,5%). The result from the examination of the amniotic fluid can be obtained after 2-4 weeks depending on the method used.

### **A normal chromosome result**

A normal chromosome result means that the number of foetal chromosomes is normal. The examination will not reveal small changes in the chromosomal structure.

Many illnesses and disabilities cannot be discovered with foetal examinations. The chromosomes can thus be normal, even though the child has some kind of illness or disability. It cannot with any screening method be stated with certainty, that the foetus is completely healthy.

### **An abnormal result in the chromosome examination**

Down's syndrome or some other chromosomal anomaly can be found in the placental examination or the examination of the amniotic fluid. If that is the case, a meeting with the geneticist or some other specialized physician is always arranged.

When the examination results are finished there is only very little time to decide about whether the pregnancy is to be continued or terminated. Some families decide after consideration that they want to continue the pregnancy and others decide to terminate the pregnancy. Also other structural defects in the foetus can govern the decision and not just the chromosomal anomaly. The family also has the right to change their mind at any point during the screening and the further examinations.

If a severe chromosomal anomaly in the foetus is discovered in the ultrasound scan, it is possible to based on this finding terminate the pregnancy with permission from The National Supervisory Authority for Welfare and Health (Valvira) until the end of the 23 pregnancy week (24+0) (Act 239/1970 5 a §).

**The pregnant woman always decides herself if the pregnancy is to proceed or to be terminated in such situations where a severe developmental disorder is discovered in the foetus.**

**Further information and peer support is available**

The result from the chromosome examination does not reveal anything about the degree of severity of the possible developmental disorder of the child. That will be determined over time. Even if remedial care is not available for many developmental disorders, both children and adults and their families can receive individually planned rehabilitation that supports the development. Families that get an ill or handicapped child often manage better than they estimated beforehand. A child always bring with it both happiness and challenges.

Further information and support can be obtained from health and welfare professionals already during the pregnancy, as well as from a number of associations for the disabled and their parents. Information and an open discussion lessen the worry and eases the decision-making. Also the staff at the hospital can help the family get started, by giving information about peer support families among other things. The varied course and recreation activities organised by associations, as well as publications and courses that help with readjustment can be useful for the families.

More information about disabilities, services, support and peer support families can be found for example from the following web sites: <http://mmm.verneri.net> och <http://www.kvtl.fi> samt <http://www.kehitysvammaliitto.fi>. Peer support can be had from <http://www.leijonaemot.org>.

## Word list

Placental sample	A sample that is taken during ultrasound guidance through the abdominal wall of the mother. With this sample, for example the foetal chromosomes can be studied.
Chromosome	A human normally has 46 chromosomes in each cell, that is 23 chromosome pairs (the chromosome pairs 1-22 and the sex chromosomes X and Y)
Sample of the amniotic fluid	A sample which is taken in ultrasound guidance through the abdominal walls of the mother. With this sample for example the foetal chromosomes can be examined.
Screening of the nuchal translucency	Measurement of the foetal nuchal translucency with ultrasound scan during pregnancy weeks 10-16+6
Pregnancy week	The time between the first day of bleeding in the last period until the moment the examination is done, the time is announced in full weeks + days (for example 12+3)
Risk factor	The estimation as calculated by a computer program of the risk to bear a child with Down's syndrome.

You can obtain further information also from the following places:

Contact information and phone numbers to Turku University Hospital

Contact information to your personal child health clinic: