



Screening for Down's Syndrome in early pregnancy – do you want to take part?

The purpose of this leaflet is to help you to decide whether you want to take part in screening for Down's Syndrome. Screening involves an ultra-sound examination and blood samples taken from the mother. Screening, even in the early stages of pregnancy, enables the detection of most of the foetuses afflicted with Down's Syndrome¹.

Screening is voluntary, as is participation in any subsequent examinations.

The purpose of screening is to identify those mothers in pregnancy with a higher probability of giving birth to a child afflicted with Down's Syndrome, and guiding them towards further examinations. Probability is calculated on the basis of the mother's age, the thickness of nuchal edema determined by ultra-sound examination, and the concentration of cell markers found in the mother's blood samples²). A computer programme has been developed for calculating the final probability. A positive screening result means there is an increased probability that the foetus is afflicted with Down's Syndrome.

Five percent of pregnancies screened produce a positive result. More than 80% of the pregnancies where the foetus is confirmed as having Down's Syndrome are detected through the screening process. It is worthwhile noting, however, that in most of the pregnancies producing positive screening results the foetus will prove not to have Down's Syndrome.

The current method is more effective than screening based only on the mother's age, and is therefore able to identify those pregnancies where the foetus possesses a higher risk of Down's Syndrome. Mothers who have been positively identified through screening can be directed for further examination if they so wish. Further examination involves chromosome testing of the foetus through samples taken from the placenta and amniotic fluid. If the foetus is confirmed as having Down's Syndrome, the family will be given the opportunity to prepare for birth of the afflicted child or to decide on pregnancy termination, permitted under Finnish law until the 24th week of pregnancy.

Screening through referral from a mother and child clinic

Screening of risk pregnancies is arranged at a maternity hospital outpatient clinic by means of referral from a mother and child clinic. An ultra-sound examination is performed first, between the 10th and 12th weeks of pregnancy, to determine the size of the foetus and thickness of nuchal edema. On the same day, or within a few days, a blood sample is taken from the mother to determine the concentration of cell markers. A computer programme calculates the degree of risk. Results are available one week after the samples have been taken.



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A negative screening result, or low probability of Down's Syndrome will be communicated to the mother by letter. This result means that the probability of Down's Syndrome in the pregnancy is lower than the probability relating to a 35-year-old pregnant mother based purely on age (a risk factor less than 1:250).

Approximately 95% of women screened obtain a negative result. A negative screening result, however, does not guarantee a completely healthy foetus.

A positive screening result, or increased probability means that the probability of Down's Syndrome in the pregnancy is higher than 1:250, and the family is given the opportunity of further examination, if they so wish, to determine the chromosome count of the foetus. **Mothers will receive confirmation of a positive screening result, by telephone or letter**, from the foetal research unit of the Department of Obstetrics and Gynaecology, which will include details relating to further examinations and the opportunity to make an appointment if required.

It is only possible to verify the presence of Down's Syndrome with certainty through examination of the cells of the foetus, obtained from a sample taken either from the placenta or from amniotic fluid. There is a small (less than 1%) risk of miscarriage attached to either method. It is therefore important that only those women identified as having an increased probability are submitted for this procedure.

As pregnancy progresses all those who attended early screening have the opportunity to undergo a second ultra-sound examination during weeks 19-21 in order to investigate foetal development. If the result is normal, the pregnancy reverts to normal monitoring at the mother and child clinic.

- 1) **Down's Syndrome** or trisomy 21 is a genetic disorder caused by the presence of an extra 21st chromosome. The reason for the extra chromosome is an abnormality in the composition of the ovum or spermatozoon which occurs already prior to conception. The incidence of children born with Down's syndrome in Finland is approximately one in six hundred. Individuals with Down's Syndrome have moderate developmental disabilities, typical facial characteristics, and in many cases congenital heart defects and an increased risk of developing leukaemia.
- 2) In pregnancies where the foetus exhibits Down's Syndrome, nuchal edema is enlarged and cell markers (HCG-beta and PAPP-A) are secreted by the placenta into the maternal bloodstream in concentrations differing from those in normal pregnancies.