



**Title:** Prenatal Screening: Down Syndrome, Neural Tube Defects, Routine Ultrasonography

**Agency:** GR, Health Council of the Netherlands (Gezondheidsraad)

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**Reference:** Publication No. 2001/11; full text (in Dutch) available at [www.gr.nl](http://www.gr.nl); 9 p. executive summary (English) also available separately

**Aim:**

- To synthesise current knowledge on methodological, psychological, and ethical aspects of various forms of antenatal screening for Down syndrome (DS) and neural tube defects (NTDs) and of routine ultrasound (US) examination during pregnancy.
- Report prepared in response to a request by the Minister of Health in view of evaluation of current policy in the Netherlands. This policy includes: age-based screening for DS, no screening for NTDs, no routine US. The gap is widening between policy and practice in this field.

**Results and Conclusions:**

- The report contains an extensive cost-effectiveness analysis comparing various strategies for antenatal DS screening. Comparison of screening based on the triple test with maternal age-based screening (on which DS screening in the Netherlands is currently based) shows that the former approach is much more cost-effective. A further conclusion from the analysis is that some newly developed risk-assessment strategies, eg, the combination of nuchal translucency measurement and a first trimester serum test, appear to be more cost-effective than triple-test screening. However, the scientific data in support of these alternative risk-assessment tests is not yet as strong in every respect as for the triple test.
- Other than the triple test, risk-assessment tests for DS in the first trimester cannot also be used to screen for NTDs. Ultrasound screening between 18 and 21 weeks has been proposed as an alternative, with the advantage of a one-step approach. It remains to be seen whether the sensitivity of this approach (for spina bifida) measures up to that of MSAFP screening; the costs will most likely be higher.
- The scientific literature does not provide sufficient hard evidence to support the use of US screening for structural abnormalities other than neural tube defects. Nor has routine US in pregnancy been shown to have any conclusive effect on clinical outcome measures. Given the limited nature of the research in this area, however, such an effect cannot be ruled out.
- No evidence shows that the psychological effects of risk-assessment screening are so serious as to make it unacceptable to offer such a test. Further research is needed into certain aspects, focusing on the quality of counselling and support. To date, scarcely any research has been conducted into the psychological consequences of risk-assessment screening in the first trimester of pregnancy (eg, nuchal translucency measurement).
- Antenatal screening for conditions such as DS or NTDs can be morally justified if the purpose is either to enable pregnant women and their partners to have the pregnancy terminated in the event of an abnormal result, or to prepare them for the birth of a handicapped child. The routine nature and the complexity of risk-assessment screening make it all the more challenging to meet the requirement of informed consent. The report advocates a phased approach.

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## Recommendations:

- Introduction of risk-assessment screening for DS, based on the triple test as the most tried and tested method, and organisationally the least ambitious, approach. The test should be offered to all pregnant women. Stimulation of research into newly emerging screening possibilities through regional trial population screening projects.
- Introduction of screening for NTDs. This should be based on the MSAFP test as long as triple-screening (including MSAFP) is the standard approach for DS. Where (in trial projects) first trimester DS screening is offered, it is advocated to offer US screening at 18 to 21 weeks as a test for NTDs, also as a trial screening program.
- As long as NTD screening is based on MSAFP, there is no scientific reason for offering routine US in pregnancy. However, the report recommends offering one dating scan to all pregnant women 'on pragmatic grounds'.
- The professional groups involved need to draw up guidelines for the various components and aspects of the screening program, including counselling. Educational programs are required for caregivers involved in the program.
- Prenatal screening for DS and NTDs needs to be subject to ongoing evaluation (ie, monitoring) to be conducted on a national level. Program results should be nationally recorded.

## Methods:

- Systematic review of published scientific literature. Expert Committee. Peer review of draft report.
- Cost-effectiveness analysis (appendix to the report) of strategies for prenatal DS screening.

## Further research/reviews required:

- Methodological/psychological/ethical: first-trimester screening for Down syndrome.
- Psychological: dynamics of decision making throughout the entire screening process; impact of false-negative outcomes of risk-assessment screening; consequences of abnormal US outcomes whose clinical significance is either unclear or uncertain.
- Ethical: informed consent for risk assessment screening.