Executive summary

Health Council of the Netherlands. Prenatal screening. The Hague: Health Council of the Netherlands, 2016; publication no. 2016/19.

Technological developments make it increasingly possible to detect abnormalities in the foetus early in the pregnancy. New tests that are more reliable and lead to less invasive tests are also available. What do these developments mean for the prenatal screening programme? The Minister of Health, Welfare and Sport put this question to the Health Council, which established the prenatal screening committee for this purpose. The committee drafted a preferred programme for prenatal screening based on the present state of scientific knowledge and the (ethical) criteria for screening.

Current programme and criteria for screening

The prenatal screening programme in 2016 is divided into two main parts. The first is a test for risk factors for the pregnancy, in the form of screening for PSIE (infectious diseases and erythrocyte immunisation – antibodies against non-endogenous red blood cells) and targeted screening of rhesus-negative pregnant women. The aim of this screening is to prevent health problems in the mother and child through timely treatment.

This advice mainly focuses on the second part of the programme which covers screening for congenital abnormalities. In the first trimester, it involves screening for Down's syndrome, Edwards' syndrome and Patau's syndrome and

in the second trimester, it involves ultrasound screening for structural abnormalities. This part is intended to provide ‘autonomous reproductive choices to the pregnant woman (and her partner): possibilities to decide themselves about the pregnancy. In many cases, the choice will be to carry the pregnancy to full term or terminate it. There are certain limitations within a screening programme. It must concern a serious condition and also in the future, there should not be screening for eye colour or talent, for example. The severity of the condition is not totally objective since not only the suffering of the child is considered but also the expectations and wishes of the parents. Trust in the pregnant woman and her partner is the starting point. Moreover, screening should not be performed for conditions that occur later in life to ensure that a child has an ‘open future’ and is not burdened with knowledge of a predisposition to a serious illness.

An important general criterion in screening is that the benefits must outweigh any harms for the participant. This means that the following criteria apply:

- it must concern a significant health problem
- screening must have meaningful outcomes (health benefit or options for action)
- there must be a reliable and valid screening method with safe-guarded quality
- participation in the screening is based on a voluntary, informed choice
- screening must make efficient use of resources (including cost effectiveness, fairness and accessibility).

Moreover, screening for congenital defects must take place as early in the pregnancy as possible, leaving sufficient time for follow-up diagnostic investigations and the choice to continue or terminate the pregnancy. The burden imposed by a possible termination increases during the pregnancy.

Screening for chromosomal abnormalities with NIPT

An important development in the screening for Down’s syndrome, Edwards’ syndrome and Patau's syndrome is the non-invasive prenatal test (NIPT). In the current programme, screening takes place by means of the combined test that indicates whether there is an increased risk of a foetus with one of these disorders. Since 1 April 2014, in the context of a scientific study, women who are at increased risk may opt for NIPT instead of directly undergoing an invasive test (chorionic villus sampling or amniocentesis). There is also a study under

preparation in which women can choose between the combined test or NIPT as the first screening test.

The committee advises introducing NIPT as a screening test instead of the combined test since NIPT performs better. NIPT almost always detects Down's syndrome and has a much lower false-positive rate. NIPT also performs better for Edwards' syndrome and Patau's syndrome than the combined test. NIPT's better performance means that less invasive follow-up testing is needed. Another advantage is that NIPT is not limited to a specific time frame in pregnancy.

It is sometimes argued in the public debate that the introduction of NIPT would reduce the acceptance of Down's syndrome. The committee is in favour of introducing the test because the aim of the screening is not to prevent the birth of children with a disorder but to offer meaningful reproductive choices to pregnant women (and their partners). The committee believes that the decision on whether to participate in the screening and the decision on what to do with the results is a personal one that pregnant women must make themselves. The committee believes that good facilities for people with a disability are a prerequisite for prenatal screening for abnormalities, giving prospective parents the choice of raising a child with a disability.

For the time being, NIPT should only screen for Down's syndrome, Edwards' syndrome and Patau's syndrome. In future, other genetic disorders could also be detected with the test.

Screening for structural abnormalities with ultrasound (SEO)

Research shows that many of the very serious defects that are now detected in the '20-week ultrasound' can already be detected at the end of the first trimester of the pregnancy. The advantage of an early ultrasound is that there is more time for follow-up diagnostic investigations and for a decision on whether to carry the pregnancy to full term. The disadvantage is that an additional ultrasound leads to higher costs. Moreover, if there are many false-positive or unclear findings, an early ultrasound can lead to more anxiety rather than more reproductive choices. The results of scientific research are promising but it is not yet sufficiently clear what the benefits of the early ultrasound are and how the advantages compare

with the disadvantages. Therefore, the committee recommends doing a nationwide scientific study on including an ultrasound in the programme starting at 12 to 14 weeks of pregnancy. This means that pregnant women can opt for such an early ultrasound within the context of a study that will look at how many disorders can be detected, how many findings appear to be false alarms, how long it takes before there is clarity for the pregnant woman and how pregnant women experience an early ultrasound.

According to the committee, the current ultrasound examination in the second trimester (the 20-week ultrasound) should be maintained since not all structural abnormalities can be seen at the end of the first trimester. This screening should provide better registration than at present. The committee recommends carrying out this ultrasound starting at 18 weeks and no later than 20 weeks to allow sufficient time for follow-up diagnostics within the period of lawful termination of pregnancy.

Selection and treatment of risk groups not yet possible

At present, the committee sees no reason to make use of risk stratification within the programme by selecting women with a high risk of a certain complication and offering preventive treatment. As yet scientific knowledge on the effectiveness is too limited. However, various studies in this field are currently underway. In future, screening may occur and treatment may take place for women with a high risk of pre-eclampsia (toxaemia), foetal growth retardation, spontaneous preterm birth and gestational diabetes. At this time, there is already strong evidence that intake of sufficient calcium may reduce the occurrence of pre-eclampsia. Many women do not manage the recommended intake of 1,000 mg per day. Therefore, the committee recommends discussing the already existing dietary advice on calcium, alongside the other preventive advice during the first midwife consultation.

More attention to counselling

Good counselling is important to ensure that pregnant women can decide voluntarily and are well-informed about whether to participate in prenatal screening. The committee believes that the current practice of counselling needs to be improved. For example, sufficient time is not always taken, counselling is

not always tailored to the needs of the pregnant woman and her partner and some pregnant women do not have sufficient support in the decision.

The committee believes that care providers should not have to ask for permission before giving information since information is needed to decide whether or not to participate. The information should also be separated from the counselling. During the counselling session, the care provider can determine whether the pregnant woman has understood the information and assist with the decision, if desired. Finally, the committee recommends presenting the screening as a coherent whole, not as individual parts.

The preferred programme

The committee considers it crucial that the accessibility of the programme is guaranteed. In the present programme, pregnant women pay themselves to screen for Down's syndrome, which appears to be an obstacle for certain groups. The committee also advises changing the name of the screening for Down's syndrome to screening for chromosomal abnormalities. That better reflects the content of the screening.

The programme preferred by the committee is as follows.

1 Screening for chromosomal abnormalities, currently limited to Down's syndrome, Edwards' syndrome and Patau's syndrome, with NIPT starting at ten weeks of pregnancy. In the event of an abnormal result, a follow-up examination in the form of chorionic villus sampling or amniocentesis is needed for greater certainty.
2 An ultrasound to detect structural abnormalities starting at 12 to 14 weeks of pregnancy within the context of a nationwide scientific study.
3 An ultrasound to detect structural abnormalities starting at 18 to 20 weeks of pregnancy. The results of this ultrasound must be monitored better.

According to the committee, this programme best reflects the latest scientific developments and offers the most options for action. A possible disadvantage is the risk of increased anxiety among pregnant women since there is an additional screening moment. That makes the scientific research important.

Future developments

For the near future, the committee sees several developments that could be relevant for the screening. Extending the screening with NIPT to other (sub)chromosomal disorders, once the test for them is validated, would be worth considering. The committee also expects that risk stratification and/or preventive treatment of complications may be part of the programme within five to ten years.

The committee recommends considering prenatal screening as part of a cohesive chain of screening to be achieved around pregnancy and birth. It endorses the conclusion of the previous advisory report on preconception care that, aside from prenatal and neonatal screening, preconception care can also make a significant contribution to the health benefits and reproductive choices concerning pregnancy and birth.