

Prenatal diagnosis: the Ethics

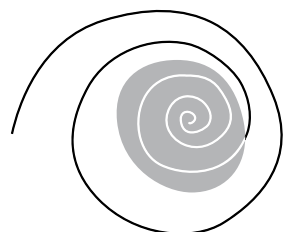
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Preface

It is not obvious that all advances in genetic technology should be put into use in practice. This opinion from the Swedish National Council on Medical Ethics (SMER) addresses the issue of what we should do, not just what we can do, in the field of prenatal diagnosis. These questions will be discussed here from a socioethical and an individual ethical perspective:

- What prenatal diagnosis should be provided by publicly funded healthcare?
- Is there any prenatal diagnosis that should not be allowed?
- To what extent can, or should, the woman influence the choice of prenatal diagnosis?
- What relevant facts must women take into account when making decisions on prenatal diagnosis?

Prenatal diagnosis brings to the fore ethical considerations involving value conflicts and conflicts of interest. In particular, there are the implications of prenatal diagnosis for our view of, and respect for, the dignity of the human being. These issues should therefore be discussed in society at large after medical experts have presented the state of current knowledge and the available technical possibilities.

The issue of how and to whom prenatal diagnosis should be offered can also be looked at from an ethical perspective, since these questions concern key values such as fairness and priority, as well as self-determination.

If we are to have broad public debate on these issues, we need openness and transparency about the development and introduction of new technologies and methods. Such an approach also serves to enhance the legitimacy of medical progress. Consequently, it is interesting to discuss what institutional arrangements are likely to promote democratic values in decisions about prenatal diagnosis.

Prenatal diagnosis is one of the issues that SMER is responsible for monitoring. This opinion does not attempt to provide guidance to individuals in their decisions: decisions of this type must be made by individual women themselves after consultations with health care personnel. The following analysis focuses on aspects of testing and treatment/action that the pregnant woman and her partner need to consider. The couple will consider many of these issues jointly; their interests will usually coincide, but not always. In cases where the couple's interests diverge, however, it is always the pregnant woman who makes the final decision. In view of this, the expression 'the woman' is used throughout, even though the decisions described also involve her partner.

The opinion is partly based on the analysis carried out by Dr Niklas Juth of the Department of Philosophy at Göteborg University, acting as secretary of a working party consisting of Professors Göran Hermerén, Christian Munthe and Jan Wahlström. This is a slightly condensed version of the original opinion.

Daniel Tarschys
Chair

1. Prenatal diagnosis in Sweden

The term 'prenatal diagnosis' covers numerous different methods used to obtain information about the health of the foetus. These include images produced by ultrasound screening, tissue samples taken from the foetus during amniocentesis, and samples from the placenta. The methods of analysis can vary from the analysis of ultrasound images by an experienced midwife or doctor to chromosome analyses and molecular-genetic examination of tissue samples.

Today, it is technically possible to use prenatal genetic screening to identify all the hereditary diseases and characteristics that an individual may develop in the future. Such analyses are currently very costly, however. A key issue in deciding what prenatal diagnosis to provide is therefore how serious a disease must be for prenatal diagnosis for it to be made available. For example, it might be considered that there is greater justification to provide genetic screening for diseases that debut during childhood, such as Tay-Sachs, than to carry out screening for diseases that do not surface until middle age. It might also seem more reasonable to provide tests for serious diseases with a high likelihood of development rather than less serious ones with a low likelihood of development. It is also usually said that the primary objective should be to diagnose diseases that can be treated.

The following terms are used in this opinion:

- Prenatal diagnosis refers to all methods used to examine a foetus.
- *Simple ultrasound scan* refers to examinations whose objective is to determine the length of pregnancy and the number of foetuses. Signs of impairment or disease may also be discovered during such an examination, leading to further examination.
- *Detailed ultrasound scan* refers to examinations that have the additional objective of looking for impairment or disease in the foetus.
- *Genetic prenatal screening* refers to analysis carried out on tissue samples consisting of cells from the foetus.

Ultrasound and serum screening do not involve any risk of physical injury to the foetus or the mother-to-be. These methods are used to discover certain types of foetal defects, but can also be used to calculate the risk that the pregnant woman may have a child with a chromosomal abnormality such as Down Syndrome.

Sampling that involves use of foetal cells for analysis is an invasive procedure and carries a miscarriage risk of about 1%.

In future, it might be possible to look for most known hereditary diseases and many abnormalities – whether in the foetus or which may develop later in life – thanks to detailed ultrasound diagnosis and genetic prenatal screening. For some of these diseases, there is a high risk that they will develop while for others

there is a low or undetermined risk. Some can be treated, but effective treatment is so far lacking for most.

When prenatal diagnosis was introduced in Sweden in the late 1960s and early 1970s, there were three main motives for it: 1) the woman's health, quality of life and self-determination, 2) the child's health and quality of life and 3) economic considerations.

1. The pregnant woman may have various reasons for wanting prenatal diagnosis. Prenatal diagnosis enables the woman to choose whether she thinks she could cope with and manage to look after a sick or impaired child. Prenatal diagnosis can reduce general anxiety about having an impaired child. A further reason for prenatal diagnosis is that the woman can prepare herself to look after an impaired child in the best possible way.

2. The child-to-be can benefit, since impairments to the child-to-be can be avoided by treatment before, during or after delivery. However, as yet this is only possible in a very limited number of cases. Prenatal diagnosis can also be said to prevent abortion in some cases: a woman who knows that she has a higher risk of having a sick child may choose to undergo prenatal diagnosis, and if the foetus proves not to have the impairment that the woman fears, she can abstain from abortion.

3. One result of the discussion that began in the 1970s is that the economic motive for prenatal diagnosis is nowadays not deemed ethically acceptable. Prenatal diagnosis should not be assessed in terms of the potential financial 'savings' to be made by aborting foetuses with a higher risk of functional impairment or disease.¹ Nor can the potential benefits of prenatal diagnosis in the shape of greater quality of life and autonomy be measured in monetary terms.

Sweden's Genetic Integrity Act (2006:351) which came into force on 1 July 2006 sets out basic regulations on the use of prenatal diagnosis. Under this Act, all pregnant women are to be provided with general information about prenatal diagnosis. A pregnant woman with a medically diagnosed elevated risk of having a child with an impairment is to be provided with further information about genetic prenatal diagnosis. The Act stipulates that the decision to undergo prenatal diagnosis or not is made by the woman, in consultation with her doctor, after she has been provided with information.

1 See Kommittén om genetisk integritet (Committee on Genetic Integrity) (SOU 2004:20, p. 266), *Vetenskapsrådets konsensusuttalande om tidig fosterdiagnostik* (Swedish Research Council Consensus Opinion on Prenatal Diagnosis) (2001, pp 25–26) and SMER's "Yttrande om införande av en ny fosterdiagnostisk metod" (Opinion on the introduction of a new method of prenatal diagnosis) (20 December 2004, p. 3).

After prenatal diagnosis, the pregnant woman is to be given all the information about the health of the foetus that was obtained by the examination. Information about the foetus that does not have any bearing on its state of health may only be given if the woman asks for it.

The result of a prenatal test may cause the parents to choose to terminate the pregnancy by abortion. Under the Swedish Abortion Act (1974:595), abortion after the 18th week of pregnancy may only be carried out with the consent of the National Board of Health and Welfare. Such consent may only be granted where there are exceptional grounds. Until the end of the 18th week of pregnancy, a woman may have an abortion without having to state her reasons.

2. Points of departure for ethical analysis

Summary

- There are two main traditions in ethics which provide a strategy for solving conflicts of interest. These are the ethics of consequences (consequentialism) and the ethics of duty. Both stress the importance of principles in ethics. Alongside these traditions there is situationalism, which emphasises that each case is unique and stresses the importance of the situation.
- The principle of human dignity is a fundamental ethical principle which is important when analysing which type of prenatal diagnosis is to be offered to a pregnant woman.
- Other important ethical principles to be taken into account in an ethical analysis of prenatal diagnosis are perceptions of human nature, integrity, autonomy/self-determination, informed consent, quality of life and the principles of need and solidarity.
- The moral status of the fertilised egg is another point of departure. In previous opinions, SMER has held that the genesis of human life is a process in which the fertilised egg is a life in the making with a certain moral status. This moral status gradually increases as the foetus develops. At the point in time when the foetus can survive outside its mother's body, its moral status becomes human dignity.

2.1 Background

The task of ethics is to systematically examine and analyse the standards and values that can be used to defend or criticise the actions of people or interested parties (such as organisations or groups) in the matter being analysed. Morals are reflected in the concrete acts of individuals; the failure to act in certain situations also reveals the morals of those concerned. Ethical issues become particularly prominent when the subject involved has to do with human dignity and perceptions of human nature, and when integrity, fairness and solidarity are at issue.

2.2 Tools for ethical analysis

Since antiquity, there have been two main traditions in ethics which provide a conceptual framework and a strategy for resolving conflicts of interest. One is the *ethics of consequences*, the main idea of which is that the consequences of one's actions determine what is right, wrong, or one's duty. This presupposes that it is possible to make a meaningful comparison of the consequences of the course of action chosen in a certain situation with the results of other possible options.

The other main tradition in ethics is sometimes called *duty ethics*. The main idea here is that there are some duties or rights that apply regardless of the consequences of the alternative actions open to

a certain person in a given situation. The point of departure is that people have a certain dignity, certain rights and should be treated with respect. The German philosopher Immanuel Kant, for example, stressed that people should always be treated as an end in themselves, and never simply as the means to an end.

There are deep-seated conflicts between these two prevailing ethical theories. Various attempts have been made to bridge these conflicts. One of the most interesting is the theory presented by the British philosopher Richard Hare in 1993, that moral thinking takes place on two levels. At what Hare calls the *intuitive level*, we use rules of thumb and principles of the type that duty ethicists talk about. When making decisions in everyday life, under time pressure and using incomplete information, we stick to certain rules since experience has shown that this generally works well.

However, when we are not relying on our intuition but are choosing principles and then justifying this choice, or choosing between principles that are in conflict with each other, the situation is different: careful analysis of the situation is required. At this level, which Hare calls *critical thinking*, utilitarianism is preferable in Hare's view. Here, rules and principles must be assessed in accordance with the extent to which they have good consequences in the long run, for example, the extent to which they help satisfy the interests of those involved.

One practical approach might be to try to formulate some general principles which appear credible. We can then examine the extent to which their application on a case-by-case basis agrees with our intuitive ethical convictions. If there is a conflict between the principles and our intuitions, we must critically examine both. We should ask whether the principles need to be modified so as to fit our intuitions. We should also ask whether intuitions that come into conflict with some principle, on closer reflection, need to be revised or abandoned. By a process of mutual adaptation of principles and intuitions, we can then arrive at a reflective equilibrium in which our principles and basic ethical intuitions are in harmony with each other.

In this opinion, SMER has primarily used this method of ethical analysis to clarify problems and as an aid towards a solution that can be broadly accepted.

Use of this method requires that interests be weighed against each other. We must take into account the positive and negative consequences of various alternative courses of action, as well as long-term and short-term consequences, and not forget the interests of any of the groups involved. What might be a positive consequence for one person might be negative for someone else. As in other areas, there may be winners and losers, and they are often different people or groups whose interests must be weighed against each other.

2.3 Some basic ethical principles and concepts of importance for prenatal diagnosis

2.3.1 Perceptions of human nature

A perception of human nature is often thought to be the same thing as human dignity, but it is important to distinguish between the two. The concept 'perception of human nature' is larger and more wide-ranging than human dignity. The fundamental principle in Western culture of the equal worth of all can be part of some perceptions of human nature but not all, such as Nazi or other racist views of humankind.

2.3.2 The principle of human dignity

The principle of human dignity is a fundamental ethical principle with an integral place in duty ethics, particularly in the Kantian tradition. SMER has discussed the issue of human dignity in a large number of contexts. In its 1993 publication *Det svårfångade människovärdet* (Human dignity: an elusive concept), SMER stated that:

- 1 Human dignity is attached to existence, not to functions or characteristics
- 2 Human dignity is an axiom and cannot be proved by empirical study or tests
- 3 Human dignity means that all people have certain fundamental rights that must be respected, and nobody takes precedence in this regard
- 4 Human dignity does not rule out the evaluation of people's characteristics, suitability or qualifications in a given context.

It has been asserted that human dignity² could be under threat because prenatal diagnosis impacts negatively on the perception of human nature, as this activity is the expression of an outlook which does not accord the same dignity to all.³ This is true in that the discovery of an impairment or disease in the foetus often leads to termination of the pregnancy. Prenatal diagnosis could thus be regarded as a means of 'discarding' certain individuals who are deemed less desirable than others due to their disability or disease. The key issue is whether prenatal diagnosis violates the principle of human dignity, and if so, how the activity should be organised so as to preserve human dignity.

Greater discrimination against the disabled?

Fears are sometimes expressed that prenatal diagnosis is not just an expression of negative attitudes towards sick people and individuals with disabilities, but that the activity itself reinforces these attitudes, exacerbating the stigmatisation of sick and disabled people and discrimination against them.

Another interpretation is that the explicit or implicit objective of prenatal diagnosis is to avoid certain

types of people. That is why it is a good thing if information about the purpose of prenatal diagnosis is made clear: that it is not a matter of systematically discarding some categories of people. This concern is an argument against screening programmes for diseases and impairments for which there is no treatment. It is also an argument in favour of society providing good support for people with disabilities, so that carrying the pregnancy to term is a realistic alternative for a woman when prenatal diagnosis reveals an impairment or disease.

2.3.3 Autonomy or self-determination

One of the fundamental concepts in medical ethics is autonomy, or the right to self-determination. According to this principle, each person has the right to make decisions about their own life in accordance with their own view of what a good life is. An individual must, as a matter of principle, be entitled to choose what she wants to know, or does not want to know, about the risk of future diseases in herself and her future children. The individual should thus decide for herself whether to accept the offer of prenatal diagnosis, and must also be free to act on the result.

Autonomy can be interpreted in a number of ways. It is important to distinguish between autonomy as an ability (a psychological term), as a right (a normative term) and as a value (an axiomatic term), the purpose of which may be (a) instrumental – valuable as a means of achieving something else – or (b) intrinsic, i.e. valuable in itself. Each of these terms can in turn be defined in various ways.⁴

Autonomy has traditionally not been regarded as a value to be promoted, but rather as something that gives rise to 'negative rights', or restrictions on how one may be treated by others. The idea is that if an individual is adult and capable of making decisions, then others may not prevent the individual from making independent decisions and acting on them, at least insofar as the individual does not actively violate someone else's rights⁵ or injure someone else.⁶ Under the Swedish Health and Medical Services Act, people are entitled to know what their treatment involves, how risky and painful it will be and the probable consequences of undergoing or not undergoing

2 See also Article 1, 'Human dignity', in the European Bioethics Convention.

3 This has been asserted in the international debate (see Asch, 1999) as well as in the Swedish debate (see Gustafson, 1980).

4 See e.g. Fleischhauser and Hermerén, Chapter 6.4, page 372 et seq (2006).

5 As envisaged by Locke, 1689, and Nozick, 1974.

6 As envisaged by Mill, 1859 and Glover, 1977.

7 See Beauchamp and Chilss, 2001, pp. 176–177; Juth, 2005, p. 53, Chapters 2 and 3, passim; Tännsjö, 1999.

the treatment, after which they have the right to say yes or no to it.

Recently, it has become more common to assert that various medical activities should also strengthen patient autonomy. This applies to activities such as genetic testing and assisted fertilisation, but also to prenatal diagnosis. Autonomy is seen as a goal which the health services must achieve, or a value which they are to reinforce. The idea is that prenatal diagnosis can aid the woman in planning her life. Generally, having more information gives the individual a better basis for making decisions in accordance with her own wishes and plans. This is of course also the case with family planning. A woman who has previously had a child with a certain impairment may find the information provided by prenatal diagnosis helpful.

This need not be the only purpose of prenatal diagnosis, however. A woman may be prepared to welcome a child even with an impairment, and in that case her motive for prenatal diagnosis may be that she wants time to prepare for the arrival of such a child. In all these cases, prenatal diagnosis can help a woman make her own decisions about her own future and that of her family. In these contexts, then, we are not just talking about autonomy as something that is to be respected, but autonomy as a goal to be achieved or a value to be strengthened. This idea is newer and more controversial than the traditional idea that the goal of autonomy is the health and well-being of the patient.⁷ Objections to the idea that the health services should promote the autonomy of the patient stem from the fact that such a goal can come into conflict with other ethical values. Possible conflicts might involve human dignity, the integrity of other individuals and prioritisation aspects.

The fact that individual self-determination is accorded great importance means that potentially life-changing decisions are also left in the hands of the individual. This is very much true of decisions associated with prenatal diagnosis. Information provided along with the offer of prenatal diagnosis must not, however, be understood to mean that health care staff are to provide information and then leave the woman alone to process it in order to arrive at a decision. 'Information' in this context is not the right word; it is more a question of a communicative process in which health care staff and the woman together look for the decision that is right for her. When the woman, along with health care staff, has arrived at the decision which is right for her, the staff must continue to support her in her decision regardless of their own view. This is known as a non-directive approach.

In this context, it should also be stressed that the woman's autonomy is of course limited by the framework society has established for prenatal diagnosis.

2.3.4 Informed consent

The requirement that a medical procedure is to be preceded by informed consent is extremely important in medical ethics. If a person is to act independently, make independent choices and take moral responsibility for her actions, she needs to have had access to factual information about the conditions and consequences of different courses of action, to have understood this information and to have given her consent on the basis of it. It is equally important that the patient has understood what it means to abstain from knowledge about something, for example their genes.

The difficulty of arriving at positions based on free and informed consent should not be underestimated. Some factors that may cause difficulty are expectations, use of language and the social and psychological situation.

2.3.5 Integrity

Integrity comes from a Latin word which means untouched, whole. The concept is linked to value and dignity, and refers to the intrinsic worth of every human being. A distinction can be made between physical and mental integrity. In physical integrity, the 'whole' referred to is the body. Nobody is entitled to examine someone else's body without that person's consent. Mental integrity refers to the individual's complex sum of values, ideas, views and wishes, as well as their religious beliefs and mental life.

Integrity is also intimately linked to identity in that each person is unique and that it is this which makes human identity important. Personal integrity means inviolability: the right not to be violated. The hallmark of integrity is that it does not cease to exist simply because the individual is unable to assert it. Integrity differs from autonomy in this respect. The ability to make decisions that are in one's own long-term interest may vary through life, but the right to integrity does not vary in the same way.

Genetic integrity is included in personal integrity and is particularly worthy of protection because information about genes and hereditary disease says a lot about who we are and what we will become. This means that the results of detailed ultrasound examination and genetic prenatal diagnosis are particularly worthy of protection.

2.3.6 Quality of life

Quality of life can be defined in many different ways. When talking about prenatal diagnosis, it is useful to distinguish between the terms 'health' and 'quality of life'. Health can be defined in quantitative terms, for example in terms of life expectancy, while quality of life is a qualitative concept which is ultimately based on subjective assessments. Because there are two main stakeholders – the expectant mother and the expected child – and because the health and quality of life of both are affected, ethical analysis is easier if we use a two-by-two table. Filling in positive and

negative factors in the table can yield a valuable resource in an ethical analysis.

	Life expectancy	Future quality of life
Expectant mother		
Expected child		

The different squares are filled in; for example, whether the expectant mother's life expectancy will become longer or shorter. Squares are filled in based on the conclusions of the person doing the analysis.

2.3.7 The principle of need and solidarity

A number of need principles can be identified, depending on which needs are being met. In this context, it is reasonable to focus on health-related needs. The Swedish Health and Medical Services Act states that the aim of the health and medical services is to assure the entire population of good health and care on equal terms. This is an expression of solidarity. Solidarity does not just mean equal opportunities to receive care, but an ambition that the outcome of care will be as equal as possible given the conditions, i.e. that everyone will achieve the best possible health, life expectancy and quality of life.

The principle of need and solidarity is part of the ethical platform which provides the basis for the guidelines for priority-setting in health care adopted by the Swedish Riksdag (Parliament). The principle of need and solidarity is formulated as follows: "Resources should be devoted to those areas where there is greatest need". The need and solidarity principle is a further development of the fairness principle. The import of the principle of need and solidarity is that if priorities must be set between effective measures, then more care resources should be given to those who are most in need, those with the most serious diseases and those with the poorest quality of life.

2.3.8 The moral status of the fertilised egg

A further key issue in analysing human dignity in prenatal diagnosis contexts is that of the moral status of the fertilised egg.

There are diverging views in society about what moral status should be accorded to a fertilised egg. Three different positions can be distinguished:

- 1 Human life begins at conception and the fertilised egg has full human dignity, i.e. a right to protection and an unconditional right to life.
- 2 The genesis of human life is a process in which the fertilised egg is a life in the making and has a certain moral status. This moral status increases gradually as the foetus develops. At the point in time when the foetus can survive outside its mother's body, its moral status becomes human dignity.
- 3 The fertilised egg has development potential but has no moral status in itself.

SMER has dealt with this issue in previous opinions and documents, and has agreed with the second position; this position is consistent with current abortion legislation and also with how prenatal diagnosis has so far been applied in Sweden.

3. Why perform prenatal diagnosis?

Summary

- Prenatal diagnosis can be viewed from a number of perspectives. For the woman, an important issue might be to be able to independently decide whether she feels able to take care of an impaired or sick child. From the perspective of society, it is a matter of offering prenatal diagnosis to pregnant women within a given framework and in a way that safeguards women's self-determination.
- There is a link between prenatal diagnosis and abortion, since the result of prenatal diagnosis may present the woman with the choice of whether or not to have an abortion.
- The woman's quality of life, in terms of health and psychological well-being, can be advanced by prenatal diagnosis, but can also be damaged, above all if she receives knowledge that she did not request. Information ahead of the decision on whether to have prenatal diagnosis is therefore important.
- For the foetus, it is doubtful whether health, life expectancy or expected quality of life can benefit at all in cases where there is no treatment for the diagnosed disease.
- The motive for prenatal diagnosis can also help answer the question of which type of prenatal diagnosis should be offered.

3.1 Stakeholders and values

The overarching purpose of prenatal diagnosis can be described from a number of different angles. From the point of view of society, the aim is to offer information about prenatal diagnosis to the pregnant woman within a certain framework, to allow her to take a position on whether she wants to undergo any type of prenatal diagnosis. Because the result of prenatal diagnosis can lead to the woman's having to decide whether she wants to have an abortion, it is important that she is given the necessary information about possible consequences if she accepts the offer of prenatal diagnosis. Swedish abortion legislation gives the woman the right to make her own decision about terminating the pregnancy up until the end of the 18th week of pregnancy. Because the law gives the woman this right, a woman who is informed after prenatal diagnosis that the foetus is sick or impaired can make her own decision on whether or not to have an abortion.

For the pregnant woman, the aim of prenatal diagnosis might be a general health check. There is no ethical controversy here because it benefits both the pregnant woman and the foetus. This applies to the health checks carried out at prenatal clinics and to the simple ultrasound scan which focuses on establis-

hing the duration of pregnancy and how many foetuses the woman is carrying.

Detailed ultrasound examinations, which involve going through checklists to discover foetal impairment or disease, as well as all genetic prenatal testing designed to find abnormalities and disease in the foetus, can, on the other hand, have certain ethically controversial aspects. The most important motive, however, is to give the pregnant woman the opportunity to make an independent decision on whether she wants an abortion because she feels unable, on the basis of her experience and life situation, to look after a child with the impairment or disease that prenatal diagnosis has disclosed in the foetus.

The following questions should be answered for a more in-depth analysis:

- What are the benefits of prenatal diagnosis?
- Who might benefit from carrying out prenatal diagnosis?

The most usual response is that prenatal diagnosis can benefit the pregnant woman and the child-to-be.⁸ The woman's self-determination benefits from prenatal diagnosis, since she can gain a better basis for a decision on whether or not to carry the pregnancy to term when prenatal diagnosis shows that the foetus is impaired or diseased. The issue involved here may be whether or not the woman feels able to look after an impaired child. The woman's quality of life also benefits in terms of mental well-being (reduced anxiety) and health. In the child's case, life expectancy and quality of life are the main areas of potential improvement if an identified disease or impairment can be prevented or treated.

Many fundamental values other than self-determination are important in the ethical evaluation of prenatal diagnosis. A number of these have been discussed in Chapter 2. Quality of life and self-determination are well-defined concepts in medical ethics.⁹ If these values are promoted, if quality of life is improved and if self-determination benefits from certain types of prenatal diagnosis, then the activity can be justified.

3.2 Quality of life and prenatal diagnosis

Expected quality of life can be improved for the pregnant woman in that prenatal diagnosis improves the chances of preparing for the birth so that neither the woman nor the child suffers injury. Psychological well-being can also benefit. More precise information about what can happen to the foetus and the woman herself during and after pregnancy can reduce some of the anxiety caused by uncertainty about these matters. Prenatal diagnosis can also reduce women's wish to have abortions because of general concern about foetal impairments.

8 SOU 2004:20, and The Nuffield Council, 1993.

9 Beauchamp and Childress, 2001, Chaps. 3 and 5; Fleischhauer and Hermerén, 2006.

For the pregnant woman, complications in the shape of miscarriage due to genetic prenatal diagnosis can in the first instance affect quality of life. Anxiety provoked by questions about prenatal diagnosis might impact negatively on the quality of life of the pregnant woman undergoing diagnosis. For pregnant women who decline the offer of prenatal diagnosis outright, the fact that the issue was raised at all might boost anxiety and thus lower quality of life. Quality of life can also be reduced if a risk assessment using nuchal translucency diagnosis and biochemical markers shows that a pregnant woman has a low risk of having a child with chromosomal abnormalities, she therefore refrains from further prenatal diagnosis, and it later turns out that despite this low risk she still has a child with a chromosomal abnormality. The quality of life of such a woman may be negatively affected.

The issue of the quality of life of the expected child is more complicated, however. If there is treatment available for the diagnosed impairment or disease, prenatal diagnosis can benefit the quality of life of the child-to-be. This primarily applies to the life expectancy of the child, however, since few diseases which can be identified at the foetal stage can be treated at that stage at present. Symptoms of some conditions can, in rare cases, be treated if and when the child has actually been born. There is seldom any medical advantage to the unborn child in diagnosing the disease at the foetal stage.

In the international and national ethical debate about prenatal diagnosis there is some discussion of conditions which entail a very short and painful life for the child itself and for which no real cure or alleviation is possible.¹⁰ One example of this is Krabbe disease, which results in cramping and discomfort on being touched only months after birth. The disease usually results in death around the age of one. In cases like this, it has been argued that it would have been better for the child never to have been born at all. If we accept the view often applied in health care that it is the individual's own experience of life that must be decisive, then this way of thinking may perhaps be accepted.¹¹ The argument is so controversial, however, that it cannot serve as a basis for policy decisions on prenatal diagnosis. Conditions such as Krabbe disease are very rare.¹²

Despite the fact that arguments of this type are highly controversial, they should still be mentioned,

for two reasons. Firstly, because they often come up in discussions about what type of prenatal diagnosis is justified.¹³ It has sometimes been asserted that the limit for defensible prenatal diagnosis is 'a life not worth living', at least when there is no treatment for the foetus or child-to-be. Secondly, because this argument clearly shows how prenatal diagnosis raises fundamental and central ethical questions. If prenatal diagnosis is to be defended by referring to the quality of life of the child-to-be, then we cannot duck the issue of what quality of life is, and whether it is self-evident that a life's 'quality' is always good. And thirdly, an important issue is who gets to decide what lives are worth living? And what criteria or principles do we use to approach such issues?

We can envisage a scenario in which a woman who has prenatal diagnosis and thereafter has an abortion because the foetus proved to have an impairment later gets pregnant again and has a healthy child. If she had not chosen to terminate the first pregnancy, she would probably not have had the second child. The result of prenatal diagnosis was that another child was born instead of the one on which prenatal diagnosis was first done; a child which can be expected to have greater opportunities in life because it is not impaired or deformed. This could be interpreted as meaning that this woman makes comparisons between the life that an aborted foetus could have had, and the one which the foetus that is later born actually has. However, it is not possible in practice to make this sort of comparison. The pregnant woman chooses whether she wants prenatal diagnosis or not in each individual pregnancy, and her decisions take place at different points in time and are thus in this respect independent of each other. It is, however, a fundamental point that prospective parents want to give their children the chance of as good a life as possible, regardless of which individuals actually become their children. The whole issue can be expressed in theoretical moral philosophy terms as an idea of impartiality – it does not matter which actual individuals come to exist, as long as they have the best possible opportunities to live the best possible life¹⁴.

No theoretical borderline as to which diseases we should be able to do prenatal diagnosis for can be based on an assessment of the child-to-be's opportunities to live the best possible life. As organisations representing the disabled have pointed out, these opportunities depend at least as much on the design of the community as on the functional impairment of the sick or disabled person.¹⁵ Resources in the community should therefore be allocated so that prospective parents can realistically envisage having a child with a disability when they are deciding about the offer of prenatal diagnosis. The issue of how serious a disease is becomes important for this reason, although ideas about the seriousness of diseases are largely subjective.

The issue remains of what weight the 'interests' of the foetus being diagnosed should be accorded.

10 See e.g. Buchanan et al., 2000, Harris, 2000, pp. 232–242, and Tännsjö, 1991, pp. 50–60.

11 See Munthe, 1996, pp. 47–48.

12 Typical examples of conditions like this are Tay-Sachs and Lesch Nyhans Disease.

13 In the USA, parents have sued care institutions for not having prevented the birth of their child with the type of condition named above (known as 'wrongful life suits'). See Buchanan et al., 2000, pp. 232–233.

14 Parens and Ash, 2000, pp. 23–26.

Against the moral philosophy perspective of impartiality and accordance of equal weight to all imaginable future people, ideas may be set that we have special obligations towards foetuses whose development has begun. In reality, a kind of compromise takes place between these two approaches. From the perspective of moral philosophy, the interests of the foetus are accorded some weight, while at the same time the opportunity of giving another child a better chance in life is regarded as an argument for terminating a pregnancy involving a foetus that will probably be sick or disabled later in life. The attempt to draw a line between 'serious' and 'less serious' conditions can be regarded as an attempt to find a compromise between these apparently incompatible standpoints.

3.3 Conflicts of interests and values

Since there are different parties involved and different values at stake, there are naturally conflicts of interests and values. The quality of life goal, for example, can come into conflict with the goal of autonomy, since the offer of prenatal diagnosis, despite its guidance function, can create more worry than it alleviates. Conflicts can of course also arise between the child's life expectancy and/or quality of life and the woman's expected quality of life. The woman may, for example, decide not to abort an impaired foetus even if most of those around her feel that an abortion would be the best thing for her. A woman may also choose to have an abortion after prenatal diagnosis even though the child would most probably have lived a good life if the pregnancy had not been terminated.

Conflicts can also arise from disagreement between the pregnant woman and the prospective father. The basic rule is that the woman has the right to make the final decisions, even if the father is opposed. Since all examinations and interventions take place in the pregnant woman's body, it is difficult to take issue with the reasonableness of this rule. On the other hand, the father's opportunity to put pressure on the woman to make certain decisions against her will can be problematic bearing in mind the woman's right to self-determination. It can be difficult for health care staff to determine whether or not such pressure is being exerted. The problem can be managed in most cases, however, if there is a dialogue from the outset between the woman and health care staff who are familiar with the problems and used to talking about them. The woman should also be offered support if she chooses to decide in accordance with her own will when the father is opposed.

The trend in Sweden and other countries is that the objective of promoting self-determination is gaining increasing priority; among other things, it has a legal basis in the Swedish health care legislation.¹⁵ The scales are increasingly tipped in favour of autonomy in cases of conflict, at least with respect to reasons for what is offered and the forms in which it is offered. This is particularly true of new fields in health care, such as assisted

fertilisation and genetic screening.¹⁷ Since prenatal diagnosis is partly based on genetic screening, the goal of autonomy will in future probably be more important in genetic prenatal diagnosis as well. One example of the increasingly prominent role of autonomy as a motive for procedures is the growth of genetic counselling. Key to this activity is that the counselling should not steer the individual's decisions; these should be based on the free will and own values of the individual.

That autonomy has come to be accorded greater weight in the event of conflicts with other values is of course not an argument that it *should* be accorded greater weight. In prenatal diagnosis, however, a decision in favour of autonomy need not mean that autonomy in general is the most important value.¹⁸ It is also possible to argue that the woman should be allowed to control her reproduction as she herself thinks best by referring to other, more traditional considerations, such as well-being.¹⁹ So, even if autonomy in the end should only be regarded as an instrumental value, even as such it can be an important objective for prenatal diagnosis. The fact that value shifts take place underlines the need for continual evaluation of activities, both to examine what actual priorities are made in value issues and to decide which are reasonable.

15 See Bartels et al, 1993; Chadwick et al, 1997, Chaps. 1 and 6; Juth, 2005, Chaps. 2 and 3.

16 Juth, 2005, Chap. 2.

17 Even if it of course is possible. See Robertsson, 1994.

18 See Tännsjö, 1999, for this argument. In that case, the conflict is at individual, rather than general, level.

3.4 Goal-based analysis

Goal-based analysis of an activity assumes that its goals have been identified and clarified. An examination then takes place of the extent to which various proposed actions or approaches are consistent or inconsistent with these goals; the extent to which they help promote them or counteract them. The goals of prenatal diagnosis have, as described, varied over time but can here be summarised in the following points:

- to enable assessment of foetal health
- to enable parents to make informed choices about how, when and what children they want (family planning)
- to enable the pregnant woman to make an informed choice about whether to carry the pregnancy to term
- to prevent individual suffering by treating a discovered impairment or disease in the foetus
- to prevent individual suffering by preventing the birth of a child with profound disabilities
- to improve public health by reducing the number of people suffering from severe diseases or disabilities
- to improve public health by reducing the number of carriers of severe hereditary diseases
- to save society money by reducing the cost of caring for people with severe disabilities and diseases.

Some of these goals are controversial and heavily criticised. This applies not least to the last few, which have helped fuel controversy about prenatal diagnosis. These goals have been criticised because they can lead to discrimination against people with disabilities.

The most important goals of prenatal diagnosis today are to promote the woman's autonomy and the health of the expected child. These goals, too, can sometimes collide, however. If this happens, they must be ranked and the analysis will involve deciding which goals are most important.

The goals are of course underpinned by values. The argument for trying to achieve a certain goal is of course that if you do achieve it, you have done something worthwhile. The values concerned here include the woman's health and the health and quality of life of the expected child. What promotes the woman's autonomy need not automatically be the same as what is best for the expected child, and vice versa.

What, then, is the advantage of a goal-based analysis of prenatal diagnosis? If we have agreed about the goals that are to be achieved by prenatal diagnosis, then we also have a means of evaluating the prenatal methods, both those which are already established and new ones whose possible introduction requires decisions on our part. We can then ask ourselves whether a method lives up to the original goals. In other words, the goals can help us tackle

the problem of drawing the line, i.e. answering the question of what type of prenatal diagnosis should be provided. The goals can also teach us something about *how* prenatal diagnosis should be provided. The ability to provide prenatal diagnosis in a way that is consistent with the goals also sets the limits for what prenatal diagnosis should be offered.

The goals of prenatal diagnosis can be achieved to a greater or lesser degree. Since autonomy and quality of life are relative terms, achievement of these goals is also relative. That is why the evaluation question always arises of whether a given method of prenatal diagnosis adequately achieves the goals. It is, then, important to follow the development of methods in the field and to provide scope for ethical analysis in the decision-making process when new methods are to be introduced into health care.

4. What type of prenatal diagnosis?

Summary

One of the basic value conflicts in prenatal diagnosis is that between the integrity of the foetus and the woman's self-determination. Other conflicts may concern our perceptions of human nature and autonomy.

If the purpose is to promote the woman's ability to make her own decision, how the line is drawn as to which diseases prenatal diagnosis will be allowed to test for will differ from a situation where the decision is based on how severe the presumed foetal disease or impairment is.

In public health care, more serious conditions are generally given greater priority than less serious ones, according to the principle of need. This principle, however, is in conflict with a consistent application of the goal of autonomy, according to which the activity should be steered by the pregnant woman's need of information and guidance. The latter might however lead to 'indication drift', i.e. an extended area of use for a given method.

4.1 Drawing the line: what values are to be taken into account and how should they be weighed against each other?

One condition for being able to offer prenatal diagnosis is that respect for human dignity is not violated, and one point of departure when analysing the consequences for human dignity is the previous SMER standpoint on the moral status of the fertilised egg (see section 2.3.8).

There are a number of different value conflicts in prenatal diagnosis. One is that between the integrity of the foetus and the autonomy of the prospective parents. Another conflict is whether there is a difference between abortions that are primarily done for social reasons and those which are done as a result of prenatal diagnosis. As described previously, there are similarities and differences between these two motives for abortion. The person best placed to make decisions of this nature is, as previously emphasised, the woman.

Other conflicts might be between perceptions of human nature and autonomy. The value that all people are of equal worth, for example, comes into conflict with use of prenatal diagnosis to diagnose characteristics such as sex. In a discussion about which diseases prenatal diagnosis is to be provided for, the *degree of severity of the disease or impairment* is important.

It is impossible to draw a theoretical line marking out what diseases or conditions it should be possible to carry out prenatal diagnosis for. This is because nobody is 'perfect' in every way, and different characteristics influence people's life expectancy and quality of life in different ways. This means that the interpretation

of what benefit to the child-to-be means does not justify a theoretical delimitation between different serious conditions, or even between characteristics and diseases.²⁰ Serious disabilities, however, affect each individual's opportunities to such a degree that in practice this is an important factor in the quality of life of both parents and children. Generally, then, the reasons for providing prenatal diagnosis are stronger the more serious the disease or impairment in question.

Does the goal of favouring the pregnant woman's autonomy provide guidance in drawing the line as to which diseases or abnormalities the health services should look for? Probably not, because what is irrelevant for one woman might be crucial for another, so we cannot generalise. It is therefore difficult, simply by thinking about autonomy, to draw a theoretical line concerning what conditions may be looked for and what measures may be considered on the basis of information about the foetus.

Nor can references to the woman's *well-being* be used to draw a theoretical line between various conditions or between diseases and other characteristics. The reason is similar to that for autonomy – different women, and different couples, worry about different things.

Tests which are in some way unreliable or have low predictive value are difficult to justify. Tests can be unreliable in a number of ways, such as being associated with a high risk of false positive or false negative results.²¹ Use of such tests is difficult to reconcile with the goal of boosting the autonomy and quality of life of the pregnant woman, since prenatal diagnosis only boosts the woman's autonomy when the diagnosis is correct. If it is not correct, then it obstructs rather than improves the woman's chances of making a decision based on her own wishes. Quality of life is also a relevant factor, since there is a great risk that false positive results will lead to unnecessary anxiety.

Tests can also be unreliable in that they give an uncertain diagnosis. The test might say that there is a *low* and/or *indefinite* probability of the presence of a given disease, or might say that there is a low and/or indefinite probability of how serious the disease is. This type of uncertainty will probably be characteristic of future tests for multifactorial hereditary disease.²² In all these cases, the test's ability to provide guidance is reduced. This type of test risks contributing to uncertainty and confusion, thus making it harder to make decisions about the future. Uncertain tests also risk contributing to unnecessary concern for the future health of the expected child, and the child

19 See Munthe, 1999, pp. 154–158.

20 A test result is a false positive if it shows the presence of a condition where none exists, and false negative if it does not show the presence of a condition which in fact is there.

21 Wahlström, 2002, pp. 24–26.

may be unnecessarily stigmatised by being regarded as sick without cause.

Even if there are therefore very good grounds for not providing unreliable and uncertain tests, it must be stressed that the reliability and certainty of tests is *relative*. The more reliable and certain tests are, the greater the chance that they might guide parents' decisions. However, prenatal diagnosis that does not provide absolutely certain answers can also sometimes provide guidance; examples of this type are nuchal translucency tests and serum screening, which in the first stage are a risk assessment. Pregnant women who undergo this test and are estimated to have a high risk of having a foetus with a chromosomal abnormality are offered supplementary testing and subsequent analysis which can establish with a high degree of certainty whether the foetus has a chromosomal abnormality.

4.2 Fairness and priority

One perspective which is relevant for prenatal diagnosis is that of fairness or priority.²³ In this context, fairness means that all citizens in the community should basically receive the same offer of prenatal diagnosis, regardless of where they live. Consequently, in the first instance, the need and solidarity principle should be applied. Generally, this aspect touches on the issue of how health care resources should be allocated. It assumes, however, that the principle of human dignity has already been taken into account. The question here is primarily whether prenatal diagnosis should be provided by society, not which type of prenatal diagnosis society wants to allow.

The issue of priorities becomes particularly pressing in a field in which knowledge and technology are progressing rapidly. In these contexts, it is important to point out that we must distinguish between the issue of the allocation of resources between prenatal diagnosis and other health care (usually called *horizontal priorities*) and the issue of the allocation of resources within prenatal diagnosis (usually called *vertical priorities*).

Priorities in health care are normally set within different types of treatment whose success *presumes* diagnosis, and not between different types of diagnostic method. Progress in genetics and in ultrasound diagnosis boosts the ability to diagnose conditions that previously could not be diagnosed at the foetal stage.

If we consider using these methods in clinical application, the question arises as to where the resources

are to come from to introduce these methods.

In view of the fact that resources in society – and in health care – are limited, it can be asserted that it is reasonable to give priority to advanced prenatal diagnosis for very serious conditions ahead of diagnosis for minor anomalies. The provision of prenatal diagnosis for conditions that cannot be counted as diseases, impairments or anomalies (sex, for example) and which are therefore not otherwise treated by public healthcare is neither ethically nor economically defensible. Wider opportunities to analyse genetic or other factors for characteristics that are not regarded as health-related could also lead to *indication drift*,²⁴ meaning the trend that a diagnosis or treatment is requested, and provided, for increasingly trivial conditions or even matters that are not regarded as medical conditions today.

Regardless of how priorities are set in practice, it is important to not just take costs into account. If the introduction of a new method means that human dignity is threatened, then that is a price in ethical terms. It is therefore important to take a position on which values can be realised by the introduction of new methods. Consequently, it is important that there is a body in society with the knowledge required to evaluate these ethical aspects.

The problem of indication drift can also occur because opinions about what conditions the medical services should deal with change over time. Today, the prioritisation decisions of the health services involve giving priority to serious diseases ahead of less serious ones. What should be regarded as a serious disease depends on factors including whether the disease can be treated. The concept of 'disease' itself can also undergo shifts in meaning and value.²⁵

The problem of indication drift can also be assumed to be increasing due to globalisation and the greater opportunities for 'medical tourism' that this brings. If we in Sweden draw a line as to what type of prenatal diagnosis is provided, the diagnosis that is not provided here will probably be provided somewhere else in the world. This of course undermines the practical impact of limits.

The necessity of priorities gives rise to fundamental value conflicts in prenatal diagnosis. Consistent application of the goal of autonomy means that the pregnant woman's values and wishes should determine what type of prenatal diagnosis is carried out. It might seem inconsistent to only allow the woman to decide as long as this is in accordance with widely held views. This is the case if the goal of self-determination is given a superior status in conflicts with other values and goals. Society does, however, set limits for what is permitted in other contexts with respect to actions and decisions that are the result of the values and cultural background of individuals. Prenatal diagnosis involves many ethically controversial questions about where to draw the line and if we are to

22 This aspect was dealt with by the Swedish Government Official Report SOU 1995:5 Vårdens svåra val (The Tough Choices of Health Care).

23 See SMER's opinion "Preimplantatorisk genetisk diagnostik" (Pre-implant genetic diagnosis), 23 January 2004, p. 17.

24 Brülde and Tengland, 2002, pp. 95–118.

cope with them, we need active ongoing analysis and discussion of these issues too.

The development of prenatal diagnosis in itself also gives rise to fundamental value conflicts. On the one hand, as we have said, consistent application of the goal of autonomy means that the activity should not really be guided by the conditions that can be discovered using prenatal diagnosis, but by the pregnant woman's need of information and guidance. This, on the other hand, gives rise to a need for greater resources, which is a question of priorities. The need in this field should therefore be continually reviewed against other health care activities in setting horizontal priorities.

5. Prenatal diagnosis: How should it be provided?

Summary

- One of the conditions on which the pregnant woman's autonomy depends is that she is able to provide informed consent to prenatal diagnosis and to the decisions that might result from this procedure; but also that she is able to decline the offer.
- The way in which prenatal diagnosis is offered is important. It is helpful if the information is provided in two stages. The first stage is when the woman decides whether to say no or whether she wants to know more about prenatal diagnosis. Those who want to know more should then, without time pressure, receive sufficient information from trained personnel, and should be provided with it in such a way as to allow the woman herself to make the final decisions.
- Information about prenatal diagnosis and follow-up information provided to the woman must be correct, comprehensive and understandable so that she is able to make the necessary decisions.
- Genetic prenatal diagnosis in the shape of screening for diseases for which there is no treatment should not be introduced because this would threaten the human dignity principle, the goal of autonomy and the quality of life of the woman and the child.

5.1 How should information about prenatal diagnosis be offered to the pregnant woman?

When we have decided what conditions or diseases prenatal diagnosis should be offered for, the question remains as to *how* it should be offered.

To be able to request prenatal diagnosis at all, the pregnant woman must know that the opportunity exists. The woman should have been informed of this before getting pregnant; this means that society has a responsibility to communicate this knowledge in schools or by other means.

At the first visit to the prenatal clinic, the midwife should ask the pregnant woman if she wants to know more about prenatal diagnosis or if she wants to decline further information. If she chooses to decline, in future she is only to be offered medically motivated examinations, including ultrasound examination.

Forcing information on women that they do not want is unacceptable, and may restrict their self-determination. The type of information that women

want, and what information they do not want, varies. It should then, be the woman herself who requests more information about prenatal diagnosis.

Pregnant women who want to know more should be offered an appointment for more information.

This should be provided at a special clinic on a particular occasion, and by specially trained staff. The information provided to the woman at this clinic should contain all relevant information about prenatal diagnosis and the possible options that may be open to her depending on the results of the diagnosis. The same applies to information about all the alternative forms of diagnosis that she can be offered, as well as the option of declining prenatal diagnosis; it also applies to possible courses of follow-up action such as abortion or giving birth to the impaired child.

Information about prenatal diagnosis should be provided as early in the pregnancy as possible to give the woman as much time as possible to arrive at a well-considered decision.

It is also important to not just bring up medical information and medical consequences of different decisions. Psychosocial factors can be just as important. What might life as the parent of a disabled child be like? What limitations will my child face in life? What care does society provide for the disabled? Will my child be able to have a family and children of its own? Can I insure the child?²⁶

The advantage of such an approach is that more time can be spent on providing information and allowing the woman to ask her questions. It is easier for the woman to ask questions of staff who are specially trained in the field. Such a clinic should also be responsible for following up women who terminate the pregnancy because the examination has shown that the foetus had an impairment. The woman's ability to control the decision according to her wishes and

5.1.1 Genetic counselling

One useful approach in giving information about prenatal diagnosis is that used in genetic counselling. The main purpose of genetic counselling is to help people who have a risk of hereditary disease in their families, and this includes the option of prenatal diagnosis.²⁷ Genetic counselling attempts to safeguard patient autonomy. Autonomy, then, is an important objective of prenatal diagnosis in general and genetic counselling in particular. It is important, however, that any conflicts between autonomy and other goals are brought to light and analysed and so it is important that staff who are to guide the woman through the prenatal diagnosis process are well versed in ethics, and knowledgeable enough to be able to identify goal conflicts. More specific ethical expertise should also be available so that a satisfactory analysis can be carried out of any goal conflicts, as a basis for further guidance.

25 See for instance Platt Walker, 1998; Munthe, 1999, p. 83; Juth, 2005, pp. 84–85. values is decisive from an autonomy point of view.

26 The classic formulation of the central components of genetic counselling is found in Fraser, 1974, p. 637. For a history and analysis of these components, see Juth, 2005, pp. 72–79.

5.2 Genetic screening

5.2.1 Background

So far, our point of departure has been that it is the woman who takes the initiative in the question of possible prenatal diagnosis, because she is worried about having a sick child. Another possible alternative is that the initiative comes from the health services or the community. Examinations of this type are called screening, and are generally provided to all individuals in a certain group, or at a certain time in life. If genetic techniques are used in screenings, the genetic integrity of the individual may be at risk. If the screening refers to genetic methods carried out during prenatal diagnosis, the individual's genetic integrity may be even more under threat and the question becomes even more important.

The World Health Organisation (WHO) has established ten criteria for the screening of neonates and these may also be important in screening during prenatal diagnosis. The criteria include a requirement that a discovered condition should be treatable. Genetic screening programmes can only be justified if effective treatment is available for the hereditary disease in question.

There is reason to believe that genetic screening in prenatal diagnosis may provoke more worry than it alleviates. The majority of those who would undergo such an examination have not had any reason to suspect an elevated risk, so they have no concerns that would be alleviated by prenatal diagnosis. If the health services inform them about, and offer, an examination which they have not requested, this may generate unnecessary anxiety.

Screening also reduces the autonomy of individuals in that they are deviations from the rule that care should be initiated by the individual. When health services staff offer care and testing, the patient may feel that she is being urged to undergo the procedure, since care personnel are regarded as authorities in these matters. If the patient must take the initiative of saying no to avoid a test, or if staff use words that suggest they are urging the patient to undergo a test, the way in which patients are informed may put them under pressure, and thus be a departure from the principle of voluntary consent.

Genetic counselling may help the individual make an independent decision and thus promote patient autonomy, but such counselling can hardly be regarded as realistic in genetic screening because of its prohibitive cost.

5.2.2 Examples of screening in other types of prenatal diagnosis

In Sweden, ultrasound is a screening examination that in principle is carried out on all pregnant women. The goal has not primarily been to look for foetal impairments but rather to establish the duration of the pregnancy and whether the pregnant woman is carrying more than one foetus. The Swedish

Council on Technology Assessment in Health Care (SBU), however, concludes in its report on routine ultrasound scans in pregnancy that 'the scientific basis indicates that prenatal diagnosis should be routinely offered as part of screening activities'. It is this position, among other things, that has led to the use of checklists which mean that ultrasound diagnosis can be divided into a simple examination with the original objective, and a detailed examination for the purposes recommended by the SBU. It is important, however, that information ahead of this examination gives the woman a chance to understand that even the simple ultrasound scan might lead to the discovery of serious deformities and disease in the foetus, and that she can decline the scan if she so wishes.

6. Who should set the framework of prenatal diagnosis?

Summary

- Methodological developments in prenatal diagnosis are rapid, among other things because of progress in molecular genetic research. This is why prenatal diagnosis should be assessed on an ongoing basis. We must continue to discuss the defence of human dignity.
- It is important for the legitimacy of prenatal diagnosis that continual follow-up and evaluation is characterised by openness and influence for the groups involved.

6.1 Value shifts

Assessment of the terms on which prenatal diagnosis should take place is based on values of various types. These include our view of human dignity and integrity. Factors that must be considered include assessments of how safe a method of prenatal diagnosis must be for its use to be defensible. Values, and hence the limits for what is deemed defensible, also shift over time, depending in part on our view of disease.

6.2 The basic prerequisites for prenatal diagnosis as laid down by the Riksdag

Issues of this type should be dealt with in a democratic process. The strength of the democratic process is that it is a forum for the airing of contradictory values, such as those involved in this issue. The Genetic Integrity Act which came into force on 1 July 2006 is a decision made on democratic grounds which sets out the fundamental prerequisites for the prenatal diagnosis to be provided in Sweden. However, society must specially monitor and assess new methods to prevent undesirable developments in a field that encompasses so many ethically and socially controversial issues.

To be able to capitalise on the benefits that can come from the rapid developments in the field, however, some *flexibility* is desirable. The general conditions laid down by the Riksdag can only be altered by amending the legislation. Since this is a process which needs time, and indeed must be allowed to take time, it would be desirable to have a level which can monitor and assess new methods before they are brought into use, but which can still create legitimacy. There is otherwise a great risk that new and improved methods which are available and are requested by pregnant women will be introduced into care without there having been time for any ethical analysis. It is, however, not just the demands of pregnant women that might lead to the introduction of new methods without prior ethical analysis; there is also a significant risk that methods will be introduced because they are felt by the health services to have

many advantages. If society wants to influence the introduction of new methods in the field, experience shows that such assessments must be carried out before the method is introduced. If a new method which proves ethically dubious gains a foothold in clinical activity, it is very difficult to then withdraw it.

The range of prenatal diagnosis on offer can also be influenced by commercial interests. In practice, this may for example mean that commercial prenatal clinics offer ultrasound diagnosis to find out characteristics of the foetus, a service that is not offered by public health services.

If, then, prenatal diagnosis is to have legitimacy, we need ongoing evaluation and assessment characterised by transparency, discussion and influence for the groups involved. The issue is how society can promote such a process.

7 Summary and conclusions

7.1 Introduction

Prenatal diagnosis brings difficult ethical issues to the fore that concern many people. It involves our perceptions of human nature and our values. It also involves our attitude to disease and disability. Prenatal diagnosis can also provoke anxiety, particularly in the pregnant woman. A woman who is to decide on whether or not to undergo prenatal diagnosis is faced with a life-changing decision which in the first place affects her and her family, but which is also fraught with basic ethical values such as human dignity, self-determination, quality of life and life expectancy.

Against the background of rapid developments in prenatal diagnosis, the question has primarily been which prenatal diagnosis methods society and the health services should offer. The answer depends on factors such as the values prenatal diagnosis can promote and the risks associated with the activity. Important ethical values involved are the perception of human nature, human dignity, informed consent, fairness, need and solidarity and the moral status of the fertilised egg. One value which is usually particularly emphasised is the woman's right to autonomy.

The woman's self-determination is limited by the framework established by society. This framework should be based on an ethical analysis of existing value conflicts from a society-level perspective. Such an analysis is necessary for e.g. early discovery of indication drift which might threaten human dignity, integrity or other ethical values. According to the goal of women's self-determination, it is not the condition that prenatal diagnosis may discover which is to steer the activities, but rather the pregnant woman's need of information and guidance. The information provided to the woman prior to prenatal diagnosis is one of the prerequisites for her self-determination. The health services must have the requisite skills to be able to answer the woman's questions, thus reducing her concerns. Information should be provided to the woman in two steps.

Screening in prenatal diagnosis raises difficult ethical issues. One of the problems is that it is the care services, and not the woman herself, that take the initiative. From the perspective of the principle of human dignity, the goal of autonomy and the patient's well being, genetic prenatal diagnosis in the shape of screening should not be introduced if there is no treatment for the condition that can be discovered by the screening process.

The Swedish National Council on Medical Ethics wishes to underline in particular that:

- the conflict between different ethical values needs ongoing analysis
- self-determination is important, one reason being to avoid undesirable consequences for society
- self-determination can be promoted through increased information and through the means selected for providing the information
- analyses which yield uncertain information or which are unreliable should not be used
- the risk of indication drift needs to be monitored
- prenatal diagnosis methods should, like all other health care, be subject to discussions of priorities, which also limits the types of prenatal diagnosis to be offered.

7.2 Proposed measures

We need continual monitoring of prenatal diagnosis and a broad public debate, in which society takes official positions.

As an advisory body to the Government, SMER has the task of monitoring and analysing the consequences of issues that can affect human dignity in particular. SMER does not, on the other hand, have the task of examining the shape that concrete organisational measures should take in order to safeguard fundamental ethical values. SMER's examination has, however, pointed to a number of issues that may require consideration and more concrete action. What SMER wishes to point out in particular is that there is a need for well-considered arrangements for managing the introduction of new methods of prenatal diagnosis and for the decision-making process when these methods are to be introduced into health care. SMER also wishes to stress the necessity of continual monitoring of these issues so that there is preparedness to take action when human dignity comes under threat.

If a woman is to be able to make independent and well-considered decisions, she must have the necessary knowledge on which to act. This, however, in turn assumes educational measures on a number of different levels, both for the woman who is to make the decision and the health care staff who are to pass on the knowledge to the pregnant woman, and for others who might be affected in other ways by prenatal diagnosis. Further measures are also necessary to investigate how the health services can design appropriate arrangements for providing information.

References

- Arrhenius G (2005), *Vem bör ha rösträtt?* (Who Gets to Vote?), *Tidskrift för politisk filosofi*, 2005, 2: 47-63.
- Beauchamp TL & Childress JF (2001), *Principles of Biomedical Ethics*, 5th edition, New York & Oxford: Oxford University Press.
- Brülde B (2003), *Teorier om livskvalitet (Theories of Quality of Life)*, Lund: Studentlitteratur.
- Brülde B & Tengland PA (2003), *Hälsa och sjukdom – en begreppslig utredning* (Health and disease: an examination of the terms), Lund: Studentlitteratur.
- Buchanan A et al (2000), *From Chance to Choice – Genetics and Justice*, Cambridge University Press, Cambridge, UK.
- Bartels DM, LeRoy BS & Caplan AL (1993) (eds.), *Prescribing Our Future. Ethical Challenges in Genetic Counselling*, New York: Aldine De Gruyter.
- Chadwick RF, Levitt M & Shickle D (1997) (eds.), *The Right to Know and the Right not to Know*, Avebury: Aldershot.
- Clarke A (1994) (ed.), *Genetic Counselling: Practice and Principles*, London & New York: Routledge.
- Dahl, RA (1989), *Democracy and its Critics*, London: Yale University Press.
- Fleischhauer K & Hermerén G (2006), *Goals of Medicine in the Course of History and Today. A Study in the History and Philosophy of Medicine*, Stockholm: Vitterhetsakademien/Royal Society of Letters etc, Almqvist & International (distr).
- Fraser FC (1974), "Genetic Counselling, *American Journal of Human Genetics*, 174, 26: 636-659.
- Glover J (1977), *Causing Death and Saving Lives*, London: Penguin Books.
- Gustafson S (1980), *Fosterdiagnostik ? för vem?* (Who is prenatal diagnosis for?), Stockholm: LT's Förlag.
- Hares, RM (1993). *Essays on bioethics*. Oxford: Clarendon Press.
- Harris, J (2000), "Is There A Coherent Social Conception of Disability?", *Journal of Medical Ethics*, 26, 95-100.
- Hermerén, G. (1994), *Informed Consent from an Ethical Point of View. I: Westerhäll, L, Phillips, C* (eds): *Patients' Rights: Informed Consent, Access and Equality*. Stockholm: Nerenius & Santerus.
- Juth N (2005), *Genetic Information – Values and Rights: The Morality of Presymptomatic Genetic Testing*, Göteborg: Acta Universitatis Gothoburgensis.
- Locke J (1689), *Two Treatises of Government*, Laslett P (ed.), New York: Cambridge University Press, 1988.
- Mill J S (1859) *On Liberty*, Suffolk: Penguin Books, 1974.
- Munthe C (1996), *The Moral Roots of Prenatal Diagnosis. Ethical Aspects of the Early Introduction and Presentation of Prenatal Diagnosis in Sweden*, Göteborg: Centrum för forskningsetik.
- Munthe C (1999), *Pure Selection. The Ethics of Preimplantation Genetic Diagnosis and Choosing Children without Abortion*, Göteborg: Acta Universitatis Gothoburgensis.
- Munthe C, Wahlström J & Welin S (1998), *Fosterdiagnostikens moraliska rötter. Goda handikappomsorger avgörande för den etiska kvaliteten* (The moral roots of prenatal diagnosis. Good care of the disabled is key to ethical quality), *Läkartidningen* 1998, 95: 750-753.
- Nozick R (1974), *Anarchy, State and Utopia*, Basic Books: New York.
- The Nuffield Council on Bioethics (1993), *Genetic Screening and Ethical Issues*, London: The Nuffield Council on Bioethics.
- Parens E & Asch A (2000), *Prenatal Testing and Disability Rights*, Washington, D.C.: Georgetown University Press.
- Platt Walker A (1998), "The Practice of Genetic Counselling", i Baker DL et al (red), *A Guide to Genetic Counselling*, Wiley-Liss Inc, ORT: 1-26.
- Robertsson JA (1994), *Children of Choice. Freedom and the New Reproductive Technologies*, Princeton, N.J.: Princeton University Press.
- Saltvedt, S (2005), *Prenatal Diagnosis in Routine Prenatal Care – A Randomised Controlled Trial*, Stockholm: Kongl Carolinska Medico Chirurgiska Institutet.
- Sidgwick H (1907), *The Methods of Ethics*, Edinburgh: R & R Clark Ltd.
- Scott R (2005), "Prenatal Testing, Reproductive Autonomy, and Disability Interests", *Cambridge Quarterly of Healthcare Ethics*, 2005, 14: 65-82.
- Schickle D (1999), "The Wilson and Jungner Princip-

- les of Screening and Genetic Testing”, Chadwick R et al (red), *The Ethics of Genetic Screening*, Dordrecht: Kluwer Academics Publishers: 1-34.
- Tännsjö T (1991), *Välja barn. Om fosterdiagnostik och selektiv abort* (Choosing a child. On prenatal diagnosis and selective abortion), Borås: Sesam.
- Tännsjö T (1998), *Vårdetik (Ethics of care)*, 3:rd revised edition, Falun: Thales.
- Tännsjö T (1998b), *Hedonistic Utilitarianism*, Edinburgh: Edinburgh University Press.
- Tännsjö T (1999), *Coerceive Care: The Ethics of Choice in Health and Medicine*, London & New York: Routledge.
- Wahlström J (2002), *Hälsa, ärftlighet och genetiska undersökningar – medicinska bakgrundsfakta* (Health, heredity and genetic examination – the medical background), in *Genetisk screening – om hälsa och ärftlig sjukdomsrisk* (Genetic screening – on health and the hereditary risk of disease) , Stockholm: Swedish National Council on Medical Ethics.
- Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine, Council of Europe, European Treaty Series, no. 164, Oviedo, 4.IV.1997.*
- Abortfrågan. Remissyttranden över 1965 års abortkommittés betänkande Rätten till abort* (Abortion. Response fo the 1965 abortion committee report The Right to Abortion), SOU 1972:39, Stockholm 1972: Department of Justice.
- SOU 1989:51, *Den gravida kvinnan och fostret – två individer. Fosterdiagnostik, abort m.m.* (The pregnant woman and the foetus: two individuals. Prenatal diagnosis, abortion etc.), *Socialutskottets betänkande* (Report of the Committee on Health and Welfare) 1994/95:SoU18, Stockholm: Sveriges riksdag.
- Fosterdiagnostik och abort* (Prenatal diagnosis and abortion) (Govt. bill. 1994/95: 142)
- SOU 1995:5, *Vårdens svåra val (The Hard Choices of Care)*.
- SOU 2004:20, *Genetik, integritet och etik (Genetics, Integrity and Ethics)*.
- Govt. bill, *Genetisk integritet mm* (Genetic Integrity, etc.) 2005/05:64
- Vetenskapsrådets konsensusuttalande om tidig fosterdiagnostik (Swedish Research Council Consensus Opinion on Prenatal Diagnosis), (2001). Swedish Council on Technology Assessment on Health Care, *Rapport om Rutinmässig ultraljudsundersökning under graviditet* (Report on routine ultrasound examination during pregnancy), (1999).

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