12  Prenatal screening for Down syndrome  
Why we shouldn’t?  

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**Introduction**

At the turn of the century, several Western countries introduced prenatal screening for Down syndrome. Technology became widely and easily available, and the combination of ultrasound and then blood tests made it possible to screen the entire pregnant population. For the first time in history, there was a chance of detecting, and eventually aborting, most instances of Down syndrome, and at relatively low cost.

Some countries chose not to offer early screening during pregnancy, using value-based arguments, often claiming solidarity to weaker members in society. Yet, the same countries typically accepted abortion and prenatal diagnosis based on previously established maternal-age criteria.

This chapter investigates and discusses from a philosophical perspective whether there is a relevant difference between offering prenatal tests to high-risk groups as opposed to the entire pregnant population. Is this an ethical question, or a matter of more effective ways to introduce and use new medical technology? To screen or not to screen, that is the question.

**Autonomy: a primary argument for screening**

An illustrative case is the recent history of maternity care in Denmark, where the Danish National Health Service produced the report *Prenatal Diagnosis and Risk Assessment* (Sundhedsstyrelsen 2003) proposing a ‘paradigmatic change in prenatal practice’. This report described the former paradigm as one centred on the prevention of disability, a mind-set said to have emerged from twentieth-century eugenic ideas. This report further suggests that prenatal diagnosis has been continuously contaminated by this way of thinking until fairly recently, ideas which should be abandoned and replaced by a new paradigm inspired by one prominent value in particular: autonomy. This would mean that the success criteria for prenatal screening would not be prevention of disability through a greater number of selective abortions, nor in economical expenditure brought about by a reduction of children born with impairments. The new sole criterion for success would be that pregnant women (and their partners) could exercise freedom of choice when it came to prenatal testing for Down syndrome.
Autonomy and screening apparently then belong together? If the primary justification for having a test is to increase freedom of choice and deciding future life directions, then, in principle, anyone who could benefit from such tests should have equal access to it. While the risk of having a baby with Down syndrome apparently increases with the age of the mother, every pregnancy involves a certain degree of risk, and thus, screening for Down syndrome seems to represent a fair distribution of both medical technology and information for individuals to be able to make autonomous decisions. In Denmark, as well as in most countries introducing screening for Down syndrome, the larger question about the relation between autonomy and the presumed ‘burden’ of eventually having a child with Down syndrome has not been a major consideration. Since prenatal diagnosis had been offered to Danish women over the age of thirty-five years since the beginning of the 1980s, this debate appeared to have been settled. If one agrees with A, one has to agree with B, the argument went. And in 2004, prenatal screening for Down syndrome was introduced in Denmark.

At the same time, the expert group producing the report emphasised that autonomy does not always mean ‘a free choice from the uppermost shelf’ (Sundhedsstyrelsen 2003). Autonomy and free choice are always defined within certain limits, a claim few philosophers would protest. Relevant limits in this case concerned the choice of prenatal tests. Pregnant women were offered these tests based on a certain risk assessment. They could choose whether they wanted to test for Down syndrome or not, but not which tests they wanted. Interestingly, this leads us to another argument for screening.

The not so hidden agenda: improving the accounts

Whether the world is changed by ideas or whether changes are caused by more materialistic forces is a set of classical philosophical and political questions and arguments. The reason why western countries put prenatal screening for Down syndrome on the agenda has at least something to do with the development of new technologies, most notably breakthroughs in ultrasound diagnosis in the 1990s. High-resolution pictures, improved interpretive skills, and the detection of more ‘soft markers’ for disease and impairments enabled the way for ultrasound implementation early in pregnancy. The so-called ‘thick neck-fold’ has been central in identifying Down syndrome (Nicolaides et al. 1992; Spencer et al. 1999; Taipale et al. 1997), and an increased amount of fluid in the neck of the foetus is also a possible sign of Down syndrome. Why this technology made the ‘autonomy paradigm’ possible becomes more visible if contrasted with its test-predecessor, amniocentesis.

Amniocentesis was and is an invasive technology. The needle enters the womb, and such an intervention increases the risk of miscarriage. The risk is not dramatically high: about one out of a hundred pregnant women tested will abort as a result of this pinprick. But the total number of losses would be unbearable for health services worldwide if every pregnant woman were offered the possibility of amniocentesis.
Denmark, along with Italy, has had the greatest reported use of amniocentesis in Europe, something which has been morally quite costly for Danish health services (Sundhedsstyrelsen 2003: 127). The number of foetuses without Down syndrome whose lives were lost as a consequence of amniocentesis was much higher than the number of foetuses detected with Down syndrome (Getz and Kirkengen 2003). By contrast, ultrasound in combination with blood tests could be offered to the entire pregnant population without having a negative impact on the balance of results: more foetuses with Down syndrome could then be detected while fewer foetuses without Down syndrome would be lost (Sundhedsstyrelsen 2003).

The point I am making here is that Denmark and other countries offering early ultrasound screening did not introduce it in order to rid themselves of people with Down syndrome. The new technological solution had its own internal logic: more (and even most) cases of Down syndrome could be detected, and at a lower cost of human life. No matter how one chooses to view this, either from an economic or human perspective, the balance was better than it had been.

Of course, this might be good news if plans were intended to reduce the number of future people with Down syndrome. But based on the lesson learnt from Danish eugenic history (and other countries), such intentions were increasingly discredited. More and more, the dominant values concerning prenatal diagnosis in most western societies today are those of free choice and self-determination, and the new technological possibilities suit these ideals very well. After all, the more risk-free tests available, the more choices are made available. Giving everyone the choice of having (or not) a baby with Down syndrome by offering non-invasive tests increases the autonomy of pregnant women and/or their partners, no matter what choice they make. To test or not to test, carrying an affected foetus to term or aborting, became solely all about autonomy! For the Danish National Health Service, this seemed to be a win-win situation: the transition from invasive to non-invasive technology combined with the expansion of free choice seemed to represent some sort of ethical progress.

**Arguments against screening**

Scrutinising technology sheds light on how the screening debate is contextualised in different ways. For instance, Denmark’s neighbour country Norway has had stricter maternal age criteria than Denmark (thirty-eight years old), and as a consequence, the number of amniocentesis procedures performed in Norway and the number of unwanted losses has been fewer. If Norway were to introduce prenatal early screening for Down syndrome, the total number of amniocentesis procedures might increase dramatically and so too the number of unintentionally lost healthy foetuses. The ‘internal logic’ of the technology then, did not clear the way for autonomous choice. In practice this meant that the ‘push’ from the technomedical establishment towards screening was not as great as it would have been if it had represented a clear step forward when compared to previous patterns.

In fact, resistance towards introducing early ultrasound screening was quite high in Norway, even though the Norwegian National Health Service offered...
testing for Down syndrome for women in the ‘high risk’ category. One example of this resistance is that the Norwegian Minister of Health recommended in 1999 that a leading Norwegian ultrasound specialist stop a planned study on early ultrasound (NTB 1999), fearing that the study would encourage early ultrasound screening.

A central argument against screening in Norway and elsewhere is a version of an expressivist argument. The expressivist argument is articulated, among others, by Adrienne Asch’s ‘any–particular distinction’ (Asch 2000): to abort a foetus when the pregnancy is unwanted is to abort ‘any kind’ of foetus, whereas to abort a foetus with (for instance) Down syndrome is to abort a ‘particular one’ with characteristics that are shared by other members of society. While an abortion can belong to the private sphere, concerning primarily the woman and the foetus in her womb, a selective abortion affects other people or groups of people in society and is then a public and societal matter. This is the ethical-political dimension of selective abortions, according to Asch. In a selective abortion, certain ideas about the anticipated quality of life (or absence thereof), about burdens or happiness (or not), and about the meaning of family life provide premises for making the decision. This line of thinking leads to a conclusion that selective abortions express discriminatory or offensive attitudes to (particular groups of) disabled people.

Nancy Press (2000) argues not to focus on the message from a specific abortion or on the woman having an abortion, but rather on the meaning of the offered possibility of prenatal testing itself. Her point fits well with the public debate in Norway, and opens up a possible ethical distinction between a high-risk strategy and screening. The offer of testing to a small high-risk group has different implications than the offer of testing to a larger number of low-risk pregnant women. In the case of the former, prenatal tests can be legitimated on the grounds of a conscious knowledge and anxiety about one’s own ‘natural’ risk. In the case of the latter, the message from a National Health Service would seem to be much clearer: Down syndrome is such a potentially severe threat to a ‘good family life’, that abortion might be a better solution, at least for some.

The expressivist argument has a strong standing in Norwegian public debates, occurring in various versions. One creative version was presented in the spring of 2007 on national Norwegian television, when Marthe Goksøyr presented her own video diary (NRK 2007). Marthe has Down syndrome and filmed herself entering the laboratory of a medical genetics department with her video camera, asking the staff there why parents wanted to terminate people ‘like her’. The result (an extremely uncomfortable situation for the geneticists) did not in itself constitute proof that the expressivist argument is correct, but the uncomfortable situation needs to be understood and explained.

A credible response from the geneticists might have been one provided by several bio-ethicists: terminating a foetus is not terminating Marthe or someone else, but rather only about terminating a foetus. This position is argued by several well-known bioethicists, among them John Harris (1998) and Peter Singer (1994). Tom Shakespeare formulated such a position as follows: ‘it seems intuitively true that if it is permissible to terminate pregnancy at all, it is permissible to terminate in
the case of disability’ (Shakespeare 2006: 93). One perhaps tends to forget that we are thinking and talking about foetuses here, and that many of us would find it permissible to terminate a pregnancy even when it does not involve a potential disease or impairment.

But such a response does not seem to consider all of the messages regarding the central question of this chapter, that of screening: some messages and signals are more intense than others. Perhaps it is not the permissibility of terminating that is necessarily at issue but rather the permissibility of actively arranging situations where terminating is one of the endpoints? Such a step goes too far judgementally for the (family) lives of people with Down syndrome, according to critics. But is this critique a sustainable one?

**The struggle for recognition**

Many people with Down syndrome and their families feel that the message from prenatal screening hurts, diminishes and devalues them in various ways. One mother quite well known in Norway who has a child with Down syndrome formulated it this way in the media: ‘We find that health professionals talk about people with Down syndrome as if they were defect cars that shouldn’t have been on the market’ (Borud 2000).

If this were a representative experience for disabled people and their relatives, it would be easy to conclude that prenatal screening is at least perceived as offensive. But isn’t reality more complex? Disabled people and their families do not always experience a sense of diminishment when confronted with prenatal screening and diagnosis. Perhaps the variable here is not primarily one of ideas or values, but rather which impairment is being considered? After all, the conditions of spina bifida and cystic fibrosis have not led to the same degree of controversy and intensity in public bioethical debates as has Down syndrome. A simple but convincing way to establish this claim is that a recent search in the Retriever archive for Norwegian media provided 513 hits for the combined terms ‘Down syndrome’ and ‘prenatal diagnosis’, whereas a search for ‘spina bifida’ and ‘prenatal diagnosis’ resulted in twenty-four hits, and ‘cystic fibrosis’ and ‘prenatal diagnosis’ in only fifteen hits (A-tekst 2008). One reason for this might be that the overall incidence of Down syndrome is higher than the incidence of spina bifida and cystic fibrosis. But this may not be the main reason. The main reason, in my opinion, has something to do with the fact that some impairments and disabilities constitute what Stainton (2003) has called ‘strong identity characteristics’, and this then makes the expressivist critique more relevant when considering Down syndrome compared to other impairments.

Stainton’s point is that prenatal screening for intellectual disabilities can in itself be viewed as an expression of an identity-based oppression. The fact that people with intellectual disabilities neither objectively suffer, nor experience any more or less sadness or joy than the rest of us seems to count for little, even among those who are otherwise concerned with the implications of testing and elimination for other types of disabilities, according to Stainton (2003: 538). In his
view, then, prenatal screening for Down syndrome may not only have potentially negative consequences for people with Down syndrome and for their families. We could instead turn it around: prenatal screening for intellectual disabilities may be an indication that something has gone wrong in society with regards to identity construction. And Stainton suspects that the origin of the problem is in the history of modernity, where the intellect is so highly valued.

I believe Stainton is correct, at least in his focus on identity. Down syndrome is different from spina bifida, well illustrated in Norway where a former Minister of Environmental Protection, Guro Fjellanger, had spina bifida, while no one with Down syndrome is likely to become a minister. Fjellanger was dependent on crutches and wheelchairs and accessible settings in order to do her job. Having Down syndrome is not an impairment that can be compensated for so easily, at least not to the extent that one could perform most duties of a governmental minister. A person with Down syndrome would not just be different, but too different to include in politics at that level.

Some people might then draw the conclusion that prenatal screening for, and the abortion of, foetuses with spina bifida is morally more problematic than Down syndrome, because the latter is potentially a much more severe condition than the former. The former is not necessarily even a hindrance to becoming a minister. But this logic fails to explain public controversies about Down syndrome and the lack of public controversies about spina bifida. The point is rather that prenatal screening for Down syndrome highlights the problematic nature of intellectual impairments in our societies. People with Down syndrome and their families face what Charles Taylor (1995) has called a ‘struggle for recognition’ to a larger extent than people with spina bifida and their families.

**Terminating foetuses, terminating burdens or terminating identities?**

John Harris (1998: 215) has subjected such expressivist relations to critique in his ‘argument from Beethoven’. Beethoven, it will be recalled, was deaf. But to abort a foetus with ‘Beethoven syndrome’ is not to abort Beethoven, but rather as Harris contends, it is just to abort a foetus. Harris’s argument can be clarified thus: everyone would prefer to have babies with two legs instead of one leg. If we could prevent having a one-legged baby by postponing conception by one month, we would and should postpone conception. To then claim that our actions were offensive to people with only one leg, or that we were expressing oppressive identity characteristics, would be absurd. So in Harris’ view, regarding debates on disability, respect and recognition are non-starters: it is ‘better’ to have children without Down syndrome in the same way that it is better to have children with two legs instead of one. According to Harris, that is what prenatal diagnosis is all about.

Harris presupposes that a foetus is not a moral person: it is, then, ‘a nothing’. And thus, preventing impaired children by abortion or postponed conception amounts to one and the same thing. This position is of course valid if you happen
to believe that an embryo or a foetus has no moral value whatsoever. But since this position represents only one possible interpretation of the moral standing of the foetus, and one that is marginal in most western cultures, it is a position that does not adequately provide meaning to the widespread feeling that prenatal diagnosis and selective abortion are related in important ways to the politics of disability.

An alternative point of departure would be that most people think we should ascribe some moral value to the foetus. Even where women can have abortion on demand, without having to justify their decision, there is still a tacit requirement for a kind of ‘private’ justification. To abort because the pregnancy collides with an unexpected holiday in the Greek islands is not sufficient justification in the minds of most people. The (presumed) burden does not outweigh the kind of moral value that is typically assigned to the foetus. This vague notion of morality strongly suggests that preventing disability by abortion has a ‘moral cost’. I am not claiming that the moral cost is the killing of a person, but simply preserving the common intuition that some moral cost is involved.

In contrast, if future people with Down syndrome could be prevented by postponing certain conceptions or taking a pill before conception, there would be no moral cost involved in such prevention, because abortion is excluded from the options. The expression of such prevention is that forthcoming parents want children without Down syndrome. But everyone knows that forthcoming parents as a basis want children without disease and impairments. The message to be inferred from prenatal screening, on the other hand, is that (family) life with Down syndrome is so negatively valued that the morally problematic action of abortion could be preferable.

Since opinions differ as to what extent any abortion is morally problematic, this means that the grade of offensiveness also has to differ. I call this the ‘gradualist view of offensiveness’. This means among other things that it is logically impossible to defend a position where one denies any kind of moral value to the foetus, while simultaneously claiming that prenatal screening is offensive. Here, Harris is quite correct. A provoked abortion must be viewed as an event that involves a ‘moral cost’ if one is to claim that there is a message sent from prenatal screening programs and selective abortions to disabled people in society. And in most cases, this would be viewed as having a moral cost.

On the other hand, a gradualist view of offensiveness is not just dependent on the moral judgement of abortion. It is a necessary but not sufficient condition. In addition, it is dependent, as stated earlier in this chapter, on the nature of the disease or disability in question. When Norwegian women still chose selective abortion on the basis of spina bifida, after Guro Fjellanger became a national Minister and a public figure, they were not ‘terminating the minister’. They still knew there might be potentially substantial burdens connected to (family) life with a child with spina bifida, and that the spectrum of the impairment could vary dramatically. So, most of them opted for termination. To abort a foetus with Down syndrome, on the other hand, is probably to a certain extent to abort the person that is too different to become a minister. Down syndrome has a stronger identity characteristic than spina bifida. When the majority
culture is able to become acquainted with a person with spina bifida as a minister, or a university student, an artist or a gay person, the same majority culture will in all probability and in most cases still ‘know’ a person with Down syndrome as a person with Down syndrome. The task of getting beyond the impairment will be much larger. Of course one could argue that there are often significant burdens related to having a child with Down syndrome (and the burdens will not disappear when the child grows up). But to a larger degree, it is the identity that is terminated in a prenatal screening for Down syndrome, and not first and foremost the future burden. The difference, and not the anticipated or potential burden, is at stake. One proof of that is the shock women get when they unknowingly give birth to a child with Down syndrome. Getting a postnatal Down syndrome diagnosis, the parents feel shocked, angry, devastated, overwhelmed, depressed, stunned and helpless (Skotko 2005). This shock is probably not primarily caused by the thoughts on the burdens lying ahead but rather provoked by the difference between the wanted child and the real child.

The strong identity-forming character of Down syndrome is based partly on the fact that people with the syndrome typically have intellectual/cognitive impairments and partly on the fact that this impairment is at the same time thought to mean a happy life for the person with Down syndrome, such as a cheerful way of being. Additionally, a person with Down syndrome is recognisable on the street because of her or his facial characteristics. All of these factors add up to Down syndrome being constructed as ‘pure difference’ in societies such as Norway. Whereas Fragile X syndrome is understood as a disease, for instance, and is understood to belong to the ‘domain’ of medical geneticists and genetic counsellors, Down syndrome is a symbol for a different way of ‘being human’ in the public sphere: a way that is vulnerable but also one valuable and worth protecting.

If this argument is correct, people with Down syndrome and their families have good reasons to be offended by prenatal screening offers. It is a basic need to feel that you live in a society where you and your child are welcome. It seems then that the expressivist position has something relevant to contribute, especially with regards to the larger implications of intellectual impairments such as Down syndrome. Under attack here are the identities and descriptions that people with Down syndrome can create and thrive with. The greater the human and economic costs, coupled with societally sanctioned prenatal tests and service-supports designed to give parents the choice of preventing babies with Down syndrome, the more difficult it becomes for people with Down syndrome to have the positive aspects of their identity recognised by society.

At the same time, there is a paradox here: precisely because Down syndrome has been successfully constructed as a ‘difference’ in the Norwegian public sphere, the termination of foetuses with trisomy 21 has become problematic. Framing impairment in order to make it a question of identity may be an empowering strategy in many arenas in society, whereas in prenatal diagnostics it leads to confrontation and a feeling among some people that the practice is offensive. As Lynn Gillam has remarked, to have someone look at your life from the outside,
making judgements about how fulfilling and happy you are – or not – must be deeply offensive. But at the same time Gillam warns against equating this feeling of offence with discrimination. She writes, ‘The fact selective abortion is offensive to many people with disabilities does not in itself make selective abortion discriminatory to those who are offended by it’ (Gillam 1999: 170).

‘Offence’ is a rather vague kind of harm, and there are other powerful ethical considerations that deserve consideration. If critical voices have been thoroughly heard in a country such as Norway, could it be that the Danish voices for autonomy and maternal care have been ignored? If it is a fact that intelligence is valued in modern societies, and if we recognise that, then shouldn’t autonomy and not just a discussion on ‘burdens’ be relevant?

**Autonomy as trump**

Returning to my claim, perhaps the paradigm shift in Denmark was mainly motivated by the efficiency of new technology and not by considerations of autonomy. But even if this claim is correct, it does not make the issue of autonomy irrelevant or invalid. Perhaps quite the opposite: references to the value of autonomy provide a very common and powerful response to new technological possibilities.

There are several important reasons why autonomy should play a central role in medical genetics. Many western countries, and indeed the Scandinavian countries, have histories of eugenic practice, where violation of autonomy and a lack of respect for individual preferences and choices were at the core of mistakes in medical genetics (Broberg and Roll-Hansen 1996). Autonomous choice seems to be a bulwark against oppression. Autonomy and self-determination also seem to represent a highly relevant perspective in choices that will affect individuals dramatically. If we are allowed to choose between different toothpastes, there seems to be little sense in restricting individual choice in more important matters. The logic should instead be the other way around: autonomy and self-determining freedom are primarily important when there is something very significant at stake.

What is seen as offensive about prenatal screening for Down syndrome is precisely the same factors that make autonomy and choice extremely relevant to this practice. Down syndrome represents not a disease, but rather a radical difference. This difference challenges the idea and the purpose of reproduction. No one denies that having a baby with Down syndrome is a shock, although it is known that most parents eventually adapt to the situation. It is a potentially greater challenge to become reconciled to the fact that your foetus has Down syndrome, than to become reconciled to the fact of an unwanted pregnancy. But it is only in the last instance that autonomy and choice become ‘obvious’ relevant factors in, for example, a country such as Norway. Having a seriously impaired child provides major and long-term challenges to parents. It seems odd to deny, then, that autonomous choice is a matter of concern in such instances when it is a matter of course that an unplanned child will have a serious and similarly long-term impact.

There is increasing evidence for the argument that Down syndrome is not a threat to a good family life. Many myths exist about such family lives, a prominent
one being that the parents of disabled children divorce more often than other parents. In fact, the opposite is true (Lundeby and Tossebro 2007). One might further argue that people who believe that a family member with impairments is not conducive to a good (family) life do not know what they are talking about. But the problem is that exactly the same argument can be used against autonomy and choice with regards to an unplanned child. Yet, it is not the case that one tries to convince women about to undergo abortions that a life with an unplanned child would in fact be productive and fulfilling. One accepts her/their autonomous choice, partly because these questions may have more than one answer, and partly because we believe that the person whose life and body are most centrally affected has the right to come up with the answers and choices that feel right for her.

Prenatal screening for Down syndrome provides all pregnant women with equal rights and alternatives. It represents the fair distribution of technology and information – and such universal access in Denmark prevents unequal treatment based on resources, education, age or place of residence. In Norway, in 2007, pregnant women who had higher education and were under the age of thirty-eight knew that if they told their doctor that they suffered from severe anxiety during pregnancy, they would be referred for an early ultrasound check. If this were not the case, a woman would ordinarily be offered ultrasound only in the second trimester, which is a poorer predictor of Down syndrome. If we consider the neck-fold scan and the blood test as social goods, there seems to be a fairer distribution of these goods in Denmark than in Norway.

Shifting the focus from offence to autonomy, it seems as if the Danish prenatal screening programme reflects and enhances important values in western cultures. The possibility that people with Down syndrome and/or their families might be offended to some degree must be weighed against the harm done by suppressing pregnant women’s freedom to exercise choice, as Edwards (2004) has argued. Since the feeling of being offended is a rather vague one, and since it is unlikely to result in concrete and harmful consequences such as a worsening of attitudes towards disabled people or a reduction in the standards of care, as Shakespeare has pointed out (Shakespeare 2006: 96), autonomy does seem to be the more important consideration. Offering early ultrasound and medical genetic services seems to enhance the autonomy of every pregnant woman and also improves the quality of their pregnancies. Each year in Denmark, about 60,000 pregnant women will enjoy a pregnancy that involves less anxiety and allows them increased control over their future lives. The chance of a small number of families with Down syndrome taking offence, and possibility of criticism from disability movements, cannot be decisive, if we accept this version of priorities as the correct one. Following this line of reasoning, it seems that we have arrived at a position whereby the expressivist critique of prenatal diagnosis is acknowledged as relevant but outweighed by the argument that to suppress the autonomy of pregnant women would be far more harmful.

However, could it be that the case for autonomy is too simple and superficial? Do we really know that prenatal screening for Down syndrome increases the autonomy of pregnant women or leads to reduced levels of anxiety and an
increased quality of pregnancy? Since our apparent conclusion rests substantially on the autonomy-argument, we need to investigate the empirical basis for this argument before arriving at more certain judgements.

**Ethics and screening: about pregnancy rather than disability?**

There are certain challenges related to all sorts of screening projects. In the end these challenges can be summed up under the category of ‘information’. But what gives rise to this information problem is closely connected to the phenomena of false positive and false negative screening results. Screening is often compared to fishing with a net. If the mesh is too wide, some of the fish you want to catch will escape. If the mesh is too narrow, fewer of the fish you want will escape, but more unwanted fish will be caught. So there needs to be a compromise of some kind.

In prenatal screening for Down syndrome the ‘unwanted fishes’ are the healthy babies that are unnecessarily assigned a risk label, where the neck-fold scan and blood tests lead to the mothers of these babies being identified as belonging to a high-risk group. But they are ‘false positive’, in the sense that amniocentesis would reveal that their foetuses did not have Down syndrome.² Continuing with the fish-net metaphor, there are other ‘fish’ representing another problem: the neck-fold scan will not be able to detect all foetuses with Down syndrome. About 10–15 per cent will not be discovered, in spite of this test, thus representing the ‘wanted fish that pass through the net’, those that test ‘false negative’.

False positives and false negatives generate at least three separate medical-ethical problems. When screening the entire Danish pregnant population of nearly 60,000 women, about 3,000 of them will be ‘unnecessarily’ worried, according to estimates from the Ministry of Health (Sundhedsstyrelsen 2003). So, the first problem is the one of unnecessary anxiety and a spoiled quality of pregnancy. Second, about thirty of these women will lose their ‘normal’ baby at a later stage as a result of amniocentesis procedures used only to establish that the positive ‘diagnosis’ was false; this second problem is the more well-recognised one of unwanted loss brought about by invasive diagnostics. Third, between ten and fifteen women will still give birth to a child with Down syndrome, even though they go through all the scans and blood tests; the third problem is the one of getting exactly the ‘sort’ of baby you thought you had decided not to get.

What is the relation between these three clinical-ethical challenges and autonomy? Well, suddenly ‘autonomy’ becomes a field of empirical investigation. Without going deeply into the philosophical debates on the meaning of autonomy, it seems reasonable to assert that autonomous choices have something to do with informed and free choices. A lack of information or lack of understanding of the purpose of a medical test does not foster autonomy. Similarly, a choice made on the basis of irrational anxiety and fear is not what we usually think of as the celebrated ‘autonomous choice’.

There is a small but rapidly growing body of empirical research on this subject. A 2006 study by Müller et al. concluded that nuchal translucency (NT) screening
(early ultrasound scan) for Down syndrome does not increase anxiety or depression levels in pregnancy. In fact, the study showed that women who underwent screening were less likely to be anxious compared with those who were not offered screening (Müller et al. 2006). This finding so enthused the ethicists Chervenak et al. that they felt compelled to state the following: ‘We have argued that routine obstetric ultrasound is an important autonomy-enhancing strategy … This is further evidence that first-trimester risk assessment enhances the autonomy of pregnant women without biopsychosocial harm’ (Chervenak et al. 2006: 355).

But ‘empirical evidence’ points in different directions. One study that Müller et al. use to support their findings is a 2003 Swedish study which concluded that early ultrasound screening does not cause more anxiety or concerns about the health of the baby than a routine scan later in pregnancy does. But the authors are nevertheless in doubt as to whether their conclusions are fully reliable. They observed that levels of anxiety among respondents in the control group were significantly higher than levels presented in other studies, including another study performed in Sweden at the same time. The authors suggest that information about the aim of the study with a strong focus on foetal abnormality may have made all the women more aware of the possibility that something may go wrong (Öhman et al. 2004). These elements of doubt led the authors to carry out a new study some years later using a qualitative design which focused on women’s reactions to a false positive test, with a conclusion that totally contradicts that of Müller: ‘A false positive test of foetal screening for Down syndrome by ultrasound examination may cause strong reactions of anxiety and even rejection of pregnancy. The prevalence of such reactions and possible long term effects need further investigation’ (Öhman et al. 2006: 64).

A 2007 study of the Danish screening programme found that the pregnant women’s motives for having an NT-scan were based on rationales that hindered an informed choice being made (Lou et al. 2007). The most important motives for wanting an NT-scan were to do with reassurance, choice, expectations about the scan being a happy event and, last but not least, on the idea that the test was right because of its approval by the Danish health-care system.

Put more simply, the Danish study showed that a typical behavioural pattern for pregnant women is to have an NT-scan in order to ensure that their child does not have Down syndrome. Most of the women are fairly certain about this before they come for the scan, and this contributes to the perception that the scan is a happy event for the mother and the father. From the perspective of Chervenak et al., these factors would not argue against the rationale of enhanced autonomy. Having the possibility of an early ultrasound in order to get in touch with the foetus at an earlier stage through the medium of screen and sharing a positive experience with one’s future child can be seen as an expression of increased autonomy.

Although such an understanding of autonomy might be in line with the thinking of Chervenak et al., it is definitely not in line with the view of the medical establishment, as evidenced by the strong reaction of the European Committee for Medical Ultrasound Safety (ECMUS) among others to ‘souvenir scanning’. In 2006, they stated that ultrasound scans should not be performed solely for the
production of souvenir images or for the recording of a foetus or embryo, arguing that, ‘Very little information is available regarding possible subtle biological effects of diagnostic levels of ultrasound on the developing human embryo or fetus, and the possibility of developmental effects in the brain cannot be ruled out’ (ECMUS 2006).

Of course from a medical point of view, an NT-scan has a diagnostic benefit because it can assess the risk of Down syndrome, and getting an early scan in order to have this information increases the autonomy of the individual. But if Danish women are primarily interested in prenatal screening because they want to experience a happy event, are they choosing the scan for the ‘wrong’ reason? The only acceptable motivation from a medical perspective is that women take the scan because they want to rule out the possibility of having a baby with Down syndrome and are prepared to take the ‘rational’ consequences of that risk estimate. So again, the ‘autonomous choice’ can be questioned according to whether it is (actually) informed, rational and free.

Feminist perspectives also question the idea that the greater the range of medical-technological choices, the greater the autonomy one has. The supposition is that women want tests and technological assistance in order to be better informed and more in control. But at the same time, the battery of tests and scans performed can only be interpreted by experts. The ultrasound machine devalues the former perception of risk, that of high maternal age, and a woman’s own opinion about the due date (Saetnan 2000). Only ultrasound can provide a correct answer to these questions. But not every pregnant woman is capable of understanding this technology in order to interpret the status of the foetus. The ultrasound pictures ‘lies’ in many ways, and what you see is not necessarily what you get, in the sense that one cannot really understand these images without years of advanced training. So, as this line of critique goes, women again become dependent on medical experts (mainly men) who can interpret the advanced technological results, and tell them what is going on in their own pregnancies, including what they should fear or not.

From a philosophical view, the link between choice(s) and freedom is questionable. We know that in some areas of life, more choices do not generate more freedom or autonomy. In some instances, it is in fact the opposite, with marriage being a highly illustrative example. The quality of a marriage is not increased by introducing new and freely chosen partners every day. If you believe in marriage, you believe that the practice has value precisely because it does not permit choices of this type. The same type of argumentation could be applied to parenthood. As Simo Vehmas has pointed out, parenthood is essentially an unconditional project (2002). You do not become a more autonomous or freer parent by being given the choice of throwing away your children when you become tired of them. Again, it is the opposite: good parenthood is good precisely because it is unconditional.

However, this last point raises the question of when one becomes a parent in relation to prenatal diagnosis. It is not necessary to open that discussion here but rather to make the point that there is no inevitable link between introducing the choice of neck-fold screening in pregnancy and that of enhancing autonomy.
The link can be questioned from several empirical and normative perspectives. Indeed, two other ethical challenges that follow from screening strengthen this conclusion. Hall et al. concluded in 2000 that ‘a false negative result on prenatal screening seems to have a small adverse effect on parental adjustment, evident two to six years after the birth of an affected child’ (Hall et al. 2000: 407). In Denmark this has already lead to wrongful-birth trials brought against the National Health Service (Skovmand 2005). And the potential harm of losing a perfectly healthy and wanted baby as a result of prenatal screening (and subsequent amniocentesis) that ‘everybody else’ utilises has not been empirically investigated yet.

All these aspects are specific to prenatal screening for Down syndrome but do not apply to the same degree for a high-risk strategy where maternal age is the reason for offering such medical genetic services. The main reason for this is that older pregnant women probably have a more conscious understanding of their risk. Being forty years old and pregnant, there is a substantial increased risk in pregnancy. Prenatal diagnosis can be perceived as the ‘treatment’ of an anxiety related to this risk, and not as society’s view on people with Down syndrome. Older pregnant women are fewer in number and can receive better counselling. Their choices might be better informed. Informed consent might be reachable.

To screen or not to screen?

What is challenging about prenatal screening is the ethical complexity of the phenomenon. Principal and empirical questions arise, and it is difficult to follow all the separate and sometimes interwoven threads, and still be able to take a general and balanced view. What follows is an attempt at summing up the sides of the argument, and moving towards a potential conclusion.

I have identified two major strands of ethical thinking in relation to prenatal screening for Down syndrome. The first can be labelled the disability discussion: my main focus in describing it has been on the impact from screening on the struggle for recognition by people with intellectual impairments and their families, the potential for screening to cause offence, and the relevance of the expressivist critique. The primary pro-argument for prenatal screening in the disability discussion is to give pregnant women the choice of not becoming mothers to impaired/disabled children. Screening, so the argument goes, allows more women not to have a radically different child or a child that will prevent a good family life or otherwise constitute a burden.

The second discussion can be labelled the good-maternal-care discussion. Since only a very small minority of pregnant women carry a baby with Down syndrome, and prenatal screening includes everyone, we cannot limit the focus only to those few. Prenatal screening changes how pregnancy and being pregnant are understood and experienced. My main focus here has been on the adverse consequences followed by false alarms and false negatives but also on the information and understanding of the entire pregnant population. The primary pro-argument for Down syndrome-screening in this discourse, on the other hand, is the fact that the test is
free of risk, hence the restrictions connected to amniocentesis did not come into play and more women could benefit from a reduction of anxiety early in pregnancy.

These two discussions remain for the most part separate and discussed in different journals. Philosophers, bioethicists and disability theorists tend to favour the first discussion. The principal ethical questions seem to lie there. In medical journals and in clinical ethics, one finds more of the second discussion. There is a long tradition in medicine for defining ‘good treatment’ of a patient as a matter of balancing risk against benefit. As long as the pregnant woman can be presented as the patient, and the more controversial topic of the foetus as the real patient is disregarded, this discussion is fruitful and functional.

Both discussions deserve the name ‘ethical discussions’. But what happens if we try to bring them together? Will it provide any guidance on the question of whether we should screen or not screen? An interesting effect of attempting to join the two perspectives is that the critical potential in each perspective is strengthened by the other. From a critical disability perspective, abortion is a problematic way of preventing the birth of a disabled child. But if the national health service provide a screening service that results in massive false-alarm problems, tragically false negative-problems, huge economical expenditures and selective abortions based on inaccurate information, the costs (in the extended meaning of the word) to a society which accepts the necessity of preventing a disabled child increase dramatically. This means a further devaluing of the disease or impairment/disability in question, in terms of ‘worth-living’ dimensions. The implication seems to be that these costs are acceptable, because it is vital that every pregnant woman has the opportunity to terminate a foetus with Down syndrome. Put in a utilitarian framework: Since the ‘costs’ of a prenatal screening program are substantial, and costs need to be justified, the burden of Down syndrome and the harm of getting a baby with Down syndrome have to increase. Only the possible prevention of great harm can outweigh the human and economical costs of the screening programme. The expressivist critique appears more relevant to a screening programme for Downs syndrome compared to a high-risk strategy.

Similarly, the critical potential in the good-maternal-care perspective is strengthened by bringing in the disability perspective. If we could agree that impairment is a part of life and not necessarily a tragic one, then pregnant women should experience less anxiety and the need for a battery of risk-estimating tests would be reduced. Medical technology would play a less central role in pregnancy, while the pregnant woman would still be in charge and could more easily indulge in this part of life.

The point of this chapter is not that prenatal testing for Down syndrome is unethical: it is rather that the supporters of early screening in the whole pregnant population for Down syndrome have had an all too easy time of it so far. Very seldom are they confronted with the combined critical perspectives of both the disability discussion and the good-maternal-care discussion. The combined critique suggests that early screening for Down syndrome may cause more harm than good. And the critique claims that there is an ethical relevant difference between a high-risk strategy and population-based early screening.
There exists a third way out of the dilemma of early screening for Down syndrome. This third way would be to implement early screening because of its medical benefit for the foetus. The ‘neck-fold’ which is a marker for Down syndrome, is also a marker for different sorts of anomalies that are associated with kinds of heart failure, among others, that may be treatable (Hyett et al. 1997). With a legitimate therapeutic focus, controversial aspects such as selection and negative attitudes to disabled people are downplayed. Even the opposite could be the case, since many malformations that would have led to an abortion decision earlier can now be treated and the babies would be carried to term. At the same time, therapeutic legitimating is more in line with the good-maternal-care perspective: mothers are not offered a test in order to find out whether the foetus is an enemy or a friend, but rather because it could be beneficial for the foetus.

Today, the medical challenge is to be able to prove the therapeutic benefit(s) from early screening. The benefits have to be significant should they override the combined critical perspective. Until such benefits are proven (if they ever will be), to screen or not to screen is a powerful ethical question involving deep identity questions with regards to impairment/disability, pregnancy and technology.

Notes

1 An additional motive for terminating a foetus with spina bifida is the risk of getting a child with an intellectual disability. But this motive is not directly dependent on how the identity of people with spina bifida is constructed in public. Intellectual disability is not an issue when it comes to well-situated people with spina bifida in Norwegian public life.

2 The term ‘false positive’ has generated controversies and misunderstandings in this debate because it easily leads one to conclude that a pregnant woman receives the diagnosis Down syndrome, and then this turns out to be false. If the amniocentesis test was positive, the mother had an abortion and it turned out that the aborted foetus did not have Down syndrome, this would then have been a false positive. But this happens extremely seldom, if at all. What we are talking about in ultrasound screening is getting a risk label. Risk is about statistics, and in that sense ‘high risk’ can be true even if the baby in the end is perfectly healthy. So it might be true that the term ‘false positive’ could be misleading, and because of that, the alternative term ‘false alarm’ has been proposed as preferable.

Bibliography


