Abstract

More and more medical technologies are becoming available for the early detection of congenital abnormalities, such as amniocentesis, chorionic villus sampling, and ultrasound scanning. Recent research has shown that maternal serum can provide a wealth of information on the unborn fetus and that the testing method lends itself to mass screening. Although screening will prevent a great deal of grief, such developments also lead to extreme medicalization of pregnancy. However, owing to the fact that people show a strong desire to avert every possible approaching calamity, the "tentative pregnancy" seems to be unavoidable.

Recently, a 36-year-old pregnant woman rang up the obstetrics department at our university hospital to ask whether she qualified for chorionic villus sampling in view of her age. The answer was yes. She then asked whether they could inform her of the sex of the fetus. Once again the answer was yes. Her reaction was "Good, because we've already got three daughters and if it's another girl, I can always terminate the pregnancy."

The ultimate goal of developments in science and technology is to improve the well-being of the population. It is clear that modern medico-technological developments strongly contribute to the realization of this aim. But it is also gradually becoming clear that not everything that is technically possible should be applied. How are the advanced medico-technological possibilities going to be used? It should be realized that the public often goes beyond what policy makers ever intend (as illustrated in the above example). Where do we draw the line?

These questions also apply to the development of screening technology: diagnostic procedures for early-stage detection of diseases or their risk factors. In the field of reproduction, more and more testing methods are being developed that lend themselves to the mass screening of pregnant women. Such technologies are always associated with "stress for many against profit for few": it is not possible for one person to profit without another losing out (the so-called prevention paradox). How far should we go with this? People show a strong tendency to avert every possible approaching calamity. Which tests will benefit people and who should decide? These questions form the central issue in this article.
Every woman hopes that her pregnancy will have a good outcome. Over the centuries, steady advances in obstetric technology have reduced the risks and difficulties associated with childbirth. Consequently, maternal and perinatal mortality rates have decreased dramatically. Until fairly recently, professional intervention in pregnancy was mainly limited to the birth itself. Today, however, the course of events has changed with the introduction of technologies that can provide information about the unborn fetus. For example, developments in the field of diagnosing chromosomal abnormalities are of fundamental importance. After it had been clear for some time that there was a relationship between maternal age and the chance of having a child with a chromosomal abnormality, it was proved in 1959 that the most frequent abnormality, Down’s syndrome, could be attributed to an extra chromosome in the twenty-first chromosome pair.1 In the late 1960s, it became possible to culture cells from amniotic fluid and subject them to a chromosome test to gain insight into whether the fetus had a chromosomal abnormality. Since then, such prenatal diagnosis gradually gained acceptance. At first, the test was offered to women at very great risk of having a child with Down’s syndrome. Such prenatal diagnosis is of great significance to a 45-year-old pregnant woman who has a 1 in 15 chance of bearing a child with a chromosomal abnormality. During the past 10 years, this technology has diffused downwards very quickly. At present in many countries, 35-year-old women are being offered the chance to undergo this test, and American researchers have stated that toward the end of the century all women 30 years of age and older will be advised to undergo prenatal diagnosis (2).

At the age of 35 years, the chance of having a baby with the Down’s syndrome is about 1 in 350. The chance that amniocentesis will cause a miscarriage—of a (most likely) healthy fetus—is about 1 in 150. It is obvious that as the age limit for this test becomes lower, an increasing number of women will undergo a rather serious investigation “for nothing.” May we—must we—set any limits? Recently, a 34-year-old pregnant woman visited our university hospital clinic who, in accordance with the rules of the Dutch Health Insurance Companies, did not qualify for a chromosome test in view of her age. When she was informed of this restriction she placed 1,700 guilders (US $800) on the table and said, “If you won’t do it I’ll go to London the day after tomorrow because they will do it.” Obviously, complying with her wishes would have serious consequences for the whole health care system: from that moment onward, all pregnant women would have to decide whether they were prepared to pay that sum for the test. It should be realized that choosing not to make use of this examination also has consequences for the women concerned. This became apparent in a study we conducted on older pregnant women who consciously decided not to participate in a test for the early detection of chromosomal abnormalities (8). Some of the women had had an argument with their partner because their expectations had been different (“I didn’t want to have the test, but my husband said, ‘you’ve got to have it, because I couldn’t bear to bring up a child like that.’ Then we got quite angry with each other and it was a very difficult time for me.”). From our interviews it appeared that many of the women had found whether to use chromosomal screenings a difficult decision to make, and a proportion of them mentioned that being confronted with the test had made their pregnancy “less enjoyable.” The women often felt that they had to defend

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their decision to the outside world ("You get the idea that it's no longer safe to have a baby if you're 36"; "Very soon you won't be allowed to have a baby once you're 30."). Sociologist Katz-Rothman (3) has commented on this point that "The new reproductive technologies are heralded for their choice-giving capacity, but there are also choices lost as the technologies develop."

In addition to amniocentesis, which is a fairly complicated examination performed in the sixteenth week of pregnancy, chorionic villus sampling (CVS) has been developed during the past 10 years. CVS can be performed in the tenth week of pregnancy; its results are available 2 days later (as opposed to 10 days for amniocentesis), and provide information about possible chromosomal abnormalities in the fetus. The disadvantages of CVS are that the risk of miscarriage is higher than with amniocentesis and that no information is obtained about neural tube defects. Although this means that women are given a wider choice, the many extra factors also lead to a more complicated decision process. This became apparent in a study we conducted on the decision behavior of "older" pregnant women. Said one woman: "First you hesitate about whether or not you should have a test at all and then you have to make another choice regarding which of the two methods would be best" (5).

NEURAL TUBE DEFECTS

The diffusion of the technology for the detection of neural tube defects is an interesting field. For some time it has been possible to test amniotic fluid for a raised protein level (alpha-fetoprotein [AFP]; the fetal neural tube is not covered with skin, so this substance can filter into the amniotic fluid). It is obvious that women who run an increased risk of giving birth to a baby with serious spina bifida will benefit by undergoing amniocentesis to determine the AFP level. In the 1970s, it was discovered that AFP may also be present in maternal serum; women with an increased serum AFP level are at increased risk for having a baby with a neural tube defect. This makes it possible to carry out a simple blood test (in about the sixteenth week) in order to detect women with an increased chance of having a baby with a neural tube defect. However, this method is rather inaccurate. For example, if 1,000 pregnant women are tested and the 30 highest serum AFP levels are taken as the starting point (3%), then further tests will have to be carried out on the women in this group (repeat AFP determination, ultrasound scan, possible amniocentesis); in 28-29 of these 30 women, the test results will be normal. The many false-positive test results cause a lot of emotional discomfort and psychological strain in the women concerned (1). If all pregnant women participated in a mass screening, this test would detect about 80% of the fetuses with a neural tube defect (the serum test also misses a few cases: about a fifth of the children with a neural tube defect would be born anyway to mothers with a normal serum AFP level [false-negative result]). Clearly it is difficult to inform women correctly about the diverse aspects of these tests, which means that misunderstandings can easily occur.

SCREENING

Screening is concerned with the active early detection of abnormalities or their risk factors. Well-known examples are the mass screening programs for breast and cervical cancer, high cholesterol levels, and hypertension. "The earlier you find out the better" is the popular belief. Screening tests are not carried out in people who seek help on their own initiative, but are offered to "complaint-free" individuals. Some take the form of mass invitation programs (via the newspaper or a personal letter), while others
are conducted routinely (neonatal screening for PKU/CHT, screening at health centers). The central characteristic of screening is that it leads to the detection of risk groups. These groups are then subjected to further tests, which in turn give rise to “real” cases (and a large number of false-positive cases). Serum AFP determination in pregnant women is a test very suitable for mass screening.

THE SCREENING OF PREGNANT WOMEN

There are a number of fundamental differences between the early detection of congenital abnormalities in the fetus and the early detection of other abnormalities. First, there is the difference between the therapeutic possibilities. If prenatal screening shows up a neural tube defect or chromosomal abnormalities, for which there are no normal treatment modalities, then we are left with only the possibility of a selective abortion. However, not all women find this an acceptable solution, which means that they have to be educated very carefully. A second aspect concerns the doctor–patient relationship. Putting a mass screening program for breast cancer into operation means interfering with the lives of “healthy” women who would not have thought of seeking help of their own accord. But a pregnant woman chooses to undergo medical supervision: to a certain extent she hands over her pregnancy to the physician. A number of (mostly implicit) expectations are associated with the patient–physician relationship formed in this way. For example, the patient expects the doctor to conduct tests for her own good and that of her baby. Blood tests aimed at rhesus antagonism, anemia, and so forth belong to the standard tests for which the doctor does not have to ask the patient’s consent. But if the blood test also aims at selecting women with an increased change of congenital abnormalities, then the test takes on a completely different character and explicit permission is required. The ease with which problems can arise became clear to us in a study we conducted on the educative and communicative aspects of serum AFP determination (7). The expectant women had received written information on the possibilities of the serum AFP screening, but many of them had problems in understanding it. Some of the women had more or less consciously cut themselves off from the information offered to them or had put the information folder away.

NEW DEVELOPMENTS

More and more tests are becoming available that are suitable for the mass screening of pregnant women. It is obvious that strongly invasive and labor-intensive—and therefore expensive—tests, such as amniocentesis and CVS, will not easily be attributed a mass (screening) character. But a simple blood test in the sixteenth week of pregnancy lends itself particularly well to mass application, and in many countries serum AFP values are being screened. Few people realize that in this way we have turned on to a course that will have serious consequences for all future prospective mothers. Technological solutions to one problem often seem to lead to new problems in other or adjacent areas. Subsequently, our rational technocratic society seems to require that new technological solutions be found for these new problems. This scenario is also valid for serum AFP determination in pregnant women. Recently it was discovered that a low serum AFP level also has a certain predictive value: tests have shown that a 30-year-old pregnant woman with a very low serum AFP level has a greater chance of bearing a child with a chromosomal abnormality than a “normal” 40-year-old pregnant woman. What is the significance of this new information? Now when a gynecolo-
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gist knows that a particular 30-year-old pregnant woman has an increased risk (after the result of the serum AFP test), it would be strange not to inform her. A doctor who does not inform a 40-year-old pregnant woman of the possibility of undergoing amniocentesis or CVS runs the risk of having charges brought against him or her—as happened in the Netherlands some years ago. So too does the doctor who is aware of a very low serum AFP level in a 30-year-old woman but keeps this information away from her.

Furthermore, a serum AFP test is not the end of the story: there are all sorts of new and relatively simple testing methods for detecting congenital abnormalities. Recent studies have shown that the serum concentrations of estriol and human chorionic gonadotropin are predictors of chromosomal abnormalities in the fetus. In combination with the serum AFP level, an increasing number of congenital abnormalities can be detected via mass screening. These developments are always introduced with a great deal of enthusiasm, but what are the psychosocial and societal implications? An October 1989 issue of Lancet (9) contains an article called “Detecting Fetal Cells in the Maternal Circulation.” Certain cells can be found in the blood of pregnant women that can provide information about the fetus. What are the limits of these technologies? On the one hand, prenatal diagnosis is liberating for women; on the other hand, women are confronted with new responsibilities and burdens.

ULTRASOUND SCANNING

The development of ultrasound scanning is of fundamental importance to prenatal care. This test will seriously change the position of all pregnant women. Originally this imaging technology was used at an early stage of pregnancy (weeks 8–10) to answer such questions as: Is this a real pregnancy or a pseudopregnancy? What is the duration of pregnancy? Is there more than one fetus? Many pregnant women find this (early) ultrasound scan agreeable and some request the test on their own initiative (and take home the photos and video images). The ultrasound images have gradually become clearer, which is one of the reasons why this method is steadily gaining popularity for the detection of serious congenital abnormalities (although there remains the problem of variation in readings of ultrasound scans). It is obvious that a woman who has had two stillborn babies with seriously dwarfed growth will wish to undergo a scan in weeks 16–18 so that the fetus' limbs can be measured. But every woman stands a chance of having a baby with serious congenital abnormalities that are visible via ultrasound. Why not therefore offer (late) ultrasound scans on a large scale? At present, material means are the limiting factor (it is time consuming, it requires expertise, and it is therefore expensive). But in the medical literature, appeals are published regularly asking that this investigation be more widely available (e.g., recent publications in the Dutch Medical Journal).

It is of fundamental importance that technologies are developed that will provide us with therapeutic means for intervention in cases where abnormalities are detected via ultrasound. Reports appear regularly on this topic (“Congenital heart abnormalities detected more quickly via ultrasound”; “Detecting urinary tract abnormalities in the fetus makes it possible to operate soon after birth”). If pregnant women are informed of such therapeutic possibilities, the technology will diffuse very quickly. It is not acceptable for a person to cause a child grief if he or she is responsible for it. This also became clear in a study we conducted on a group of our University Hospital women who had had their first baby a few months previously. We presented them with
the following situation: “Of all the babies born in the Netherlands, 1 in 1500 has a serious kidney disease. This disease often arises during pregnancy: the fetus has a (too) narrow urethra (the duct by which urine is discharged from the bladder) so the urine flows back toward the kidneys. With the aid of ultrasound scanning, this abnormality can be detected between the 21st and 26th week of pregnancy. If this abnormality is detected in this way during pregnancy, then it is possible during pregnancy or directly after delivery to perform an operation to prevent kidney damage. Would you undergo such a test during a future pregnancy if you were offered the opportunity?” Eighty-seven percent of the women said that they would undergo the test, 10% would not, and 3% were not sure. The road to ultrasound screening for all pregnant women at a later stage of pregnancy therefore lies open and will face many women with unexpected problems.

THE COST AND BENEFITS OF SCREENING

It is very difficult to evaluate screening technologies. Such evaluations involve a large number of variables. These variables have many different natures. As far as the non-material aspects of screening programs are concerned, it is clear that prenatal screening can prevent a great deal of grief. But this benefit cannot be obtained without harm to others. In order to detect a few congenital abnormalities, many women will undergo what later turn out to be unnecessary tests, many pregnant women will be troubled emotionally, and some women will have a miscarriage of a healthy fetus. How can these advantages and disadvantages be weighed against one another? Society lacks cognitive models that make it possible to deal adequately with this sort of technology. As far as the material aspects are concerned, policy makers usually resort to cost-effectiveness analyses, such as that made between the (simple) cost of care associated with people born with congenital abnormalities (“a child who has Down’s syndrome costs an average of about 1 million dollars”) and the high-technology cost of the early detection methods. Such comparisons, which have an unethical character, arise from the rational way of thinking so typical of Western society. It should be clear that nearly every form of prenatal diagnosis is cost-effective if it is calculated in this way. Such an approach will therefore lead to a spiral of science, technology, and efficiency with an uncontrollable character. Are we selling our souls to technology?

THE IMPERATIVE NATURE OF SCREENING TECHNOLOGY

The general public has a very positive attitude toward screening programs. A number of factors play a role in this. First, there is the tendency toward binary thinking: whether the chance of having something serious is 1 in 100 or 1 in 100,000, people argue that “a chance is a chance,” and even if the chance of a positive result is very small, they say “suppose that one person is me.” In a recent study on this topic, we presented the following question to a group of women who had had their first baby a few months previously: “Would you take your baby to a screening clinic and leave it there for 24 hours to undergo tests for the early detection (and adequate treatment) of a serious disease which occurs in 1 in 90,000 cases?” More than a third (38%) of the respondents said that they would participate, 28% were not sure, and the rest said they would not participate.

Another important aspect of offering screening technology is the fact that people want to prevent feelings of regret regarding the choices they make (“I do not want to blame myself later for not having tried everything I could”). This “anticipated deci-
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...gives screening a strongly imperative character (6). Such a basis for decisions means that we can go on more or less endlessly with offering “chance-related screening technologies.” The social climate also plays an important role in a person's decision on whether he or she should participate in a screening test. People are influenced to a considerable extent by others: nonparticipation must be explained to one's relations; its voluntary character is therefore doubtful. For example, pregnant women realize that nonparticipation can also have considerable consequences if they should give birth to a handicapped child (“It's your own fault.”). Some years ago we interviewed a woman who, at the age of 40, had decided not to make use of amniocentesis and had given birth to a child with the Down's syndrome (some weeks before our interview). She had the feeling that the people in the village disapproved of her refusal to undergo tests and this made it extra difficult for her to accept her situation. In this way early diagnosis has far-reaching effects on the total social system.

THE FUTURE

Discoveries in the microcosmos will (and must) continue. The question is, however, how are we to deal with all the new insights and knowledge? It is clear that the medico-technological developments will have far-reaching consequences for the way in which we are going to organize society. People will go to great lengths to avoid calamity. But how far should we go? Will the time come when it will be necessary to withhold certain choice-giving technologies from individuals in order to protect our collective interest? In Western society, it is considered an unacceptable form of paternalism to make this kind of decision for others. But can we expect everybody to be responsible enough to decide for themselves whether something is worthwhile? And what should we do if someone is prepared to pay for it him or herself?

A few years ago, Barbara Katz-Rothman (4) predicted in her book, The Tentative Pregnancy, that before long all pregnancies will have a tentative character: women will not be/feel pregnant until a series of tests have been carried out in the first few months and the results are all positive. Is it old-fashioned to start feeling nostalgic?

NOTE

1 In several parts of this article information is presented on the history of prenatal diagnosis technologies. These data are derived from: A. Mantingh, On CVS. Early experience with chorionic villus sampling (CVS) in the north of the Netherlands. Dissertation. Groningen, the Netherlands, 1988 (the author presents a timetable of the historical milestones in prenatal diagnosis technologies); and A. Milunsky (ed.). Genetic disorders and the fetus. Diagnosis, prevention and treatment. New York: Plenum, 1986.

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