

## Inescapable Decisions. Implications of New Developments in Prenatal Testing

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### Abstract

The main ethical principle in prenatal testing is the autonomous decision of the pregnant woman concerned. However, recent developments in prenatal testing undermine this model. The overall number of invasive\* prenatal examinations has dropped significantly. Yet, the amount of pathologic results has increased. Due to the improvement in ultrasound diagnostics the predictability of possible disabilities or diseases of the unborn child has increased substantially. As a result of this pregnant women can take the decision whether or not to undergo invasive prenatal examinations on the basis of personal risk "evidence" produced earlier on by means of non-invasive\* screening. It can be questioned, how autonomous decisions can be if they are increasingly pre-informed through upstream risk-assessments on the basis of non-invasive screening. Particularly ultrasound screening is often carried out without thorough counselling and sometimes even without consent. The concept of autonomy is difficult to uphold if women do not deliberately decide whether to undergo non-invasive screening, but the moment of such a deliberate decision comes only after positive screening results. Taking into account that public discourses have rather focused on other aspects of genetic or reproductive technologies such as stem cell research or pre-implantation diagnosis it is important to analyse how technological innovations transform medical practices without a re-articulation on a discursive level as I will try to show in this paper for the case of prenatal testing.<sup>1</sup>

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\* Medical terms are explained in the glossary and marked with a \* sign when first used in the text.

<sup>1</sup> Acknowledgement: The issues discussed in this paper were investigated in a research project called "Prenatal Testing: Individual Decision or Distributed Action" that was carried out by the author and his colleagues within the accompanying research programme (ELSA) within the Austrian Genome Research Programme (GEN-AU) funded by the Austrian Federal Ministry for Education, Science and Culture.

## 1 Introduction

It is generally assumed that in the current medical practice, prenatal testing requires *genetic* counselling\*. In particular this is true for so called invasive (or diagnostic) methods such as amniocentesis\* and chorionic villus sampling (CVS)\*. This counselling is to secure the individual and autonomous decision of the pregnant women concerned. It is she and only she who should decide matters of prenatal testing. Not least for that reason, genetic counselling plays a key role in prenatal testing and has become an obligatory requirement in the procedure. The main task for the counsellor is now to give proper information on the subject. This is to inform the client's decision and to provide medical knowledge that a pregnant woman may not possess. A counsellor will be keen to talk about the implications of available examinations, possible risks they include and their potential outcomes. Experienced counsellors emphasise the importance of stressing that prenatal testing is not a guarantee for a healthy child and that there are certain limits to such examinations that anybody who undergoes them must be aware of.<sup>2</sup> Furthermore, it has become a standard that prenatal testing will only be performed if a pregnant woman has given her explicit agreement to the procedure, confirming that she has received comprehensive information and has decided after thorough consideration thereof.

This framework of prenatal testing refers to specific ethical standards, which are largely shared among counsellors and physicians in the field (c.f. Wertz/Fletcher 1989: 28 and 2004: 36-43).<sup>3</sup> These principles aim to give

highest priority to the woman's right to decide for herself. There are certain measures to ensure that all decisions are left to the client. Most importantly this is through so-called "non-directiveness" in counselling. In short this means that the physician should serve as an informant, but must not influence or prejudge decisions in one way or another. This way "informed consent"\* should be achieved. All together: "The counsellor-client relationship is based on values of care and respect for the client's autonomy, identity, welfare and freedom" (NSGC 1991).

It has been argued elsewhere (c.f. Clarke 1991: 1000; Marini et al. 2002: 171; Wertz/Fletcher 2004: 38) that in practice it is not so easy to actually come close to this ideal. But despite all the difficulties in applying these ethical standards, they still remain the dominant reference point for physicians and counsellors in prenatal testing. However, some have taken a critical view of the underlying individualism. Elisabeth Beck-Gernsheim for example has argued that the opportunity to decide is also an obligation to do so. In fact the possibility to not decide no longer exists (Beck-Gernsheim 1990: 157; Beck 1990: 52). A personal decision becomes inescapable. But, whoever decides is responsible for what follows from that decision. It can be argued that there is a specific coupling between individual decision and personal responsibility.

Undoubtedly the guiding vision of an "autonomous decision" in prenatal testing is a powerful driving force that shapes clinical practice to the present day. However, in this article I will

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more than 75% of the investigated countries (c.f. Wertz/Fletcher 1989: 28). Having investigated 36 nations in 1993-95 Wertz and Fletcher point to the fact that "unbiased information" is a guiding norm for English-speaking nations in particular and similarly important for northern and western European countries (c.f. Wertz/Fletcher 2004: 36-43).

<sup>2</sup> Based on interview data as conducted by the author.

<sup>3</sup> Non-directiveness in genetic counselling is a concept that reaches highest acceptance among counsellors. In a survey on geneticists in 19 nations more than 75% of those surveyed agreed to this standard in

argue that medical practice is not *exclusively* a matter of social constructions. In particular I will question how independent such a decision or informed consent actually is. For that reason it is the role of the medical technologies that is examined more carefully. To what extent do the testing methods applied exercise an influence on prenatal testing? In providing an answer to this question it can be shown how technological innovations contribute to the re-organization of medical practices.

## 2 Methodology

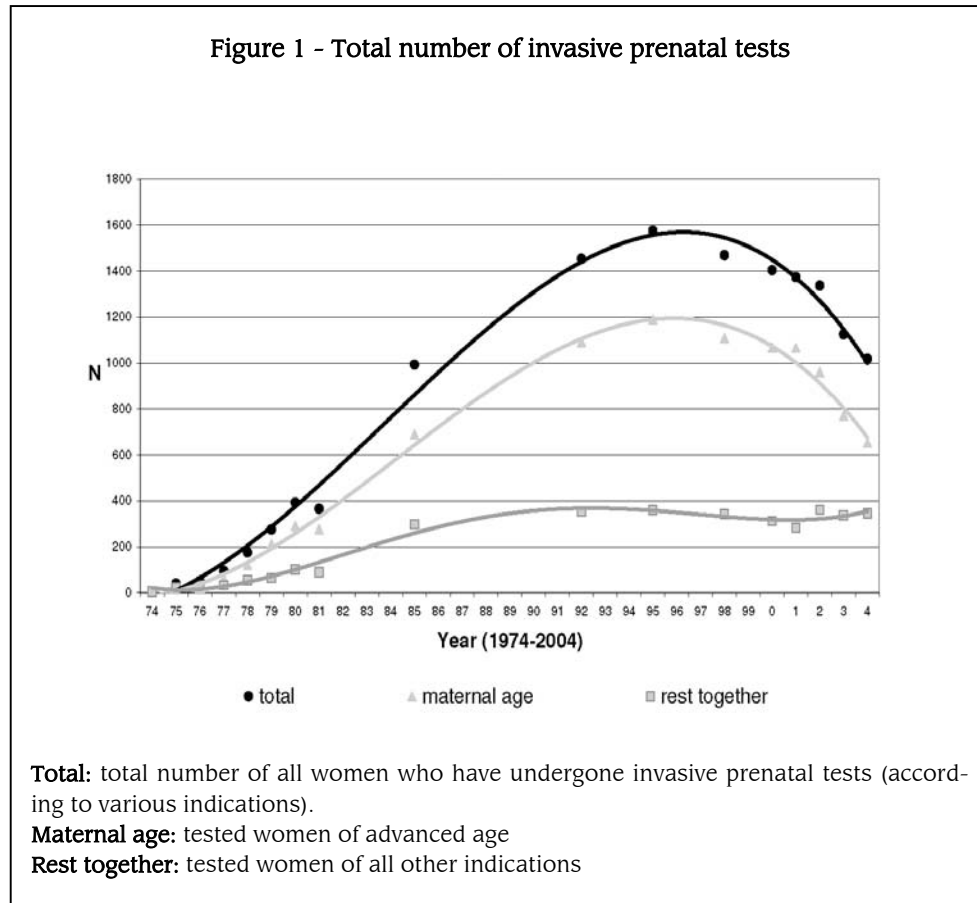
The argument presented in this paper is based on an empirical study carried out by the author and his colleagues. The study aims to examine the individual character of decisions in the course of prenatal testing and focuses on processes that contribute to an individualization of responsibility for decisions in the context of prenatal examinations. Among other research methods we have carried out expert interviews with physicians (geneticists and gynaecologists) who perform prenatal testing.

Purposive sampling was used to obtain a sample of 15 experts in prenatal testing. We have chosen interviewees who work in the major centres for prenatal testing in Austria. With one exception we could carry out interviews with physicians from all the centres for prenatal testing who run a cytogenetic\* laboratory. In addition we interviewed physicians from a province which does not have a cytogenetic laboratory on its own, but send there samples elsewhere for analysis. One geneticist was interviewed who does not carry out prenatal testing anymore, but has done so previously. Ten of our interviewees are gynaecologists who carry out the prenatal tests in cooperation with one of the cytogenetic laboratories or have one in their own department.

Semi-structured guideline based interviews were carried out by members of the research team and were audio-taped and transcribed. The interviews were carried out in the offices of the interviewees and were typically one to one hour and a half long. All interviewees were informed about the purpose of the interview and asked for consent to using the material for our study. For the qualitative analysis we used a software for data management and analysis (atlas.ti).

In the course of the interviews our interviewees provided us with data of the tests they have performed. In this paper I will analyse these data on the frequency of the tests, the indications\* they were induced, and the number of positive results. The analysis is informed by the expertise of the interviewees who explained the data to the interviewer. However, it needs to be mentioned that there are no consistent data on the *total* number of prenatal tests on a national level. Data are only available from the centres which offer such tests and willing to open up to social scientists. The figures presented in this paper are data of a particular centre, which can be regarded as a typical case. The centre is of national importance and one of the largest in the country. Our interviewees of the other centres observe a similar picture in their own context and confirm the trend we present here in the interviews we have taken. On this basis it is claimed that the presented findings give an appropriate account of prenatal testing in Austria.

The developments described in this paper can also be observed in other countries (e.g. Wray et al. 2005, Benn/Fang/Egan 2005 for the USA). However, it is difficult to generalize the practice of prenatal testing. The national health care systems are different and they provide different circumstances for antenatal care accordingly. Diffusion patterns of medical technologies vary from one country to another and also legal frameworks are



unlike to name a few parameters, which make it difficult to compare or even generalize. For that reason this paper does only claim to provide an analysis for Austria. On the other hand, medical science is highly internationalised and medical technologies are commercialised on a global market. Several of the Austrian specialists in prenatal testing have worked in other countries and learned from their colleagues abroad. For that reason developments in prenatal testing take place on an international scale, but they are situated in specific national or even regional contexts. This paper will bring together both dimensions and thereby exemplary show how international developments in diagnostic technologies are integrated in a national medical practice.

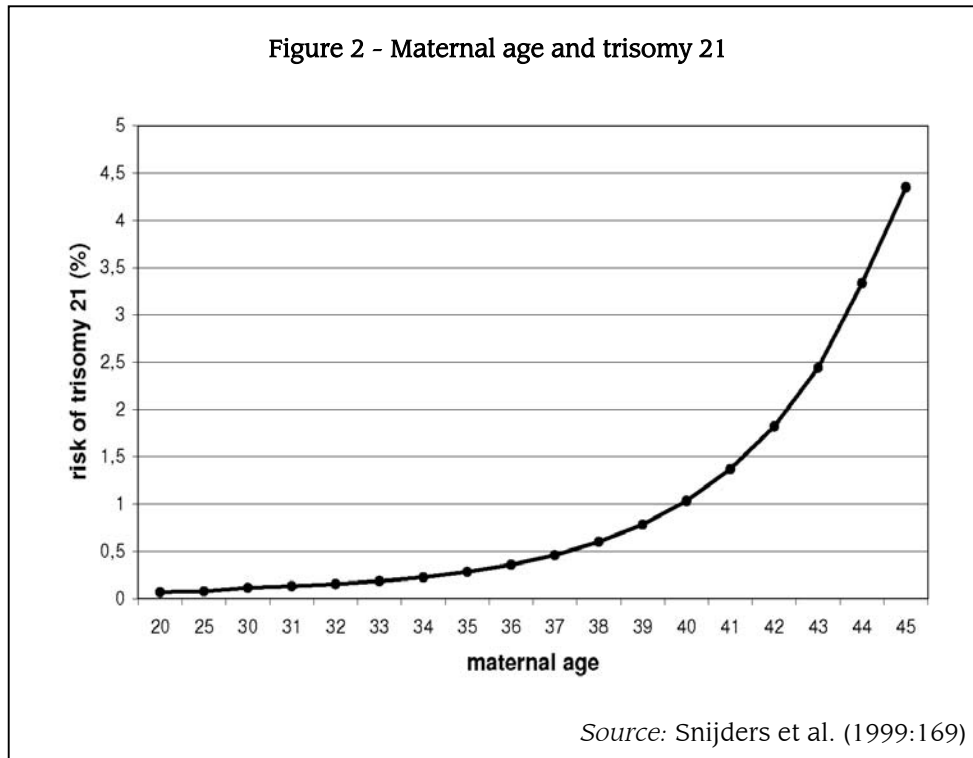
### 3 Medical background on the developments of testing methods

During the last ten years, quite a remarkable development can be observed in prenatal testing. The overall number of prenatal testing by means of amniocentesis and CVS is declining dramatically. In certain Austrian hospitals the total number has decreased by one third of its peak in 1995. Geneticists report that in Germany a decline of 50 percent can be observed<sup>4</sup> and a recent studies refers to a similar change in the USA (c.f. Wray et al. 2005: 353; Benn/Fang/Egan 2005: 328, Benn et al. 2004). In order to offer an explanation for the dramatic change in the frequency of invasive prenatal testing it is argued that the medical technologies applied play a major role. Decisions of pregnant women and

<sup>4</sup> Based on interview data as conducted by the author.

related counselling processes cannot be understood appropriately if technologies and the medical data they produce are seen as neutral or even negligible as an influence.

age. Women of 35 years and older show a higher risk of chromosome aberrations\* (trisomies\* or translocations\*). As one can see from the diagram above (figure 1) up to three



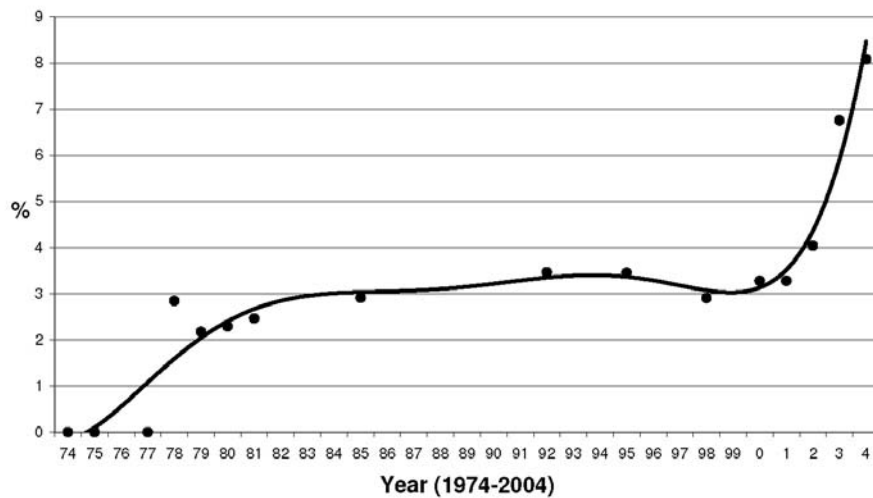
Taking a closer look on the diagram above (figure 1)<sup>5</sup>, it becomes obvious that the dynamic of the total number of invasive prenatal testing is determined chiefly by just one factor: maternal age. Any test has a justification why it is undertaken and if the justification is "only" that a particular woman wants to have it. But most of the time there is a medical justification, i.e. a higher risk of foetal anomalies compared with that on average. If this is the case and there actually is such a higher risk, in medical terms this is called an "indication". For example a prenatal test could be indicated because a woman has previously given birth to a disabled child. Yet, in most cases the indication for a subsequent prenatal test is maternal

quarters of all invasive prenatal tests have been undertaken for that very reason (between 1978 and 2003 the ratio did not drop below 68% and reached its peak in 2001 at 77.6% in the presented case). Advanced maternal age is regarded as an indicator because of the correlation as described here: "Generally the probability of a birth of a child with Down's syndrome (and many other chromosome anomalies) increases with the age of the mother. The so-called age indication following from the fact that this is the most frequent reason for a prenatal chromosome analysis" (Sancken et al. 2003, my translation). A diagram (figure 2) shows the correlation between birth giving age of the mother and a higher rate of chromosome aberration; trisomy 21 in this case.

Generally it is true that women of advanced maternal age do have a higher risk. But for some reason this

<sup>5</sup> The figures refer to empirical data from a specific clinic, which remains anonymous here.

**Figure 3 - Chromosome aberration in relation to invasive prenatal tests performed**



Pathologic results with chromosome aberration in percent of all women tested with invasive methods.

correlation no longer plays such a significant role in the management of antenatal care. Advanced maternal age is not indicating an invasive prenatal test to the extent that it did only a few years ago. This becomes clear when considering the dramatic decline in the number of examinations indicated by advanced maternal age. At this point the question arises: why is that the case?

The first point to mention is that, if age is the chief indicator for a higher risk, many pregnant women will be tested. In Austria the proportion of women who are of advanced age (35 years and older) and give birth to a child is 15% (Statistik Austria 2004: 68).<sup>6</sup> This equates to 11,856 women in the whole country in 2002. However, even if, theoretically, all pregnant women of advanced age would be tested, the

<sup>6</sup> Only in 1981 the proportion of women giving birth to a child at the age of 35 or older was 6,4%; a total of 6.016 women (Statistik Austria 2004: 68). According to a recent publication in Germany the proportion of pregnant women of advanced maternal age is about 13%" (c.f. Sancken et al. 2003).

majority of trisomy 21 pregnancies will not be detected that way. This argument was already made at the time when maternal age was sharply on the rise. Wald et al. explained that "in practice fewer than 15% of affected pregnancies are detected because fewer than half of these older women actually have amniocentesis" (Wald et al. 1988: 883).<sup>7</sup>

Equally Saller and Canick pointed out in the mid 1990s, when age indication reached its peak: "by only offering amniocentesis to women older than age 35 only approximately 20% of foetal Down's syndrome will be detected" in practice, and furthermore they conclude: "This approach results in failure to detect the approximately 80% of babies with Down syndrome born to women younger than age 35." (Saller/Canick 1996: 784) Meanwhile the proportion of women who have babies at a later stage of their life has increased. Consequently the percentage of possible detection\* with age as

<sup>7</sup> This has been claimed for the UK where 5% of women are aged 36 or greater when being pregnant (Wald et al. 1988: 883).

an indicator has increased and is estimated at about 30 to 40 percent if all women 35 and older would be tested (c.f. Sancken et al. 2003). Despite the fact that maternal age has increased on average the majority of trisomy 21 pregnancies are still conceived by women younger than 35. Some have concluded on the basis of this that "with regard to the total population of pregnant women age indication is neither particularly sensitive nor very specific" (Sancken et al. 2003). In short, from a medical point of view maternal age is an indicator, but it is not a very good one. One could even say that it is not much more than a qualified guess that about four out of a thousand 35 year old pregnant women are carrying foetuses with Down's syndrome.

Following the described logic a more accurate indicator than maternal age is desirable. The point now is, that meanwhile there are better indicators that help to identify for an amniocentesis or CVS those women who have more reasons to undergo such examinations than simply being of advanced age.

When looking at the data one can observe that even though the total number of invasive prenatal tests has gone down by about 40%, the number of foetuses diagnosed with chromosome aberration is still going up.<sup>8</sup> Figure 3<sup>9</sup> shows how significant this development is by relating the number of test results with chromosome aberrations to performed examinations. If this development is a result of the availability of better indicators, so what are these better indicators? The answer to this question is: non-invasive prenatal testing is a better

indicator for subsequent invasive examinations than simply maternal age alone.

Historically it was the so-called triple-test\* that first provided a better judgement of the likelihood that a foetus would show anomalies. The problem with this triple-test is that it produces a relatively high number of false results as Babbur et al. point out in a recent publication: "For the screened population, to achieve an 88% detection rate using the triple test alone, the predicted FPR [false-positive rate\*] would be 20%. Conversely, for an FPR of 4,8% using the triple test alone, the detection rate would be only 60%" (Babbur et al. 2005: 465). For that reason, the triple-test was controversial among physicians, and some Austrian hospitals decided not to use it any longer.

Also in the literature the triple-test is contested: "The original age indication limiting prenatal diagnosis to women over 35 has now largely disappeared in practice, so that every pregnant woman is notified of the possibility of PD [prenatal diagnosis] by her gynaecologist. The expansion of the use of PD has been driven by the so-called 'triple test', which makes it possible to identify an increased risk of chromosomal anomalies in a foetus from an examination of the mother's blood. Although this test is under severe criticism from human geneticists for its lack of validation and frequent false positive (and negative) results, it is offered by many gynaecologists as a 'safety first' test, which is often followed by a (frequently unnecessary) amniocentesis" (Hennen et al. 2000: 9).<sup>10</sup>

At first maternal serum\* screening was offered as an alternative option for those women who did not accept the

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<sup>8</sup> 46 chromosome aberrations out of 1067 tested women of age indication in 2000; 76 chromosome aberrations out 771 tested women of age indication of 2003.

<sup>9</sup> The figures refer to empirical data from a specific clinic, which remains anonymous here.

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<sup>10</sup> Translated by the authors in an English summary of their working report ([www.tab.fzk.de/en/projekt/zusammenfassung/ab66.htm](http://www.tab.fzk.de/en/projekt/zusammenfassung/ab66.htm); last access 07. Mar. 2006).

risk of amniocentesis or CVS. Yet, once these tests became available, they also gained the function of providing a more profound risk assessment for those women who wanted to have additional information prior to making a decision about having amniocentesis (c.f. Newberger 2000). Indeed maternal serum screening is still advocated as a useful means to reduce the number of amniocenteses (c.f. Rosen et al. 2002).

Meanwhile, ultrasound has also become more important, largely due to recent improvements. Initially, ultrasound was not precise enough to detect Down's syndrome fetuses (or other chromosome aberrations), but this has changed with technological advancements in the picture quality. At first, ultrasound was used at a later stage of the pregnancy and it was used for Down's screening only in cases where an indication was already given. Second-trimester\* ultrasound assessment (15-23 weeks) was offered to women with increased risk "for trisomy 21 based on advanced maternal age or abnormal maternal serum biochemical marker screening, who either had declined amniocentesis or chose to have a sonogram before deciding whether to undergo amniocentesis" (Vintzileos et al. 1996: 949). In this case ultrasound serves as a second step to provide further evidence in an already ongoing risk assessment.

But, there is a tendency to turn this upside down. Ultrasound increasingly becomes the first step. "Increased fetal nuchal translucency is associated with chromosomal abnormalities, many fetal defects and genetic syndromes" (Souka et al. 2001: 9). Ultrasound measurement of nuchal translucency\* is therefore used as a first-trimester screening test. Nuchal translucency first-trimester screening has a detection rate of about 76,8% at a false-positive rate of 4,2% (c.f. Krampfl 2005: 86). In other words, three quarters of all fetuses with trisomy 21 can be identified that way, but this rate can be increased if other biochemical tests are

performed additionally (called the combined test\*). Together with an analysis of maternal serum ( $\beta$ -HCG<sup>11</sup>, PAPP A<sup>12</sup>) the detection is as high as 87% with a false-positive rate of 5% (c.f. Krampfl 2005: 86). This can be further increased if an ultrasound of the nose bone is also performed.<sup>13</sup> Geneticists claim that it can be as high as 95 to 97%.<sup>14</sup>

Not only are there several alternatives in prenatal testing, they are also linked together: "Women who are screen-positive in the first trimester can elect to receive cytogenetic testing of a chorionic villus biopsy. The first trimester test could also, theoretically, be combined with the second trimester maternal serum screening test (integrated screening) to obtain even higher levels of efficacy" (Benn 2002: 1).

The point now is that pregnant women older than 35 do not need to decide whether or not to undergo an amniocentesis or CVS just on the basis of an anonymous statistical probability. Now they can take this decision on the basis of personal risk "evidence" provided by non-invasive testing. And if this is the case – that such a non-invasive screening produces a higher risk factor – results of the screening would justify a subsequent amniocentesis or CVS.

Currently there is a trend in invasive prenatal testing that such tests are performed less often just because of a

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<sup>11</sup> Human Chorion Gonadotrophin

<sup>12</sup> Pregnancy Associated Plasmaprotein-A

<sup>13</sup> In case the result of an ultrasound examination between 11 and 14 weeks is the absence of the nasal bone (broad nasal bridge syndrome) this is understood to be an indicator for foetal abnormalities. "Research results indicate that including this marker along with the mother's age, baby's age, nuchal fold measurement, and blood tests can bring the accuracy of the risk assessment up to 97 percent." (Baby-Center 1997-2004b)

<sup>14</sup> According to interview data conducted by the author.



higher risk due to maternal age (an indirect guess based on statistics), but increasingly because of a higher risk due to risk "evidence" (based on previous tests). And this is a significant difference. Such "evidence" is still no proof, but from a medical point of view it is a much better risk assessment than a "guess" based on maternal age alone. The decline of invasive prenatal testing indicated by maternal age (figure 1) shows how outdated the latter has become as an indicator for invasive prenatal testing. The observed tendency can be related to the increased application of non-invasive screening tests. Taken together a shift can be observed. This includes a shift from maternal age indication to testing as a result of prior screening, but a series of other changes are evident, too.

1. Increasingly there has already been some sort of examination – on the basis of which a higher risk is determined – before invasive testing is carried out. Maternal age, on the other hand, is a presumption without more specific empirical evidence for the specific case. Now that maternal age has lost its importance as an indication for invasive prenatal testing, other indicators have become more important.
2. These other indicators which have become more important are produced by means of non-invasive screening methods, mostly ultrasonic tests (and the combined maternal blood test) as already mentioned. And because non-invasive tests have become more important and more reliable, the total number of amniocentesis and CVS has gone down dramatically.

*"For many years, research on maternal blood tests has been performed. A tendency towards pre-selection on the basis of non-invasive methods can be noted. This way, an earlier start, higher effectiveness and higher accep-*

*tance can be achieved as well as a distribution of prenatal testing across all stages of age" (Arbeitsstelle Pränataldiagnostik/Reproduktionsmedizin 2004).*

3. As mentioned in the previous quote, the current shift in prenatal testing represents also a shift towards testing at a much earlier stage, from second to first trimester of the pregnancy. Time is in fact a major factor in prenatal testing and many have argued (c.f. Katz-Rothman 1989) that the earlier the testing the better it will be. Not only does the time a woman has to wait for the result matter (which is a major disadvantage of amniocentesis), but also the advancement in the pregnancy makes a difference. If an abortion is to be considered, this is easier to cope with before the 24 week of the pregnancy when a curettage\* can still be performed.
4. The tendency is noted that non-invasive techniques lead to an expansion in prenatal testing. A growth in participation can be concluded since the number of invasive tests has gone down but the number of pathologic results has increased significantly at the same time (see figure 3.). Most likely this is explained by more young women deciding in favour of a non-invasive screening.
5. Another reason for the expansion of prenatal testing can also be related to the fact that invasive tests require a specialized laboratory that is able to carry out the analysis. In Austria there are only a few of such laboratories.<sup>15</sup> Non-invasive techniques, however, already have a much higher dis-

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<sup>15</sup> In fact the common practice in invasive testing is that the sample was taken not only in clinics which had a cytogenetic laboratory. Those who did not have a laboratory on their own would send it to one of these institutions for the analysis.

semination and can be performed in a number of clinics.

6. Non-invasive screening tests also lead to "inescapable decisions". Generally, all couples are at risk of conceiving a child with chromosome aberrations or other anomalies, but a statistical probability does not tell who will and who will not. Advanced maternal age is a hint that the probability increases with age, but it does not tell who will actually give birth to a child with chromosomal anomalies. Only an invasive test can answer this question. But now non-invasive screenings provide an individual risk figure. By these means, decisions are increasingly taken on the basis of individually determined risk and not any longer on an anonymous statistical correlation, which could distinguish a certain age group at best. In this sense, one could say that invasive tests have become more medicalized, because decisions no longer start from zero.
7. Finally a last shift can be noted. The decision-making process is divided into a number of small decisions. There is no such thing as "one" decision, where everything is decided at once. In other words, decisions are by no means a priori, i.e. without pre-conditions. On the contrary, decisions are taken step by step with one leading to another: first the individual risk is determined, then a definite diagnosis is produced and only then does the question arise about whether or not to abort the pregnancy.

As the presented data show the shift described has only come about only recently. In the second half of the 1990s a stagnation of performed invasive prenatal tests can be noted. However, that this development would turn into a decline became clear only after 2001 when tests induced by age indication significantly dropped.

Obviously the technologies which have lead to the mentioned changes in prenatal medicine have been developed several years earlier. Nuchal translucency measurement was developed in the 1990s and combined with maternal serum markers thereafter (c.f. Nicolaides et al. 1992; Brizot et al. 1994). Until the innovation could exercise its full impact in Austria, physicians had to be trained (in London at King's College), but also the knowledge about the existence of the new method and who is able to perform it had to be communicated. Furthermore it takes some time to change the regime of antenatal care in which pregnant women are informed about diagnostic possibilities and referred to respective specialists. However, meanwhile the shift has become evident. One of our interviewees explained that they have a participation rate of 99% of all women who give birth in their hospital. A physician from a centre in another province of the country said: Yes, we offer non-invasive screening to all pregnant women. They only come to me after the screen. And from a third centre for prenatal testing we know that the number of chromosome aberrations diagnosed (with invasive methods) has doubled since 2000. This means that also the number of tested women has at least doubled taking into account that more and more young women are screened with nuchal translucency measurement. Although comprehensive data of the entire country on the participation of pregnant women in antenatal screening programmes are not available there is clear evidence for a substantial diffusion of non-invasive screening methods.

#### 4 Discussion

It is argued that the observed shift in prenatal testing leads to an individualization process. From a medical point of view it might be desirable to have a more accurate risk assessment, which allows for a better management of invasive prenatal testing (decisions

seem to be evidence based and therefore more rational). But on the other hand the determination of an individual risk can also be seen from a sociological point of view.

The notion individualization was prominently introduced by Ulrich Beck in his book "Die Risikogesellschaft" (Beck 1986) and Elisabeth Beck-Gernsheim (1990) has discussed individualization in the context of reproduction. Already when the concept was introduced it was argued that individualization does not only lead to an increased freedom of choice, but that it would also lead to a pressure to take decisions. Furthermore Beck argued that individualization is paradoxical and includes what he has called "control" or "reintegration dimension" (1992: 128). From this point of view prenatal testing means the integration of pregnant women into a control regime. Indeed, with the expansion of ultrasound screening more and more pregnant women are integrated into a medical network, which determines their individual risk for foetal anomalies. In his Book "Discipline and Punish" (1975) the French philosopher Michel Foucault has defined individualization as the identifiability of the individual. Following this understanding of individualization it can be argued that the prenatal regime aims to differentiate pregnant women and this differentiation is predominantly carried out in terms of risk. The place they are allocated to is derived from a sophisticated risk-assessment.

Nevertheless, this does certainly not mean that pregnant women are not free to decide. It is essential for prenatal testing that it is the women concerned who decide about their own pregnancies. This results in a fundamental paradox: There are more and more possible decisions, but these possibilities are produced in a systematic way. Thomas Lemke has argued that decisions become increasingly choices between options, but the options are pre-given beforehand and

structured by pre-decisions (c.f. Lemke 2000: 243). It is increasingly impossible *not* to choose because it is harder and harder to escape the risk assessment by means of ultrasound screening. Choices promise freedom, but the confrontation with one's individual risk demands a decision which takes the screening results into account.

Whether a chromosome anomaly is actually given or not can only be clarified through an invasive diagnostic step (cytogenetic methods are not based on statistical methods, but they provide definite results at a very low rate of false results). However, such invasive tests are bound to a risk of miscarriage (c.f. Evans/Wapner 2005). For this reason many women want to avoid an invasive examination. But as it has been argued in this paper such decisions whether or not to undergo an amniocentesis or CVS are increasingly bound to an upstream risk assessment. Hence these decisions are *not* taken without conditions, they are taken under consideration of medically generated data.

The point now is that these risk-data are produced in a framework which substantially differs from invasive testing. Our interviews suggest that increasingly genetic counselling is only performed in cases of positive screening results (prior to invasive testing). Decision autonomy is still the central ethical orientation in prenatal testing, but the consent procedure prior to non-invasive screening is largely formalized and often women don't realize the significance of the examination. Women are still asked to decide and they are explained that it is their own personal decision, but the moment of such a deliberate decision comes only after positive screening results. It is questionable if such decisions can be accounted "autonomous" if they are taken against the backdrop of a risk-assessment produced by medical examinations.

Apparently the described technological transformation in prenatal testing goes along with significant changes in the practice of its application. However, the assumption that "technical change causes social change" is usually labelled as technological determinism (MacKenzie/Wajcman 1985: 5). Günter Ropohl talks about "consequential technological determinism" (1991: 193). The criticism is, that technology is nothing original, but technology itself is socially created.

Nevertheless, one may ask, why it is possible to carry out non-invasive testing in a different organizational framework than invasive testing: Why is it possible for gynaecologists to perform nuchal translucency measurement without thorough conversation about the significance of the examination, whereas geneticists still insist on profound genetic counselling prior to amniocentesis? Both test methods are predictive malformation diagnostics. Thus the differences in the practice of their application require an explanation.

The risk of miscarriage caused by the examination is a substantial difference between invasive and non-invasive testing that can be related to the technology. Bettelheim et al. specify the risk of miscarriage (including intrauterine foetal death) to be 0.44 for Amniocentesis and 0.99% for CVS (2002: 119). Not least for liability reasons an essential task of the counselling interview is to inform the pregnant women about the risks and limitations of the examinations themselves. But if the risk that the performed examination may cause a miscarriage can be precluded there is no need to seek for "informed consent" for *that* reason.

There are certainly more reasons for counselling prior to prenatal testing. Doubtlessly the ethical model of decision autonomy needs to be understood in its historical context, namely as a possibility to overcome classical

eugenics and the catastrophe of Nazism. Nevertheless, against the backdrop of the changes in the current practice of prenatal testing, it is also important not to disregard the efficacy of the technology applied.

## 5 Conclusion

The aim of this paper was to highlight the significance of the emergence of new prenatal screening technologies and to show their impact on medical practices. The role these screening technologies play, becomes evident when relating their development and dissemination to the frequency of the total number of invasive prenatal tests. By emphasising the role of technology the question arises: how autonomous can a decision be? To what extent can a decision about whether or not to have amniocentesis (or CVS) be regarded as autonomous if it is taken on the basis of previous risk assessments determined by screening. The concept of autonomy is difficult to uphold if diagnostic technologies apparently have such a strong impact.

The decision about whether or not to undergo invasive tests increasingly depends on medical evidence. Such medical evidence is produced from the first moment a woman learns about her pregnancy. It is largely through improvements in ultrasound screening and other non-invasive tests that maternal age loses its importance as the chief indicator for an invasive test. The corresponding decline of amniocenteses or CVS shows that non-invasive screening methods serve as a means of risk assessment further upstream. But it is not only pregnant women of advanced age who can now decide on the basis of a previous risk assessment. More and more pregnant women undergo such a "pre-test" in screening programmes. It has been argued that the new diagnostic technologies contribute to an expansion of prenatal testing even though invasive tests are performed less frequent. This

way the character of prenatal testing becomes quite different; it can be argued that ultrasound changes the function of invasive tests. Amniocenteses and CVS are increasingly carried out to confirm a qualified suspicion produced by a risk assessment upstream.

In the introductory section of this paper it was stated that the highest priority is given to the autonomy of the patient, and that the goal of counselling is to ensure this for individual decision made by pregnant women. Having argued that decisions are increasingly "pre-informed" through upstream risk assessments on the basis of non-invasive screening, the question arises as to how patient autonomy and individual decisions go together with current developments in prenatal testing. Is the philosophical concept of an autonomous subject still appropriate for the problem at stake, and if not, are there alternatives? What does all of this mean for the concept of informed consent? And last but not least, what does the current development mean for counselling practices? As a matter of fact there is not much counselling involved prior to ultrasound or maternal serum screening. In many European countries prenatal screening is offered routinely,<sup>16</sup> but increasingly genetic counselling is only performed in cases of positive screening results (prior to invasive tests). The practice of prenatal testing has changed considerably since such new screening methods as described above are available. Despite its significance this transformation has not received much public recognition in Austria to this point. Public discourses have rather focused on other aspects of genetic or reproductive technologies such as stem cell research, human cloning or pre-implantation diagnosis. Taking this into account it is even more important to analyse how tech-

nological innovations transform medical practices without a re-articulation on a discursive level as it was tried to show in this paper for the case of prenatal testing.

## 6 Glossary

**Aberration (chromosomal):** the medical term for an abnormal set of chromosomes. Humans have 46 chromosomes in each cell. Too many or to few chromosomes in the cells are associated with particular disabilities and diseases. This is also the case if the chromosomes are damaged, a piece is missing or too much in the set of chromosomes. See also chromosome, translocation, and trisomy.

**Amniocentesis:** a diagnostic test, usually carried out between 14 - 17 weeks of pregnancy, in which a needle is inserted through the abdomen to remove a sample of amniotic fluid containing cells from the developing baby for testing. Either the chromosomes of the cells are examined for prenatal diagnosis of genetic abnormality or DNA is prepared for analysis. Ultrasound is used to guide the needle. The procedure carries a risk of miscarriage of about 0.5%.

**Chorionic villus sampling (CVS):** a diagnostic test normally performed between 9 - 11 weeks of pregnancy. A needle is inserted through the mother's abdomen or cervix and is used to remove a small amount of placental tissue (afterbirth). Foetal chromosomes or DNA can then be examined similar to amniocentesis. The procedure carries a risk of miscarriage of about 1%.

**Chromosome:** a piece of densely packed DNA containing hereditary information of the organism. See also aberration, translocation, and trisomy.

**Combinet test:** a non-invasive screening method, which combines nuchal translucency measurement and maternal serum test. In such a way the predictability of chromosomal anomalies can be increased up to 87%. If an ultrasound of the nose bone is also performed, the predictability can be further increased to 95-97%.

**Curettage:** a surgical abortion refers to the procedure performed by a doctor to remove the pregnancy from the uterus. In early pregnancy the surgical abortion is called a vacuum aspiration or a suction curettage.

**Cytogenetics (cytogenetic testing):** the study of the structure, function, and

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<sup>16</sup> A remarkable exception to this rule are the Netherlands.

- abnormalities of human chromosomes and chromosomal anomalies. For prenatal testing a sample of foetal cells is required, gained either by amniocentesis or chorionic villus sampling (CVS).
- Detection (rate): it is not possible to identify all chromosome anomalies with non-invasive screening methods. The detection rate indicates the ratio of the cases that are identified by means of a particular test in relation to all cases that are actually given.
- False negative: test results which suggest that a foetus does not have a condition which it later turns out to have.
- False positive: test results which suggest that a foetus has a condition which it later turns out not to have.
- Genetic counselling: A process by which information is imparted to those affected by or at risk of a genetic disorder. It includes information on the nature of the disorder, the size and extent of genetic risks, the options, including genetic testing, that may help clarify the risks, and the available preventative, supportive and therapeutic measures. In the context of genetic testing it may include responding to the concerns of individuals referred and their families, discussing the consequences of a test, and help choose the optimal decision for themselves, but not determining a particular course of action.
- Indication: the reason or justification for carrying out a diagnostic or therapeutic measure.
- Informed consent: permission given by an individual to proceed with a specific test or procedure, with an understanding of the risks, benefits, limitations, and potential implications of the procedure itself and its results.
- Invasive methods: amniocentesis, chorionic villus sampling (CVS) or other procedures to take a sample of foetal cells. Such invasive methods of sample taking are necessary for diagnostic testing, i.e. the analysis of the chromosomes or sequences of the DNA.
- Maternal serum: refers to maternal blood in the context of prenatal testing. "Maternal Serum (Screening) Tests" are blood tests performed to determine the risk of foetal anomalies (such as neural tube defects and chromosomal anomalies). The calculation is based on the levels of alpha-fetoprotein, estriol, and human chorionic gonadotropin in the mother's blood during pregnancy. These indicators can be measured alone (triple test) or together with a nuchal translucency measurement (combined test).
- Neural tube: a structure in early foetal life that develops into the brain, spinal cord, spinal nerves and spine. Defects of the neural tube are severe conditions and sometimes lethal. To the present knowledge, they are not genetic, although it is assumed that they might be.
- Non-invasive methods: ultrasound examinations or maternal serum tests where no foetal cells are taken. Such non-invasive methods allow for probabilistic tests in order to determine the risk of foetal anomalies (such as neural tube defects and chromosomal anomalies). This form of prenatal testing is also referred to as screening. See also maternal serum and nuchal translucency measurement.
- Nuchal translucency (measurement): the nuchal translucency test is used to determine if a woman is at high risk of having a baby with a chromosomal anomaly.
- Translocation: an anomaly, which has moved one piece of a chromosome to a different position. Translocations can be balanced (the set of chromosomes is complete, but does not have the proper form) or unbalanced (a piece of a chromosome is missing or too much in the set of chromosomes).
- Trimester (first, second or third): any of the three 3-month periods into which pregnancy is divided.
- Triple test: a maternal serum test used to determine the risk of foetal anomalies (such as neural tube defects and chromosomal anomalies). The calculation is based on the levels of alpha-fetoprotein, estriol, and human chorionic gonadotropin in the mother's blood during pregnancy. Also called "Multiple Marker Test".
- Trisomy: three copies of a single chromosome in the cells. The most common form is trisomy 21, the so called Down's syndrome.

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