

**ETHICAL ISSUES OF PRENATAL SCREENING FOR  
DOWN'S SYNDROME**

**PhD Thesis**

**Adél Tóth, MA**

**Department of Medical Genetics  
Medical Faculty  
University of Szeged, Hungary**

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## List of publications related to the PhD thesis

### Papers published in journals with an impact factor:

- I. **Adél Tóth**, Tibor Nyári, János Szabó. Changing views on the goal of reproductive genetic counselling in Hungary. *European Journal of Obstetrics and Gynecology & Reproductive Biology* 2008;137(1):3-9. **IF: 1,273**
- II. **Adél Tóth**, Péter Szeverényi. Interpretation in reproductive genetic counseling: a methodological framework. *Journal of Psychosomatic Obstetrics and Gynecology* 2007;28(3):141-145. **IF: 1,123**

### Further papers related to the subject of the Thesis:

- III. **Tóth Adél**, Szabó János dr. A tájékoztatás legfontosabb etikai vonatkozásai a Down-szindróma praenatalis szűrése során. [Ethical aspects of disclosing information on prenatal screening for Down's syndrome.] *Orvosi Hetilap* 2005;146(6):259-265.
- IV. **Tóth Adél**, Szabó János dr. Miért nem lehetséges a semleges jellegű genetikai tanácsadás? [Why is non-directive genetic counselling impossible?] *Magyar Nőorvosok Lapja* 2005;68(0):113-120.
- V. **Adél Tóth**. The role of values in genetic counselling. *Studia Bioetica* 2005; Volume 2: 1-7. <http://utopia.duth.gr/~xirot/BIOETHICS> - accessed 10/05/2009
- VI. **Tóth Adél**, Szabó János dr. A Down-szindróma praenatalis szűrésének néhány etikai vonatkozása. [Ethical implications of prenatal screening for Down's syndrome.] *Orvosi Hetilap* 2000;141(42):2293-2298.
- VII. **Tóth Adél**. Az értelmező orvosi etika szerepe a Down-szindrómás gyermeket nevelő családok és az orvosaik közötti kapcsolat kialakításában. [Role of interpretive ethics in building relations between physicians and families having children with Down's syndrome.] *Pediáter* 2000;9(4):297-305.
- VIII. **Tóth Adél**. A Down-szindróma prenatális diagnózisát követő terhességmegszakítás etikai kérdései. [Ethical issues of pregnancy termination following the diagnosis of Down's syndrome.] *Pediáter* 1999;8(4):311-316.

### Paper related to the ethical analysis method of the Thesis

- IX. **Adél Tóth**. The birth of bioethics and its basic principles. In: Bettina Pikó (ed.): Introduction to medicine. Basic principles of behavioral sciences and preventive medicine. Szeged: University of Szeged; 2004. p. 133-145.

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## Summary

**Introduction:** In the last three decades, prenatal Down's syndrome screening, which provides women with information about their individual risk of having an affected pregnancy, has become one of the most rapidly evolving fields of medicine. The development has resulted in the introduction of formal screening programmes in several countries and has promoted the spread of *ad hoc* screening in some others.

**Objectives:** Studying the ethical impacts of prenatal Down's syndrome screening is deemed essential, as while screening creates new perspectives for the parents to tackle the problems of Down's syndrome, it also induces new complex problems. The objectives of the PhD thesis are to answer the question whether the implementation of prenatal Down's syndrome screening is ethically justifiable at all, and, if the answer is positive, to explore the moral issues that should be taken into account through its application.

**Methods:** In those territories where the Thesis has added new information to the previously existing knowledge, the biomedical ethics approach, the interpretive ethics approach, and the sociological method of a questionnaire study were applied. Where data were collected on already existing knowledge, literature review was performed.

**Results:** By analysing the competing goals of prenatal screening (prevention and enhancing autonomy), the PhD thesis has contended that the implementation of prenatal Down's syndrome screening is ethically acceptable, provided that its goal is defined as enhancing the parents' reproductive autonomy, and that the voluntary participation of women is ensured. The Thesis has revealed that the goal of enhancing reproductive autonomy has an effect on the overall process of screening. Of these implications, first, the reasons of uncertainty concerning the use of appropriate screening methods were studied, then the professionals' and the pregnant women's attitudes to screening were interpreted and a guideline was constructed to help professionals give verbal and written pre-test information. Later on, the views of genetic counsellors on the goal of genetic counselling and the genetic counselling expectations of pregnant women were explored. Based on these data, the abandonment of non-directive consultation style and the application of interpretive genetic counselling have been suggested. The new, interpretive method was elaborated in detail by the Thesis. By acknowledging the social impacts of prenatal screening, the Thesis has supported the development of those forums where the prenatal and postnatal issues of Down's syndrome are discussed jointly.

**Conclusion:** The PhD thesis can serve as an example for presenting how medical ethics theories and experiences deriving from everyday medical practice can be combined. Thus, its results can be utilised in the fields of prenatal screening, genetic counselling and academic education of medical ethics.

## 1. Introduction

Down's syndrome or trisomy 21, as it is frequently called because of its chromosomal origin identified by J. Lejeune in 1959, is the most common genetic cause of mental disability and occurs in approximately one in seven hundred births [1,2]. Besides learning disabilities, which varies from mild (IQ 50-70) to severe (IQ 35-50), individuals with Down's syndrome can have other serious disorders, e.g. heart defects, gastrointestinal problems, hearing loss, ophthalmic disorders, susceptibility to infections, early Alzheimer disease [3]. With the development of medicine, most of these diseases have become treatable and owing to early intervention programmes beginning in infancy, the physical and cognitive ability of the affected children may be improved. So, they can develop the skills of walking, talking, dressing, etc. although, with a delay compared to other children, and resulting from special education acquired in separated or inclusive classes, many affected children learn to read and write. In a supportive environment, several adults with Down's syndrome can hold regular job and can lead an assisted independent life [4,5].

The relatively high incidence of Down's syndrome and the severity of diseases associated with it have generated an increased interest of medicine towards this genetic condition, and in the last three decades, prenatal Down's syndrome screening has become one of the most rapidly evolving fields of biomedicine. Nowadays, by the use of screening and diagnostic technologies, prenatal medicine can offer women information about their individual risk of having a pregnancy with Down's syndrome, which risk may significantly differ from their age-related risk, and for women with an increased chance of having an affected foetus, the possibility of accurate diagnosis. These achievements have resulted in the introduction of nationally organised, universally provided prenatal Down's syndrome screening in a growing number of countries [6-9]. In some other states, like in Hungary, which lack a formal screening programme, the professional considerations and the parental choices have contributed to the spread of *ad hoc* screening that works outside central coordination [10-12].

### 1.1. Research objectives

In ethical terms, prenatal genetic screening for Down's syndrome constitutes a special territory of the application of modern medical technology, and these specialities derive from the characteristics that it is an antenatally performed screening procedure and it screens for a disability condition which is not life threatening but causes severe handicap. Since prenatal Down's syndrome screening touches the sensitive fields of

reproductive decisions and social attitudes towards people with disability, its history has been accompanied by constant debates. The proponents, the majority of pregnant women and obstetricians, have welcomed the new opportunities provided by prenatal screening [13], and this enthusiasm has made Down's syndrome screening to be the standard component of antenatal care and has launched the proliferation of screening methods. The opponents, feminist activists and disability rights advocates, have criticised the negative effects and have questioned the necessity of screening on the grounds that it forces women to make unwanted decisions, it has a "search and destroy" mission concerning the affected fetuses, and it includes discriminative and eugenic hints against disabled people [14-18]. The inherent dichotomy hidden in prenatal Down's syndrome screening, meaning that while it creates new perspectives for the parents to tackle the problems of Down's syndrome, it also induces new complex medical and social problems, renders the study of the ethical implications of this medical procedure essential. The ethical analysis has the tasks to answer the question whether the implementation of prenatal Down's syndrome screening is ethically justifiable at all, and if the answer is positive, to explore the moral issues that should be taken into consideration through the application of prenatal Down's syndrome screening. In Hungary, these topics have not yet been studied, although, some of them have been thoroughly discussed in the international literature. Thus, in the dissertation, I intend to present the results of my theoretical and empirical researches that have added new elements to the knowledge on the ethical issues of prenatal Down's syndrome screening. Further aims of the doctoral paper are to give practitioners an ethical guideline on prenatal Down's syndrome screening and to outline a methodological framework of reproductive genetic counselling, which follows prenatal screening in several cases. A fulfilment of these tasks necessitates studying the following topics:

- 1) **Ethical acceptability of prenatal Down's syndrome screening:** What are the aims of the application of prenatal Down's syndrome screening? Are these aims similar to the aims of other kinds of population screening programmes, or are they different? Are these aims ethically acceptable?
- 2) **Implications of an ethically justifiable aim on medical practice:** What practice should be offered to meet the ethically justifiable aim of screening? Which screening methods should be used? How much and what kind of information should be disclosed to women and couples about prenatal screening? What kind of doctor-patient interaction should be formed through genetic counselling concerning Down's syndrome? What are the counselees' expectations on genetic counselling? Can these expectations be



satisfied by applying non-directive genetic counselling, or the use of a newer method is necessary? What are the characteristics of this new method of genetic counselling?

**3) Wider social impacts of the application of prenatal Down's syndrome screening:**

What are the wider social impacts of screening? By what means can socially, culturally and morally important values be preserved through the implementation of prenatal Down's syndrome screening? What changes are required in order to adjust the Hungarian prenatal screening practice to widely agreed ethical principles?

The need to answer these questions has been strengthened by the current public interest emerging towards the introduction of universal prenatal screening in Hungary [19,20], and by the circumstance, that the importance of first trimester nuchal translucency screening, which is considered as an indispensable part of the most effective screening methods, has been discovered by physicians working at the Medical Faculty of University of Szeged, Hungary [21]. This discovery has inspired the co-operation of physicians and medical ethicists in Szeged, which is regarded unique in the Hungarian health care context, and the results of this common work are presented in the doctoral paper.

## **1.2. Research methods**

To acquire data about what is already known on the topic of the dissertation, a literature review was performed, being the basic method of every scientific research, which has covered papers dealing with theoretical medical ethics, the special ethical problems of prenatal screening and diagnosis, and the medical technology of prenatal Down's syndrome screening. In those territories where the doctoral paper has added new information to the previously existing knowledge, three types of methods were applied: the biomedical ethics approach, the interpretive ethics approach, and the empirical sociological method of a questionnaire study.

### ***1.2.1. Biomedical ethics approach***

Since biomedical ethics, or briefly bioethics, the prevailing view of recent medical ethics thinking, has been developed by the purpose of addressing and solving the ethical problems posed by technological medicine, adopting this system to the issues of prenatal Down's syndrome screening seems obvious [22-26]. A justification given by the biomedical ethics approach can ensure widespread acceptance of the use of a medical procedure, as in making moral judgements about what is right and what is wrong, bioethics combines the traditional ideas of medicine, such as beneficence and non-maleficence and the values of democratic societies, such as respect for autonomy and justice. These four

principles are also deemed as a consensus between the two main philosophical ethical theories, deontology and utilitarianism. Therefore, the moral commitments determined by the four principles can fulfil both the criteria of deontology, that a morally right action should follow the moral duties, and of utilitarianism, that a morally right action should maximise the overall welfare. A firmly justified medical decision is expected to take into consideration more than one ethical principle [27], since in biomedical ethics moral duties are perceived as *prima facie* duties that can be overridden if good reasons are found, however, their infringements leave moral traces. By relying on the ethical reasoning method provided by the four principles, in the dissertation, the traditional goal of prenatal screening (goal of prevention) has been challenged and another, ethically consistent goal (enhancing parents' autonomy) has been proposed. The bioethics approach has also been fruitfully used when studying the social impacts of prenatal screening and the issues of respect for autonomy, which principle has taken precedence over the other ones in the medical practice of Western-type societies.

### ***1.2.2. Interpretive ethics approach***

While, in the international literature, interpretive ethics is regarded as an alternative or a complementary approach relating to the dominant bioethics view [28-32], in Hungary, this approach has been hardly known yet, so, the application of interpretive ethics to prenatal Down's syndrome screening should be considered as a novelty of the dissertation.

The interpretive approach emphasises that through the medical interaction, interpretive processes take place where the physician interprets the patient's symptoms in order to set up a diagnosis and tries to explore what meaning is attributed to the disease by the patient in order to provide proper help, which is not merely medical but also psychological and emotional. By recognising the connection between medical practice and the process of interpretation, the most important doctrines of philosophical hermeneutics elaborated by Hans-Georg Gadamer [33] may be incorporated in the methods of medical science. The ideas that human understanding is always influenced by the context in which it occurs, and that in the process of understanding both the phenomenon being interpreted and the interpreter interact with each other have strongly affected medical ethics. Thus, the interpretive ethics approach contends that the moral deliberations of patients are significantly determined by their cultural and social traditions and not only by the rationally acknowledged personal values, as bioethics has previously presumed [34]. The interpretive approach places the doctor-patient relationship into a new perspective as well; it rejects the view of traditional medical ethics that considers patients as only the passive

subjects of medical interventions, and it also denies the concept of bioethics that regards physicians as merely technicians whose expertise is restricted only to technical knowledge and skills [35]. In interpretive ethics, the doctor-patient interaction is viewed as a dialogical relationship in which both parties play active roles; the physician and the patient mutually share their knowledge (professional and lay) and experience (scientific and personal) that can result in a common interpretation of the situation caused by the disease, that differs from the presumptions of both participants. When, in collaboration with colleagues, I have applied this new approach to the ethical questions of genetic counselling, a deeper insight into the dynamics of the doctor-patient relationship has been gained [36]. The novelty of this work were appreciated by a prestigious, peer-reviewed international journal that, besides the publication of the results of this research, initiated a debate on the problems of genetic counselling based on the issues addressed by our paper [37]. An elaboration of the method of interpretive genetic counselling has also been owed to the application of this new method, and our paper written on this issue has been found worthy for publication by an international journal with an impact factor [38].

### ***1.2.3. Empirical sociological approach: a questionnaire study***

In the dissertation, the biomedical and interpretive ethics approaches were applied to perform normative ethical analysis, e.g. to make considerations about what ought to be done, however, when descriptive ethical questions were studied, e.g. different moral attitudes on a given issue were explored, a sociological research method was used. Owing to a co-operation with the Department of Medical Genetics, University of Szeged, I had the opportunity to conduct descriptive ethical research on the expectations of pregnant women on genetic counselling concerning Down's syndrome. Previously, no data on this issue have been available in Hungary. Consequently, the normative moral recommendations of the dissertation are based not only on theory but also on facts coming from day-to day medical practice.

The empirical research included a quantitative questionnaire study, which was conducted among pregnant women visiting the genetic counselling clinic of the Department of Medical Genetics, University of Szeged. The questionnaire was based on the related international and Hungarian literature, was self-administered by the respondents, and was approved by the institutional ethics committee. The analysis of the empirical data required the use of a statistical programme provided by the Statistical Package for Social Sciences, version 15.0 (SPSS, Chicago, IL, USA).

## **2. Goals of prenatal Down's syndrome screening – competing principles of beneficence and respect for autonomy**

A study of the ethical issues of prenatal Down's syndrome screening should begin with answering the crucial question whether the application of this medical procedure is ethically acceptable at all. Since a response to this question is strongly affected by the goals of prenatal Down's syndrome screening, in the following, I critically examine the two competing concepts given on this topic in the international and the Hungarian medical literature. As the positions relating to the ethical acceptability of prenatal screening and to the goal of it can influence both public policy and doctor-patient relationship, examining these questions with scrutiny has of vital importance. When criticising the intuitively given aim of prenatal screening and endorsing a more sophisticated goal, I intend to help professionals to choose an ethically consistent goal for their practice.

### **2.1. Goal of disease prevention: a critique**

The first approach, motivated mainly by public health considerations, claims that the goal of prenatal screening is disease prevention; prenatal screening tests can be viewed as a form of secondary prevention, which completed with selective abortion, can reduce the number of newborn babies having Down's syndrome [39-44]. This position is influenced by the view that population screening programmes, such as mammography, colorectal or prostate cancer screening, are thought to belong to the realm of preventive medicine, which, by identifying those at increased risk, can facilitate early diagnosis and successful treatment. Furthermore, the general tendency that almost 80-90% of the parents decide against the continuation of a pregnancy affected by Down's syndrome might suggest that prenatal Down's syndrome screening serves preventive purposes [45,46]. Nonetheless, I challenge the goal of prevention by stating that this aim is intuitively given, and the lack of systematic ethical analysis reflected in the definition hinders the proponents of this view in taking into account the differences of prenatal genetic screening compared to other population screening programmes, and in recognising the wider social impacts of the goal.

#### ***2.1.1. Problems with the ethical justification of the goal of prevention***

To illuminate the differences of prenatal screening and other types of population screening, I perform an ethical justification by using the reasoning method of biomedical ethics. When the implementation of population screening programmes is justified, generally, the principle of beneficence is applied as a moral argument. Benefiting the

patient, contributing to the health-related welfare of others has been constituted a fundamental professional duty of physicians since the time of Hippocrates, and in nowadays biomedical ethics the duty of beneficence requires preventing harm (vaccination, etc.), removing harm (administering medication, surgical operations, etc.), and promoting good (biomedical researches, etc.) [23,47]. Naturally, screening programmes that are followed by early diagnosis and effective treatment can benefit the patient having the disease by preventing harm, so their implementation can be approved on the grounds of beneficence. However, in the case of Down's syndrome, no curing procedure exists and only the termination of affected pregnancies can be offered to the parents, thus, by screening, not the disease but the birth of a baby with the condition may be "prevented". Consequently, the foetus having the disease is not benefited as its abortion is expected in the sense of prevention. Moreover, it is theoretically dubious whether the foetus can be benefited or harmed at all, since, in biomedical ethics, only those can be benefited or harmed who have special moral status and until now, no consensus has been developed on the moral status of foetuses. Therefore, a decision whether or not a foetus is deemed as a moral person that can be benefited or harmed depends on the discretion of the parents, albeit among the conditions provided by the law.

As the application of the principle of beneficence has proved inadequate in the case of foetuses, now, this principle is applied to pregnant women participating in prenatal screening. The examination reveals that through Down's syndrome screening, not the health of pregnant women is promoted at the first place, but an opportunity is provided them to make a decision about the future of an affected pregnancy, and the territory of making choices belongs to the principle of autonomy and not to the principle of beneficence. The idea that, through antenatal Down's syndrome screening, the key issue is the parents' decision-making is well demonstrated by the resolutions of those parents - even though their numbers are only few - who participate in prenatal screening but refuse the abortion of an affected foetus, for which action the goal of prevention cannot give any explanation.

### ***2.1.2. Negative social implications of the goal of prevention***

While it is recognisable, that the ethical justification of the goal of prevention is problematic, since the moral rightness of this aim cannot be proved by the principle of beneficence, the goal of prevention raises further ethical problems by its possible negative implications on widely supported social values. For example, the goal of prevention fails to recognise that deciding about the future of a Down's syndrome pregnancy represents a

genuine reproductive decision and not a medical decision, where the decision-makers should be the parents and not the medical professionals. Because of the underlying presumption of the goal of prevention that the affected pregnancies will be terminated, parents may think that their reproductive liberty has suffered interference by third parties e.g. by providers of screening tests or health care decision-makers, who expect or suggest a decision which belongs exclusively to the parents' authority. It is just the notion of respect for reproductive autonomy that differentiates the expression of "*preventing the birth of a baby with Down's syndrome*" from "*avoiding the birth of a baby with Down's syndrome*" in ethical terms [48]. As, while the first expression may imply administrative, regulative or even coercive measures that constrain reproductive liberty, the last refers to allowing parents to make decisions on procreation that respects reproductive freedom.

Considering prevention as the goal of prenatal screening may violate the sensitivity and interest of families bringing up children with Down's syndrome as well [49], since it is frequently associated with the argument that the care of people with Down's syndrome imposes severe burden not only on families but also on society. The parents generally interpret this argument as discriminative and contradictory to the principle of justice, as based on social justice, people in disadvantageous situation rightly expect the support of the community in order to improve their chance for a better life.

The aim of prevention encourages the rise of eugenic thoughts too, as by emphasising the community's interests in reducing the number of newborn babies with Down's syndrome, the idea seems to be suggested that the community supports the selection of foetuses based on their genetic make-up. However, in 20th century history, eugenic thoughts served corrupt political aims and led to tragic consequences, so, refraining from eugenics constitutes an ethical imperative for recent medical genetics [50,51].

In summary, the ethical analysis has pointed out that, by contrast to other population screening programmes, the goal of prevention cannot be considered as an ethically acceptable aim of prenatal Down's syndrome screening as it cannot be justified by the principle of beneficence. Furthermore, the goal of prevention may compromise the doctor-patient relationship by giving a tacit approval to the physicians' influences on the parents' reproductive decisions and may convey negative messages on the social value of people with disability. Because of these reasons, I suggest omitting prevention out of the goals of prenatal Down's syndrome screening.

## **2.2. Goal of enhancing parents' reproductive autonomy: an approval**

The other concept defines the goal of prenatal Down's syndrome screening as enhancing parents' reproductive autonomy by disclosing information about the genetic risk of the foetus on that basis parents can make informed decision about the future of the pregnancy and about their family planning [39,43,52,53]. The ethical justification of this aim can be performed by alluding to the principle of respect for autonomy, which is a *prima facie* moral duty in biomedical ethics and requires the recognition of the right of persons to self-governance insofar as their actions do not violate the same rights of others [23]. Noteworthy, that while a rational person's right to self-governance is widely supported, a future child's rights to autonomy is ethically debated and is associated with the issue of the moral status of foetuses. Thus, promoting the reproductive preferences of parents and supporting them to control the most intimate sphere of their life exemplify respect for autonomy, and many actions belonging to this field can be morally justified.

### **2.2.1. Scope of reproductive autonomy**

In the developed countries, reproductive autonomy includes the rights to not procreate by using contraceptive methods and abortion; to procreate by using fertility treatments and assisted reproductive technologies; to decide the timing and number of children; to choose the mood of labour; and also to promote the health of a future child by using prenatal screening and diagnostic services [54-56]. Nonetheless, the scope of reproductive liberty is not without limits and the parents' reproductive preferences are not accepted without exceptions. For example, using selective abortion for sex selection based on social and not medical reasons is prohibited in the majority of countries, since this selection generally happens to the disadvantage of female foetuses and Western-type societies are not willing to support a policy which may strengthen discriminative tendencies [42,57]. However, contrary to the fact that prenatal screening services are also accused by perpetuating discrimination against people with disability, selective abortion of a disabled foetus has remained an important part of the parents' reproductive autonomy. In this social practice, according to my view, a widely shared belief about the differences of disability and discrimination is reflected. As, while gender-based discrimination is caused by social arrangements and can be solved by social measures, the learning disabilities of people with Down's syndrome are caused by objective, physiological factors and cannot be totally eliminated by anti-discriminatory social policy. (Nonetheless, its negative impacts can be exaggerated by discriminative social attitudes.) Despite feminist views asserting that a woman should have the right to terminate an unwanted pregnancy, however, if she

decides to have a child, she has to accept any kind of child [58], I contend that because of the special tasks and responsibilities families bringing up children with disability have to face, leaving decisions about the future of an affected pregnancy on the parents is morally justified.

For the reason that prenatal Down's syndrome screening aims to enhance the free reproductive choices of women, special attention should be paid to their voluntary participation as, sometimes, social or familial expectations make difficult to reject to participate in screening, or to continue with an affected pregnancy. Providers of prenatal screening should also be aware that while this procedure enhances parents' reproductive choices, its spread may involve the danger that the birth of a disabled child would be considered as a result of the deliberate choices of the parents, which may reduce social sympathy and support given to these families [59].

### ***2.2.2. Moral status of fetuses with disability***

When including the selective abortion of disabled fetuses in the reproductive rights, it is presumed that fetuses with disability possess lower moral status than persons or non-disabled fetuses [60]. The ethical literature offers moral arguments underlying this presumption; for example, the argument from "potential" represents a conservative idea, which is accepted even by some protestant churches in the case of disabled fetuses [61]. According to this view, embryos and fetuses are not human persons, however, they have the potential to develop into it. Our moral obligations towards them, meaning whether or not they can be destroyed, depend on the extent they include the potential to develop into a full human person. Since fetuses with disability possess a smaller degree of this potential than non-disabled fetuses, their termination can be ethically justified [62].

The argument of "replaceable fetuses" demonstrates a utilitarian idea, which demands that in decisions about the future of a disabled fetus, the utilitarian principle of maximising the overall welfare of all affected parties should be considered [63]. The utilitarian calculus on the amount of happiness and misery presumably produced by the continuation or the termination of an affected pregnancy is performed by comparing the quality of life of a future disabled child with that of a future non-disabled child. As the life of a future disabled child shows a lower quality than that of a future non-disabled child, according to the utilitarian principle, the parents have to choose the termination of an affected pregnancy, provided, they can conceive a non-disabled child in the future [64].

The lack of consensus on the moral status of fetuses has an influence on the issue of the moral status of disabled fetuses as well. Thus, the above-mentioned arguments are acceptable for those parents who are considering the termination of an affected pregnancy



but are unacceptable for those who, in accordance with the Catholic Church, attribute full moral status to the foetus. In the European countries, the laws on the abortion of Down's syndrome foetuses are varying greatly. Generally, although under strict control, it is allowed before 24 weeks of gestation, when the parents have received the results of mid-trimester amniocentesis, but the foetus has not yet reached the threshold of viability [6,65].

### ***2.2.3. Social implications of the goal of enhancing reproductive autonomy***

Sometimes, the goal of enhancing the parents' reproductive autonomy is criticised on the grounds that professionals of prenatal medicine have shifted the responsibility away from themselves, since by this goal, the discriminative and eugenic features of prenatal screening can be interpreted as being the unintended consequences of the parents' individual choices [18,42]. While this objection points to the undesirable consequences of discussing the issues of prenatal Down's syndrome screening only from the perspectives of the screening participants, the aim of enhancing reproductive choices should be regarded as a thoughtfully given definition that considers both the interests of the parents and the wider social impacts of screening. This aim takes seriously the parents' reproductive autonomy, as it does not include any expectation about the content of parental decisions about the future of an affected pregnancy; respects the sensitivity of families having children with disability, as it does not convey discriminatory meaning against disabled people; and refrains from eugenics, as it does not involve concerns about the condition of the human gene pool.

At the end of the ethical analysis of the goal of prenatal Down's syndrome screening, the initial question should be answered whether the implementation of this medical procedure is ethically acceptable. In answer to this question, I assert that if prenatal Down's syndrome screening aims to enhance the parents' reproductive autonomy, and if women's voluntary participation is ensured, its implementation is ethically acceptable. As the goal of enhancing parents' reproductive autonomy can be ethically approved by the principle of respect for autonomy, and as this goal takes also into account the moral requirements of the principle of justice by rejecting eugenic and discriminatory tendencies, I contend that only this goal can give an ethically consistent justification to prenatal screening. Obviously, a definition of the goal of prenatal screening can give only a frame to practice good medical care, but its realisation depends on the quality of the doctor-patient relationship and on the supportive social and health care contexts, which are indispensable in order to guarantee the really free choices of women through the whole process of prenatal Down's syndrome screening.

### 3. Confusion on the issue of what screening technology should be applied

On the account that prenatal Down's syndrome screening promises to enhance the parents' reproductive autonomy by giving reliable information on the genetic risk of the foetus, choosing the screening technology fulfilling best this task is regarded essential. The ethical problems derive from the feature of screening that it can identify women with an increased chance of having an affected pregnancy by using biochemical and ultrasound markers (Table 1), however, it can never reach the accuracy of invasive diagnosis. False positive and false negative results are inseparable parts of screening that may induce negative psychological consequences in women, and in cases of screen positive results, diagnostic procedures (amniocentesis, chorion villus sampling) may result in about 1% foetal loss. Therefore, screening technologies should meet the ethical requirement of "do no harm" concerning the health of mothers and foetuses, so they should perform low false positive rate. Furthermore, in the sense of utility, they should maximise the advantages for all who are affected, so they should present high detection rate and cost-effectiveness.

**Table 1:** Currently applied ultrasound and biochemical screening markers for Down's syndrome [66-68]

	First trimester	Second trimester
<b>Ultrasound markers</b> (based on foetal anatomy)	Increased nuchal translucency (NT) Absent nasal bone Tricuspid regurgitation Increased impedance in ductus venosus	Increased nuchal fold (NF); Absent nasal bone; Congenital heart defects; Intrauterine growth restriction; Mild cerebral ventriculomegaly; Choroid plexus cysts; Cystic hygromas; Echogenic intracardiac foci; Increased intestinal echogenicity; Duodenal atresia ("double-bubble sign"); Renal pelvis dilation; Shortened humerus and femur; Increased iliac wing angle; Clinodactyly and hypoplasia of the fifth finger; Increased space between first and second toes; Two-vessel umbilical cord
<b>Biochemical markers</b> (based on maternal blood serum components)	Pregnancy associated plasma protein A (PAPP-A) Free $\beta$ human chorionic gonadotrophin (free $\beta$ -hCG)	Alfa foetoprotein (AFP) Total or free $\beta$ -hCG Unconjugated oestriol ( $uE_3$ ) Inhibin A

#### 3.1. Debated questions of screening technology

When reviewing the relevant literature, confusion and uncertainty have been revealed on the questions of the most effective and safest screening methodology. Although, the literature has generally mentioned the great number of methods as the source of uncertainty [69], the ethical consequences of this uncertainty have not been systematically studied. Therefore, I think necessary to give an overview about the manifold reasons of the confusion emerging on the use of the appropriate screening method and to summarise the ethical requirements that screening strategies have to fulfil in order to dissipate this confusion.

### 3.1.1. Too many screening methods

The difficulty of making decisions about the use of the best technology is partly owned to the great variety of the applicable screening methods. In the last three decades, the technological development concerning antenatal Down's syndrome screening has been so enormous that currently 17-19 different screening strategies are available [70,71], which diversity of technical opportunities is without precedent in the other areas of medical care. The proliferation of methods has been fostered by the shift happened in the philosophy of screening as, while, in the 1980s and 1990s, single-type methods were used (either biochemistry or ultrasonography, and either first or second trimester tests), in the last decade complex screening methods have been introduced that apply biochemistry and ultrasonography together, or first and second trimester tests together. Thus, besides the well-known screening technologies, such as second trimester double, triple and quadruple biochemical tests and first trimester nuchal translucency scan [72-75], newer, complex screening technologies have slipped into practice (Table 2), such as the different versions of combined, integrated, stepwise sequential, and contingent screening [76-81], the advantages and disadvantages of which are not always evident for the practitioners.

**Table 2:** Down's syndrome screening tests and detection rates (5% screen positive rate) [82]

First trimester		Second trimester		First and second trimester	
Screening test	Detection rate	Screening test	Detection rate	Screening test	Detection rate
<b>NT measurement</b>	64–70%*	<b>Triple screen</b> (MSAFP, hCG, unconjugated oestriol)	69%*	<b>Integrated</b> (NT, PAPP-A, quad screen)	94–96%*
<b>Combined screen</b> (NT measurement, PAPP-A, free or total $\beta$ -hCG)	82–87%*	<b>Quadruple screen</b> (MSAFP, hCG, unconjugated oestriol, inhibin A)	81%*	<b>Serum integrated</b> (PAPP-A, quad screen)	85–88%*
				<b>Stepwise sequential</b> First-trimester test result after <i>combined screen</i> : - Positive: diagnostic test offered - Negative: second-trimester test ( <i>quad screen</i> ) offered Final: risk assessment incorporates first and second results	95%*
				<b>Contingent sequential</b> First-trimester test result after <i>combined screen</i> : - Positive: diagnostic test offered - Negative: no further testing - Intermediate: second-trimester test ( <i>quad screen</i> ) offered Final: risk assessment incorporates first and second results	88–94%‡

Abbreviations: hCG = human chorionic gonadotropin; MSAFP = maternal serum alpha-foetoprotein; NT = nuchal translucency; PAPP-A = pregnancy-associated plasma protein A; quad = quadruple.

\*From the FASTER study [see reference 79]

‡Modelled predicted detection rates [see reference 70]

### ***3.1.2. Reliability of data on screening performances of tests***

Theoretically, the good screening performances of the complex methods, i.e. they have a high sensitivity (over 85%) and a low false positive rate (under 5%), can ease the task of choosing among them, as by their use, the number of detected Down's syndrome pregnancies can be increased and the number of invasive diagnosis can be decreased. However, in the literature, doubts have emerged about the reliability of data produced by statistical models and observation studies. A great part of proposals on future screening policies have been based on statistical modelling studies using the data of well-designed trials, but critics have assumed that these tests would not perform so well in real world situation [83]. In practice, in cases of strategies containing two-step tests, the pregnant women's participation in both parts of tests could not be totally ensured, however, it was presumed by modelling studies. Concerning observation studies, it has become clear that where nuchal translucency (NT) was measured only for the sake of scientific interest, the proportion of failed or unsatisfactory NT measurement was much larger than in intervention studies where NT was measured for the sake of risk calculation [75,84]. The confusion further increased when scientists conducting large-scale studies to compare the effectiveness of diverse screening methods made contradictory recommendations based on research data. For example, the leader of the British SURUSS study categorically opposed the introduction of contingent screening [85], while a representative of the North-American FASTER study definitely proposed it [45]. An additional ethical problem has been raised by conflicts of interests, e.g. when a screening technology has been patented (as it is the case with integrated test), since in this situation, personal prejudices may take part in proposals about the application of a particular screening technology.

### ***3.1.3. Issues of using biochemical or ultrasound screening, or both***

The new technologies have caused confusion not only by their surprisingly great number but also by their complex methods that have compelled professionals to change their previous ideas on screening. Scientific data, showing that combining biochemical screening with ultrasonography can give a better screening performance than either of the two tests alone [67], have challenged the views of those who have favoured the use of biochemical screening for the reason that serum tests are not so sensitive to professional skills than ultrasound scan [78]. As, except from serum integrated screening, all of the complex screening tests include nuchal translucency measurement, ensuring well-trained professionals who have acquired the skills necessary to perform this procedure is generally expected from recent prenatal screening services. While, previously, the concerns about the reliability of NT measurement have hindered the spread of nuchal translucency screening, nowadays, in the new screening programmes, these problems have been acknowledged and

initiations have been made to solve it by setting up standards for professional skills and ultrasound equipments, by organising specific training, and by establishing ongoing quality assessment of ultrasonographers and centres [82]. Noteworthy, that the Fetal Medicine Foundation, a registered charity organisation in the United Kingdom, has supported the appropriate introduction of NT screening into clinical practice for almost a decade, and has established a process of training and quality assurance, available through the Internet [86].

### ***3.1.4. Issues of using first trimester or second trimester screening, or both***

The other professional attitude that has needed revision in light of new screening methods is the insistence on providing mainly second trimester tests. Second trimester triple and quadruple tests have been widely applied in the developed countries since the 1990s. Recently, serum and fully integrated screenings, which integrate the results of first and second trimester tests into a single risk calculation [78], also support the idea of giving risk assessment only in the second trimester, by stating that fewer women have to decide about the future of a pregnancy because of the intrauterine loss of Down's syndrome foetuses. However, in the existence of similarly effective first trimester screening methods, it is difficult to ethically justify the general use of methods giving risk assessment only in the second trimester, since they impede women in getting information earlier and in accessing to early diagnosis and early pregnancy termination. Empirical studies also suggest that pregnant women prefer first trimester screening, as, in a survey, about 70% of women stated that they would still choose first trimester screening even if all the identified Down's syndrome pregnancies miscarried before the second trimester [87,88]. Moreover, telling nothing about the findings through ultrasound scan, and remaining silent when the probability of foetal abnormality is great, which are the expectations of integrated tests concerning the first trimester part of the tests, are unacceptable for many physicians. Some of these problems are solved by stepwise sequential and contingent screenings, which offer first trimester prenatal diagnosis for women in the high-risk group, but continue screening with all the remaining women or with women in the intermediate risk group.

### ***3.1.5. Issues of offering prenatal diagnosis on maternal age***

With the development of screening technologies, the importance of maternal age in antenatal Down's syndrome screening has also been questioned, although, the association between maternal age and the risk of having a pregnancy with trisomy 21 (Table 3) has prompted a policy of offering prenatal diagnosis to pregnant women aged 35-37 years and more in the great majority of countries. In the 1970s, when this method was first applied, about 5% of pregnant women belonged to this age group who carried about 25-30% of Down's syndrome pregnancies. By the application of screening methods providing

individual risk assessment, the significance of maternal age as a screening method has seemed to be decreasing. The risk calculated for an individual person gives more reliable information than a risk estimated for an age group, as, in the first case, the maternal age-related and gestation-related risk is modified with the results of the performed screening tests. However, until now, only the American College of Obstetricians and Gynecologists recommended in 2007 to cease proposing automatically prenatal diagnosis to pregnant woman aged 35 years or more and to offer invasive diagnosis only on the results of tests providing individual risk calculation or at the request of the pregnant woman [82]. Nonetheless, the latest changes in the age distribution of pregnant women has drawn attention to the need of cautious decision-making on the role of maternal age in prenatal Down's syndrome screening. In the well-developed countries, the proportion of pregnant women aged 35 years and more is around 15-20%, so, the sensitivity of screening based on maternal age reaches 50-60% in those countries [10,89].

**Table 3:** Estimated risk for trisomy 21 in relation to maternal age and gestation [75]

Maternal age (yrs)	Gestation (wks)				Maternal age (yrs)	Gestation (wks)			
	12	16	20	40		12	16	20	40
20	1068	1200	1295	1527	35	249	280	302	356
25	946	1062	1147	1352	36	196	220	238	280
30	626	703	759	895	37	152	171	185	218
31	543	610	658	776	38	117	131	142	167
32	461	518	559	659	39	89	100	108	128
33	383	430	464	547	40	68	76	82	97
34	312	280	378	446	41	51	57	62	73

(Rate of foetal loss is about 30% between 12 weeks of gestation and term, and about 20% between 16 weeks and term)

### 3.2. Need for professional guidelines on screening technology

In answering the question what screening method should be used, the professional bodies of prenatal screening experts, obstetricians, and geneticists have fundamental responsibilities, since their statements can function as guidelines for national screening programmes, for private health care services, and even for pregnant women. The construction of professional guidelines can be supported by a summary of requirements that screening programmes have to meet according to the findings of the above analysis:

- *Based on reliable data:* Screening strategies should be built on the results of large-scale prospective and intervention studies to avoid biases connected to research methodology.
- *Presenting good screening performance:* Of the available methods, those should be used that perform a high detection rate (over 85%) and a low false positive rate (5% or less). With the decrease of false positive and false negative test results, maternal anxiety and the number of invasive diagnostic procedures can be reduced.
- *Combining biochemistry with ultrasonography:* According to data, this combined technology is one of the most effective ways of reaching good screening performance. Including first trimester combined screening (NT, PAPP-A, free  $\beta$ -hCG) in the

screening strategy should be considered. However, since obtaining nuchal translucency measurement necessitates specialised training, and nuchal translucency might not be measured successfully in an individual patient, besides combined screening, other methods should also be made available.

- *Offering first trimester screening:* This option can shorten the period of “tentative pregnancy” and maternal anxiety, can ensure early diagnosis, and can fulfil the preferences of the majority of pregnant women. Of the complex methods, not only the combined test but also some contingent methods make possible to complete screening in the first trimester even for women in the intermediate risk group [81].
- *Offering also second trimester screening:* To maintain this type of screening is necessary in the interests of pregnant women seeking prenatal care only in the second trimester. Quadruple test (AFP, hCG, oestriol, inhibin-A) is unanimously accepted as the best second trimester test for Down’s syndrome.
- *Requiring the audit of screening professionals and centres:* Where screening programmes include nuchal translucency measurement, specific training to acquire a standardised method of measurement and ongoing quality control are recommended.
- *Taking a stance on the role of maternal age as a screening method:* In this process, besides the age distribution of pregnant women, the wide public awareness about the increased risk of women aged 35 years or more should be taken into consideration.
- *Addressing counselling issues:* Counselling issues are especially important in cases of those complex methods where risk is calculated only after the full completion of tests.
- *Considering cost-effectiveness:* Some authors include the cost of care of people with Down’s syndrome in the cost of prenatal screening [45], however the acceptability of this policy is debated as prevention cannot function as an ethically justifiable aim.

In summary, because of the ethical requirements of enhancing reproductive autonomy and considering foetal safety, those methods should be offered that give individual risk assessment, present high sensitivity (85% or more) and low false positive rate (5% or less), and their results are confirmed by large observation studies. Since not only one but some methods can meet the requirements, through screening, local conditions (number of well-trained sonographers and accredited laboratories, financial limitations), and specific needs deriving from gestational age, number of fetuses, previous obstetric history, or wishes to finish the test in the first trimester can be taken into account [90]. To dissipate uncertainty on the use of screening methods, I propose that professional bodies should clearly define what technology can fulfil best the given medical, ethical or economic requirements and that screening professionals should openly disclose pregnant women the reasons on which the use of a particular method is based.

## **4. Role of comprehensive and personalised information in enhancing parents' reproductive autonomy**

When screening professionals disclose the considerations underlying the use of a particular screening method, they support the idea of giving comprehensive information on prenatal Down's syndrome screening that is required by the aim of promoting parents' reproductive autonomy. Detailed information can help women and couples in performing the following tasks: (1) deciding about the participation in screening, (2) understanding the test results and their consequences on the baby's health, and (3) deciding about the course of action to follow after screen positive results. In the accomplishment of these activities, pre-test and post-test information can provide effective support for the majority of parents, but in cases of increased risk, genetic counselling is also required. In this chapter, the factors influencing the provision of pre- and post-test information are examined.

### **4.1. Pre-test information**

Informing the candidates of genetic screening programmes is considered so fundamental that the EuroGentest, a network of excellence funded by the European Union, recommends not implementing a screening programme if comprehensive and well-planned pre-test information and counselling are not guaranteed [91]. The special problems of prenatal Down's syndrome screening, namely, that screen positive results may increase maternal anxiety, may result in risky diagnostic procedures performed on healthy foetuses, and may force women to make reproductive decisions about the continuation or the termination of their pregnancies, justify the demand of providing women detailed information prior to screening. If screening is offered as a routine part of care, or is performed without the consent of pregnant women - both types of conduct neglect the provision of comprehensive information - women and couples do not have the opportunity to prepare for the consequences of screening.

#### ***4.1.1. Factors influencing information giving: attitudes of pregnant women and medical professionals to screening***

The interpretive ethics approach illuminates that the effectiveness of information giving, meaning, the information is perceived by pregnant women and then used up in their decision-making, requires not only providing detailed information but also adjusting information to the individual needs of women [31]. The interpretive approach also highlights that the presumptions and experiences of health care professionals influence remarkably the patient-professional interaction [35]. Therefore, exploring the attitudes of pregnant women and medical professionals to prenatal Down's syndrome screening seems indispensable for ensuring the success of the information process. After reviewing literature data by the interpretive method, a brief overview of the most frequently mentioned contextual factors has been constructed.



**Table 4: Women's attitudes** influencing information perception on prenatal Down's syndrome screening

Attitudes of women	Characteristics of attitudes	Communication measures to overcome the negative effects of attitudes
<b>Compliant behaviour</b>	<ul style="list-style-type: none"> <li>Behaviour, on that basis, every medical procedure offered by a professional is accepted in order to ensure the baby's health. However, this behaviour does not promote thoughtful decision-making about the acceptance of screening [40]</li> </ul>	<ul style="list-style-type: none"> <li>Those aspects of screening should be emphasised that differentiate screening from routine medical procedures.</li> <li>Attention should be drawn to both the advantages and disadvantages of screening.</li> </ul>
<b>Beliefs about the aim of screening</b>	<ul style="list-style-type: none"> <li>Women believe that the aim of screening is detecting foetal anomalies in general, but it is giving reassurance in their own cases [92].</li> </ul>	<ul style="list-style-type: none"> <li>Emphasising the aim of screening before the beginning of examinations and giving data about the recall rate may help women prepare for both screen negative and screen positive results.</li> </ul>
<b>Pursuing non-medical aims</b>	<ul style="list-style-type: none"> <li>Some women assess ultrasound screening as a "nice baby watching" and not as a tool to search for abnormalities [93].</li> </ul>	<ul style="list-style-type: none"> <li>Before ultrasound examination, the alternative outcomes of reassurance and of detecting anomalies should be disclosed.</li> </ul>
<b>Emotional reactions to screening</b>	<ul style="list-style-type: none"> <li>Even the offer of screening can cause maternal anxiety [94,95].</li> <li>Ultrasound screening can strengthen emotional bond between mothers and foetuses, which turns decision-making about the future of an affected pregnancy troublesome [96].</li> <li>Screen positive results cause maternal anxiety [97].</li> </ul>	<ul style="list-style-type: none"> <li>Giving all the relevant information prior to screening is a general expectation as in this period, women are not so anxious and can think over the consequences of screening more carefully than after the disclosure of screen positive results.</li> </ul>
<b>Lack of knowledge on Down's syndrome</b>	<ul style="list-style-type: none"> <li>Women underestimate the prevalence of Down's syndrome, and know little about the impacts of the disorder [98].</li> </ul>	<ul style="list-style-type: none"> <li>Information should be provided not only about the medical aspects but also about, the educational and social impacts of Down's syndrome.</li> </ul>
<b>Difficulty in understanding the concept of risk</b>	<ul style="list-style-type: none"> <li>Women often find difficult to distinguish screening from diagnosis [99] and to understand the concept of risk calculation [100,101].</li> </ul>	<ul style="list-style-type: none"> <li>Prenatal screening can be compared to other, better known screenings.</li> <li>Visual interpretation can be useful for interpreting the meaning of risk.</li> </ul>

**Table 5: Professionals' attitudes** influencing information giving on prenatal Down's syndrome screening

Attitudes of medical professionals	Characteristics of attitudes	Measures to overcome the negative effects of attitudes
<b>Biased attitude to screening</b>	<ul style="list-style-type: none"> <li>Some physicians show biased attitude to screening because of their opposition to abortion [39].</li> </ul>	<ul style="list-style-type: none"> <li>Professionals should acknowledge that based on their personal values, they can refuse to perform abortion but cannot refuse to give women information about the availability of screening.</li> </ul>
<b>Offering screening as routine</b>	<ul style="list-style-type: none"> <li>Screening is offered as routine, no pre-test counselling is provided. This practice is generally justified by lack of time [94,99]</li> </ul>	<ul style="list-style-type: none"> <li>Information constitutes an essential part of screening. Time should be guaranteed for giving information.</li> </ul>
<b>Uncertainty about the appropriate screening method</b>	<ul style="list-style-type: none"> <li>The multitudinous, currently available screening strategies have resulted in confusion among practitioners [82].</li> </ul>	<ul style="list-style-type: none"> <li>Guidelines constructed by professional bodies can help practitioners choose the appropriate method.</li> </ul>
<b>Favouring a particular screening technology or strategy</b>	<ul style="list-style-type: none"> <li>Screening is offered only for women aged 35 years or more [102,103].</li> <li>Among practitioners, sometimes, an unjustified insistence on either ultrasonography or biochemistry can be recognised.</li> </ul>	<ul style="list-style-type: none"> <li>National screening programmes and private practitioners are expected to revise their screening policy regularly in order to avoid the use of outdated screening methods.</li> <li>Professional guidelines can help this process.</li> <li>The reasons underlying the use of a screening method (medical, economic, others) should be disclosed.</li> </ul>
<b>Unbalanced information on lives of people with Down's syndrome</b>	<ul style="list-style-type: none"> <li>Counselling may be unduly negative about the likely quality of life for people with Down's syndrome [104,105].</li> </ul>	<ul style="list-style-type: none"> <li>Providing women up-to-date information about the life of people with Down's syndrome is deemed important in facilitating informed choice.</li> <li>A co-operation with parent support groups can help professionals to get reliable information.</li> </ul>
<b>Communication difficulties</b>	<ul style="list-style-type: none"> <li>Communication difficulties arising from a largely different cultural background of the patient and the professional may compromise the effectiveness of information giving [106].</li> </ul>	<ul style="list-style-type: none"> <li>Not only the technical skills but also the communication skills should be taught to providers of screening tests.</li> <li>Communication training should be involved in their continuous education.</li> </ul>

#### **4.1.2. Contents of pre-test information**

The acknowledgement of the socially, culturally, and emotionally determined attitudes can assist a carefully planned information process, and can successfully serve the basic idea of interpretive ethics that the content of pre-test information should be directed by the needs of pregnant women and not by the expectations of the professionals [107]. In the following, a guideline is presented that can promote public and private screening services in giving adequate written and verbal information on the following issues [108]:

*Addressing the aim of prenatal Down's syndrome screening:* In accordance with ethics, the aim of screening should be considered as informing the parents about the chance of having a Down's syndrome pregnancy. The parents' attention should be drawn to the scenario that screening might be followed by decision about the future of the pregnancy; therefore, it is advisable to make thoughtful decision about the participation in screening.

*Ensuring respect for parents' autonomy:* Pregnant women and their partners should be informed that prenatal testing is voluntary; before screening, pregnant women's informed consent should be obtained. The whole screening process or any steps of it can be refused. Women and couples must not be subjected to pressure to undertake the test, to terminate a pregnancy, or to make decisions in undue haste [109,110].

*Description of the screened disorder:* Comprehensive and balanced information about the medical, educational and social impacts of Down's syndrome should be given, which includes data about community-based services, early intervention programmes and opportunities for integrated life [111]. In these questions, the experiences of parent support groups can be taken into account. Outdated information, negative stereotypes, or offensive terminology concerning life with Down's syndrome should be excluded.

*Practical aspects of screening:* Information involves the date of screening, e.g. in what week of pregnancy the test is performed, and screening includes one-step or two-step tests. The method of screening should also be disclosed, e.g. what kind of biochemical and/or ultrasound markers are examined, and what their values are in unaffected and in affected pregnancies. A brief explanation must be given on the procedure of individual risk calculation, and women should be informed about when and how screening results are disclosed. [98,109,112].

*Epidemiological data of screening:* Using understandable epidemiological terminology and giving comprehensible information on screening performances of tests are difficult, however, are important. These data can influence women's decisions about the uptake of screening and can prevent women from overestimating or underestimating the potentials of screening. Epidemiological information should embrace the frequency of Down's syndrome both in pregnancies and in live births, the differences of screening and diagnosis, the arbitrary nature of cut-off risk, the meaning of screen positive and screen negative results, and the possibility and reasons of false positive and false negative

screening results. On screening performances of tests - including detection rate, recall rate and false negative rate - better to give centre-specific data, if they are available.

*Consequences of screening:* Deciding about the acceptance of diagnostic tests offered after screen positive results is one of the most significant consequences of screening. Thus, women should get information about the diagnostic methods and their risks, and about the length of time, they should wait to get the result of the diagnosis. Women should also be informed about the availability of genetic counselling where they can get support in the interpretation of screen positive results and in decision-making about prenatal diagnosis or the future of the pregnancy. It should be disclosed that respecting the parents' decisions about the continuation or the termination of an affected pregnancy is ethically required. It is also advisable to mention the psychological impacts of screening, since even the offer of screening can increase maternal anxiety, and screen positive results affect negatively the psychological well-being of women, regardless how the information is presented. Addressing the name of a professional who can give advice in case of anxiety is useful for both screening programmes and private services [107,113-115].

*Evaluation of screening:* Summarising the advantages and disadvantages of a particular screening test, comparing its screening performance to that of other tests, and revealing the reasons why this particular test is offered can fulfil the expectation of giving balanced information and can facilitate parents' careful decision-making.

*Resources to get further information:* Offering information leaflets, newspaper articles, and reliable Internet addresses can help women to get further information [39].

In literature, the combination of information leaflets and personal communication is endorsed as the best form of pre-test information [116-118]. A readable, regularly updated information leaflet can be taken home where enough time is ensured to think over the impacts of screening. Personal communication can tailor information to individual needs.

## **4.2. Post-test information**

Post-test information differs from genetic counselling, since the first is provided by a screening professional to every woman participating in prenatal screening, while the second is provided by a specially trained genetic counsellor to only a group of women who are assessed as having a high-risk pregnancy.

### **4.2.1. Risk presentation**

Disclosing risk constitutes one of the most important parts of post-test information, and data suggest that screening results can be more effectively conveyed through personal communication than through phone or letter [98]. In connection to risk communication, the aspects of interpretation should be taken into account, as, for example, the parents of children with Down's syndrome have proposed the use of the word "*chance*" instead of "*risk*", as in their views, the meaning of chance is neutral, however, the meaning of risk is

negative [119]. Another debate addresses the dilemma whether words, numbers, or both should be applied when probability information is disclosed to patients. Concerning the results of prenatal Down's syndrome screening, the most preferred expressions are: screen positive - screen negative, increased risk - decreased risk, or requiring further tests - not requiring further test. When screening results are expressed in numbers, fractions (1/250, 1:250) or percentages (0.4%) are used. Fractions or percentages should be consistently applied as it can be misleading if the screening result is given in the form of fraction and the risk of amniocentesis-related miscarriage is given in the form of percentage [120]. Although many physicians believe that words are easier to process, many others think that numbers may increase awareness of residual risks and may encourage deliberative decision-making [121]. According to an additional professional opinion, numbers alone are not enough; their meaning should be interpreted by words [122]. It is also admitted that information about the arbitrary nature of cut-off level of risk promotes thoughtful decision-making, therefore, parent can be given the example that the risk 1:300 counts high in one country but not high in another [123]. The idea of giving balanced information can be served well by framing risk in both terms of loss and gain, e.g. "You have 10% chance of having an affected baby, or you have 90% chance of having an unaffected baby." [124].

#### **4.2.2. Risk perception**

The aspects of interpretation appear in risk perception as well, which is determined by more complex factors than only risk presentation. For example, reasoning methods have an effect on risk perception; the same risk, if it is expressed variously as a percentage or a fraction, is understood differently according to whether the patient uses numerical reasoning, e.g. numbers serve as anchoring points, or person reasoning, e.g. numbers function as representatives of persons [125]. Assessing the comprehension of women more frequently may facilitate more effective risk communication. Besides the probability of outcomes, risk perception is also influenced by the value the parents attach to the outcomes and by the opinion the parents hold about the seriousness of the disease. Literature has drawn attention that perceived risk, rather than the communicated risk, predicts pregnant women's medical decisions [121]. Consequently, discussion about risk should be a two-way exchange of information. Simply providing risk information without discussing the pregnant women's circumstances and perceptions does not help make informed choices.

In summary, giving information constitutes an essential part of screening for which time and financial resources should be guaranteed. Because of the essential role that comprehensive information plays in decision-making, an information leaflet should be given to all women prior to screening and communication training should be included in the post-graduate education of screening professionals, where the data of tests and the ways of giving information on risk are taught. Where prenatal screening lacks central coordination, relevant professional organisations can help physicians acquire these skills.

## **5. Genetic counselling relationship of counsellors and women with an increased chance of having an affected pregnancy**

When prenatal screening results refer to an increased chance of having a Down's syndrome pregnancy, or when a pregnant woman is aged 35-37 years or more, or when she had an affected pregnancy previously or has a positive diagnostic result now, genetic counselling is offered to help her make informed decision. Over the last decades, the notion of non-directiveness has governed the ideal genetic counselling relationship of counsellors and clients. In the sense of non-directiveness, genetic counsellors are expected to give "objective", value-neutral information on the genetic risk, the genetic disorder, and the available screening, diagnostic and treatment possibilities [126-128], and they are also expected to refrain from offering advice in value-laden problems, e.g. pregnancy termination, acceptance of diagnostic procedures carrying the risk of pregnancy loss [129]. However, recently, studies have highlighted that it is impossible to provide neutral information, since genetic facts may not be insulated from social, professional, and personal values [130-132]. Additionally, experience with genetic counselling has revealed that an impartial communication style often hinders counselees in gaining adequate information and in perceiving the problem [133-135].

As these new ideas have illuminated that the individual value orientations of counsellors exert an indispensable influence over the course of counselling, and that without the fulfilment of counselees' expectations, the counselling process may prove to be useless, I thought demanding to examine the everyday practice of genetic counselling in Hungary. Thus, by an ethical analysis of the relevant literature, the views of counsellors on the goal of genetic consultation were studied, and by an empirical research, the genetic counselling expectations of counselees were explored [36]. The results of these studies were expected to answer the questions that besides non-directive genetic counselling, what kind of methods were practiced by counsellors, and what kind of counselling style could fulfil the expectations of counselees.

### **5.1. Views of counsellors on the goal of reproductive genetic counselling**

Although, in Hungary, the professional body of clinical geneticists has given an official statement on the goal of genetic counselling [136], other definitions have also survived that have been constructed since the 1970s, when a wider spread of these services began.

#### **5.1.1. Preventing diseases—an information guidance approach**

The approach that dominated the 1970s and 1980s and has remained recognisable even in today's counselling practice was introduced by Endre Czeizel, who defined the

goal of genetic counselling as to prevent severe diseases and to promote the birth of healthy children in the interests of families and society [137]. To achieve this aim, the so-called information guidance approach was applied; counsellors made unsolicited recommendations to parents on reproductive decisions, e.g. to undertake a pregnancy or to refrain from it because of the high genetic risk to the foetus [138]. The application of the nondirective counselling method was rejected, since counsellors believed that it did not fit the Hungarian medical context. They thought that clients needed direct advice on what to do. Although accepting the offered solution was not mandatory [139], the information guidance approach was criticised on international forums for its features of violating the reproductive autonomy of the parents. Owing to the criticism, the overt support of this approach became inconvenient by the second half of the 1980s, nevertheless, sometimes reports have been heard even today about counsellors giving selected information and recommending the prevention of birth defects without the clients' request for advice.

#### ***5.1.2. Preventing diseases and respecting reproductive autonomy—a transitory approach***

The next approach, invented in the early 1990s and having been favoured from that time, supports the adoption of the non-directive method. Zoltán Papp, the main advocate of this view, has emphasised that in a non-directive counselling relationship, the counsellor's tasks include providing information and advice, but do not involve decision-making [140]. By its moral standpoint, that the right to decide about the future of a pregnancy belongs solely to the parents, this view has contributed to the abandonment of recommending parents a particular course of action. The commitment to the respect for the parents' reproductive liberty, however, has not been reflected in the definition concerning the goal of genetic counselling, since Z. Papp has assumed that the aim of counselling is to ensure the birth of a healthy child for families at high genetic risk [141]. Nonetheless, this transitory view of combining the goal of respecting the autonomy of counsellees with the goal of disease prevention involves an essential contradiction. While the promotion of the autonomous choice of counsellees requires the provision of balanced information, the goal of prevention constrains parents' free choice as the information disclosed is permeated with the counsellor's personal view about the importance of preventing diseases. The focus on prevention limits the counselling interaction as well, since it does not encourage the recognition of the parents' needs differing from disease prevention, such as understanding probability information, or finding coping strategies for the situation of being at risk.

#### ***5.1.3. Enhancing the autonomous choice of parents—an informative approach***

From the beginning of the 1990s, another type of genetic counselling practice has also been prevalent in the country, the goal and method of which unambiguously

correspond to the ethical norms of non-directiveness. The proponents of this type of approach, e.g. Olga Török, Csaba Papp, and Ernő Tóth-Pál, have considered reproductive genetic counselling as being fundamentally a communication process involving informing and educating parents to make family planning decisions [53,142]. They have omitted the prevention of diseases from the goal of genetic consultation, and have restored the original meaning of non-directiveness by demanding from the counsellors not only to allow parents to make choices, but also to ensure the impartiality of information on which parents base their decision. This approach has also tried to restrict the possibility of imposing the values of the counsellors on the clients; thus, counsellors have been expected to abstain from proposing a solution, but, on exceptional occasions, they have been allowed to inform clients about what the majority of parents decided in the same situation [143].

#### ***5.1.4. Enhancing the careful deliberation of parents—an interpretive approach***

In recent years, a concept has been espoused by me and a colleague [144] that the goal of genetic counselling should be shifted from giving impartial information to the promotion of the careful deliberation of counsellees. This aim takes into account the philosophical consensus on the impossibility of value neutrality; and in order to help counsellees reach well-informed and well-considered decisions, it demands from the counsellor not only to give counsellees detailed medical information but also to explore their values with considerable effects on their decision [145,146]. To accomplish this duty, the so-called interpretive counselling method has been proposed, which provides a useful aid in identifying the value orientations of the counsellor, the counsellee, and the community that are relevant in the counselling interaction [38]. Through open dialogue, in which the counsellor does not dominate, the counsellor and the counsellee can discuss facts and value-laden problems as well, and finally, they can find a solution that is acceptable to the counsellee. Similar to the non-directive view, the interpretive counselling approach emphasises that forcing a particular decision on the counsellee is impermissible; thus, the non-prescriptive communication style remains the basic feature of genetic consultation.

## **5.2. Genetic counselling expectations of counsellees**

After a review of the genetic counsellors' views, the counsellees' expectations were studied and to gain data on this issue, pregnant women attending the genetic counselling clinic in Szeged, Hungary between September and December 2006 were asked to answer a questionnaire containing 21 items. The study was approved by the institutional ethics committee. Of the 181 eligible participants 170 responded (response rate 94%), and prior to counselling, the following data were collected (Table 6):

**Table 6:** Sociodemographic and health-related characteristics of the respondents (n = 170)\*

	n (%)		n (%)
Age		Previous pregnancies	
– 19-24 years	9 (5)	– 0	50 (29)
– 25-29 years	24 (14)	– 1	57 (34)
– 30-34 years	46 (27)	– 2 or more	63 (37)
– 35-39 years	76 (45)	Previous counselling experience	
– 40 years and above	15 (9)	– Yes	47 (28)
Residence		– No	123 (72)
– Village	43 (25)	Reasons for counselling	
– Small town	77 (46)	– Maternal age $\geq 35$ years	89 (53)
– City	49 (29)	– Positive screening test	23 (14)
Education		– Previous genetic anomaly	16 (9)
– Primary school	24 (14)	– Drug or environmental exposure	41 (24)
– Secondary school	89 (53)	Participants of counselling	
– College/university	55 (33)	– Pregnant woman alone	78 (46)
		– Pregnant woman with partner	75 (44)
		– Pregnant woman with family member	17 (10)

\* Numbers do not all sum to 170 because not all respondents answered all the questions.

Pre-counselling, the counselees' expectations on medical information, psychological support, types of decision-making, and possible decisional aids of knowing the decisions of other parents and the opinion of the counsellor were also inquired. Post counselling, the respondents were asked to evaluate the effect of actual genetic counselling from the viewpoints of comprehensiveness of information, the calming effect of psychological support, the types of their actual decision, and their satisfaction with the overall process of counselling. Statistical analysis was performed by the Statistical Package for Social Sciences, version 15.0 (SPSS, Chicago, IL, USA). The Chi-squared test was used to search the correlations of expectations, the fulfilment of expectations, the actual decision-making, and the satisfaction with the overall process of counselling with the characteristics of the respondents and with one and other.  $P \leq 0.05$  was considered statistically significant.

### 5.2.1. Pre-counselling expectations of counselees

Prior to counselling, the majority of women wished to receive detailed and new information, to get psychological support, and to know the counsellor's opinion (Table 7).

**Table 7:** Counselees' expectations on genetic counselling

Expectations	Counselees n=170	
	n	(%)
Detailed information	167	(98)
New information	140	(82)
Psychological support	115	(68)
To know the decisions of other parents	77	(45)
To know the counsellor's opinion	119	(70)
Shared decision-making	106	(62)
Independent decision-making	56	(30)
Counsellor's decision-making	10	(6)

Wishing to know the counsellor's opinion presented a direct connection with the reason for counselling ( $p = 0.051$ ), with education ( $p = 0.032$ ) and with the number of previous pregnancies ( $p = 0.019$ ), since women who had screen positive results, or who were



pregnant with their first baby, or who had a college or university degree expected more frequently the counsellor's advice than the others. Answers concerning the expected type of decision-making showed that 62% of the counselees intended to rely on the counsellor's support and wanted to reach a shared decision, 6% expected the counsellor to make the decision on their behalf, and 30% insisted on an independent decision. Of the sociodemographic variables, only education presented a statistically significant relationship with the expected type of decision-making ( $p = 0.026$ ). Pregnant women with college or university degrees expected a shared decision, while respondents with a lower level of education expected an independent decision more frequently than the others.

### 5.2.2. *Impacts of expectations and their fulfilment on actual decision-making*

According to the reports of the counselees, 45% of the consultations resulted in shared decisions, 35% in independent decisions, and no decision was reached in 20% of the cases (Table 8). In the study, women did not report the counsellor's decision-making.

**Table 8:** Evaluation of actual decision-making

	Total n=170	Shared decision was made n=76	Independent decision was made n=60	No decision was made n=34	p value
	n (%)	n (%)	n (%)	n (%)	
Education					0.072
– Primary school	24* (14)	12* (16)	10* (17)	2 (6)	
– Secondary school	89 (53)	38 (51)	36 (61)	15 (44)	
– College/university	55 (33)	25 (33)	13 (22)	17 (50)	
Expected type of decision-making					<0.001
– Shared	106 (62)	61 (80)	22 (36)	23 (68)	
– Independent	51 (30)	6 (8)	36 (60)	9 (26)	
– Counsellor's	10 (6)	7 (9)	1 (2)	2 (6)	
– Uncertain	3 (2)	2 (3)	1 (2)	0 (0)	
Expecting to know other parents' decision	77 (45)	33 (43)	26 (43)	18 (53)	0.606
Expecting to know the counsellor's opinion	119 (70)	56 (74)	37 (62)	26 (76)	0.263
Unanswered questions have remained	9 (5)	2 (3)	2 (3)	5 (15)	0.024
Calming affect of consultation	110 (64)	56 (74)	38 (63)	16 (47)	0.025
Overall satisfaction with counselling	160 (94)	72 (95)	57 (95)	31 (91)	0.730

\* Numbers in the column do not sum to the total because not all respondents answered the question about education.

Expectations on the type of decision-making strongly affected the actual shared or independent decisions, as 80% and 60% of them respectively followed the expected type. Wishes associated with the mode of decision-making, however, did not influence the success of resolution, since the proportion of consultations that failed to agree on a decision was almost the same for each expected type of decision-making, and varied between 18 and 22%. As the possibility of getting to know the other parents' decisions and the counsellor's opinion was hypothetical, we were able to examine the impacts of these expectations, but not those of their fulfilment on decision-making. Data showed that the proportion of women wanting to know the preferences of other parents and of the

counsellor was greater in the group that did not reach a decision than in the other groups, though this difference was not significant statistically.

The evaluation of actual genetic counselling has explored that statistically significant correlations were found between the satisfaction with information and psychological support, and the result of the decision-making process (Table 8). According to the analysis, in the group with unsuccessful resolution, the proportion of women who had unanswered questions was higher, and the proportion of those who felt the calming effect of genetic counselling was lower than in the groups with successful resolution. Education affected with borderline significance the type and success of the actual decision. Among women with a college or university degree, the frequency of independent decisions was lower, and the frequency of not reaching a decision at all was higher than among women with a lower level of education. The satisfaction with the overall process of the consultation was high, independent of the type and success of resolution. Only the failure to answer all the counsellee's questions correlated significantly with the overall satisfaction ( $p = 0.008$ ), and had a negative effect on it.

### ***5.2.3. Evaluating the expectations of counsellees and comparing them with the views of counsellors***

Similar to studies conducted in Western countries, in our study the respondents have perceived the tasks of genetic consultations as not only to give comprehensive information, but also to provide psychological support and to facilitate decision-making [147,148]. Of the counselling types, the information guidance, the transitory, and the informative approaches have not received wide support. The counsellor's decision-making, the essence of the information guidance approach, was almost unanimously rejected (92%), and the majority of women intended to take part in the resolution process. Contrary to the transitory view, counsellees wanted more help than just information about the options of preventing diseases, e.g. they expected psychological support (68%). The appropriateness of non-directive counselling, the guiding idea of the informative approach, was questioned by those counsellees who wished to know the counsellor's preference about the course of action to follow (70%). The usefulness of neutral communication was also challenged, since the counsellees' successful decision-making required a type of consultation in which information and psychological support were tailored to the counsellees' individual needs, as in opposite cases the decision-making was postponed.

Compared with a study conducted in the UK in which 30% of counsellees expected help in making decisions [149], in our study this ratio was 68%. The extensive need for the counsellor's involvement in the decision-making has illuminated the women's uncertainty about the process of resolution, behind which the lack of knowledge or the wish to share

the responsibility of decision-making with the counsellor may be found [150]. However, in Hungary the lack of deep traditions concerning the autonomous decision-making of patients has also played an important role in the clients' behaviour of refraining from independent decision-making. Deriving from this circumstance, in our country the role of the counsellor in the independent and well-considered decision-making of the counsellee is more essential than in countries with old democratic traditions. The importance of the counsellor's activity in the process of resolution has been reflected in the data reporting that counsellees were apt to modify their preconception, and that they could even reach an independent decision as a result of the consultation (40% of the independent decisions were made in this way). The proportion of highly educated women expecting and making shared decisions reassured the findings of Wertz and Sorenson [151], which demonstrated that counsellees with higher education reported more frequently the counsellor's influence on their decision than clients with lower education. Data suggest that better educated women appreciate highly the counsellor's professional knowledge and can better utilise the information provided them. However, according to our research, the supposedly greater knowledge led to greater uncertainty as well, since the ratio of highly educated women reaching no decision was above that of groups with lower education.

#### ***5.2.4. Characteristics of genetic counselling meeting the expectations of counsellees***

The results of the study have given support to the kind of counselling method that adjusts professional help to the individual needs of the counsellees, allows the counsellor to play an active role in the course of decision-making, and excludes the dominance of the counsellor. Of the approaches promoted by genetic professionals, the interpretive counselling approach promises fulfilment of these requirements. As this method requires the counsellor to understand the counsellee's particular situation, the counsellee's individual wishes can be recognised and responded to, which could be especially useful for those women who remained uncertain about the decision after the consultation. The dialogical relationship, the main characteristic of the interpretive method, gives the counsellor the opportunity to distinguish cases in which the independent decision of the counsellee is a realistic option from those in which more help is necessary and shared decision-making should be aimed for. The application of the interpretive method allows the counsellor to disclose her opinion as well, which was expected by the majority of the respondents. This action would not encourage the revival of paternalism, since, in the interpretive counselling relationship, the values of the participating parties are explored and openly discussed, which can serve as a guarantee of finding a proposal that corresponds with the values of the counsellee and not with those of the counsellor.

## **6. Method of interpretive genetic counselling promoting the careful decision-making of counselees**

The results of our study on the counselees' expectations have strengthened the views of those who have demanded the implementation of new consultation methods that differ from the non-directive approach. It has also been revealed that counsellors should gain understanding about the situation of counselees in order to help them reach well-informed and well-considered decision. As in human understanding, besides psychological rules, philosophical characteristics play an essential role, I contend that the theory of interpretation, being explicated by Hans-Georg Gadamer in his book, *Truth and Method*, offers physicians an aid to reach the true understanding of counselees and to facilitate their decision [33]. Since V. Árnason systematised the Gadamerian concept according to the aspects of the patient-professional interaction, his views were used when I and a colleague were elaborating the methodological framework of interpretive genetic counselling [35], which could assist counsellors to incorporate the interpretative approach into their practice.

### **6.1. The Gadamerian four-openness model of interpretation**

Summarising Gadamer's theory on interpretation, Árnason has asserted that during dialogues four aspects of openness are necessary to understand the other and to take seriously her claims, such as openness to oneself, openness to the other, openness to the subject matter, and openness to tradition [33,35].

*Openness to oneself:* In Gadamer's view, during the process of understanding we all use our prejudgements and anticipations to project some meaning to the text, to the situation or to the person as a whole. These various cultural, personal or theoretical presuppositions constitute our horizons, without which any human understanding would not be possible. Openness to oneself requires the acknowledgement of these prejudices, and by this type of openness, dogmatic thinking may be escaped.

*Openness to the other:* This phenomenon means the listening to the other person and the perception of her difference. Gadamer has pointed out that the method of objective observation does not help us to recognise the differences of the other, since, if we remain emotionally unaffected, we can grasp only the typical features of the other person and we fail to take into consideration her subjectivity. It also does not lead to true understanding but only to the domination of the interpreter's perspective if the interpreter believes that she understands the other person better than the other understands herself. Gadamer has asserted that, as we are not able to fully comprehend the other, the best an understanding

person can do is to think with the other and to undergo the situation with her. In dialogues, the interpreter puts at risk her prejudices and is willing to accept the opinion of the other.

*Openness to the subject matter:* This idea reveals that during open dialogues, partners let themselves be conducted by the subject matter of the conversation and their dialogue is exclusively governed by the dialectic of questions and answers concerning the topic. Gadamer has drawn our attention to the role of questions in the process of understanding, stating that the act of asking questions expresses both the interpreter's awareness of the limitation of her perspective and her real interest in the opinion of the other. This attitude can generate the "fusion of horizons", and by the end of the conversation, partners can form an agreement on the situation, differing from the prejudgements of both of them.

*Openness to tradition:* This idea contains the notion that we should be conscious of our historical affectedness, which indicates that past traditions are built into present ones, so it is impossible for us to escape our traditions. We should know that in the course of interpretation, the meaning we project to the situation is directly related to our traditions. In Gadamer's view, tradition functions as a relevant authority, since it has preserved the wisdom of subsequent generations, and by listening to tradition, we can reach authentic understanding. In dialogue with the other person, the awareness of our tradition helps us to show openness to the tradition the other represents.

## **6.2. Process of interpretation in reproductive genetic counselling**

In the following, the four-openness model of interpretation is applied to the process of genetic counselling, where both the counsellor and the counsellee influence each other, thus, they should understand the other person and the other's role in the interaction [27]. Actually, double interpretation is performed; the counsellor interprets the counsellee, and the counsellee interprets the counsellor. An interpretation directed by the four-openness model yields the advantage of considering the four most important factors that affect the course of genetic counselling, i.e. the counsellor, the counsellee, the genetic risk or disease, and the contextual circumstances. Since, in the counselling relationship, the counsellor bears greater responsibility for the success of the consultation than the counsellee, the counsellor is expected to facilitate the counsellee's interpretation process as well [28].

### **6.2.1. Openness to the counsellor**

In genetic counselling, the openness to oneself demands the self-understanding of the genetic counsellor whose influence on the counsellee cannot be ignored. Thus, the

counsellor has to examine whether she accepts the promotion of the counsellees' interest as the goal of her activity, and if she interprets her professional task as to discuss moral, social, and emotional questions besides the medical ones [152]. The counsellor should make clear for herself her view about the severity of the disease and the quality of life affected by the disorder. These value judgements are unavoidably reflected in the counsellor's interpretation of genetic risk and in the description of the disease, so they may modify the counsellee's perception of the problem. The counsellor has to admit her preliminary concept about the course of action to be followed by the counsellee as this preconception could cause difficulties if the counsellee's idea differs from it. The counsellor's communication style is also worthy of self-examination, since she is expected to have an ability to generate an atmosphere where open communication can occur. This solid self-knowledge makes possible that when the counsellee asks for advice, the counsellor can explore the social, religious, and emotional backgrounds on which her solution is based. By disclosing explicitly her value orientation, the counsellor assists the counsellee's interpretation process, in which the counsellor is the other person whom the counsellee has to understand. On the basis of this transparency, women and couples can compare the counsellor's value system to their own ones, and in accordance with this assessment, they can determine if they accept or refuse the counsellor's advice [153].

### ***6.2.2. Openness to the counsellee***

The openness to the counsellee, the recognition of her difference is paramount in genetic counselling, because it determines the form of information and help. By showing openness to the counsellee, the counsellor does not remain untouched by her life problem, but she shares the troubles and difficulties the counsellee feels during the perception of the genetic information and in the decision-making process. The willingness of thinking with the counsellee gives authority to the counsellor to discuss questions belonging to the field of privacy. The counsellor needs to gain a notion about the vocabulary the other understands and the reasoning method she uses. Relying upon this knowledge, the counsellor has to adjust her communication style to that of the counsellee as the framing of information and the mode of reasoning affect the understanding of the situation of being at risk [154]. The counsellor explores the counsellee's knowledge on the given disease and her perception about the severity of it. The counsellor gets impressions about how firm or vague the value system of the counsellee is, and how much help is necessary in its clarification. As, in the majority of cases, the parents visit the genetic counsellor together, the counsellor should involve both of them in the course of counselling. The counsellor pays attention to the dynamics of the couple's relationship, since the couple's disagreement

or their responsibilities to other children have a definitive effect on the decision. Emotions can also come to the surface, and the counsellor can assess the level of anxiety and its role in the process of understanding, which may indicate demand for psychological support [155]. The issues posed by the counsellor contribute not only to the counsellor's effort to understand the patient, which is the basis for providing effective support, but also to the self-understanding of the counsellee, which is a pre-condition of careful decision making.

### ***6.2.3. Openness to the disease and its impacts***

In reproductive genetic counselling, the openness to the subject matter, i.e. to the genetic risk or disease, means that each life situation is unique, each parent reacts specially to the situation of having a high-risk pregnancy, so the counsellor should go through the interpretation process with each counsellee, even though the genetic disorder is the same. The counsellor presents the medical problem and the counsellee's inquiry forces the counsellor to personalise the medical information to the counsellee's needs. The counsellee's questions are not restricted to the factual nature of the disorder but extend to value-laden problems too. For example, the counsellee would like to know the counsellor's knowledge about life with a disabled baby or the counsellor's choice if she were in the other's place. The intention to enhance the counsellee's deliberation allows the counsellor to introduce unsolicited information and to challenge poorly reasoned choices [156]. This professional behaviour does not violate counsellees' autonomy but promotes cautious deliberation by drawing attention to unexplored problems. By the end of the open dialogue, the counsellee has completed her understanding with the counsellor's wider knowledge about the medical aspects of the disorder, and the counsellor has enriched her understanding about the disease with the counsellee's experience, which may be used as a starting point to understand other counsellees [32]. The common understanding of the situation enables the partners to find a solution together that is acceptable for the counsellee [157].

### ***6.2.4. Openness to the cultural and social circumstances***

During open dialogues, the counsellee and the counsellor open up together the wider social and cultural contexts in which reproductive autonomy has got its meaning. The collective opinion on the acceptance of prenatal tests, on the termination of pregnancy, on living with disability, and on the parental responsibilities concerning the upbringing of a severely disabled child indicates to the parents the ways of conduct the community could accept. In Hungary, for example, the public opinion supports the termination of an affected pregnancy, honours families caring for children with Down's syndrome, but rejects the

behaviour of the parents who abandon their disabled children and leave their care to state-financed institutions. The social or familial expectations may be assessed as constraints on autonomy—although, only if autonomy is perceived as absolute freedom—but their acknowledgement helps parents prepare for unpleasant reactions and find coping strategies if they decide against the public opinion. In connection to medical genetics, the respect for traditions calls attention to the importance of two social value commitments, the rejection of eugenics and the respect for reproductive autonomy [158], which demand that the genetic counsellor should abstain from determining the content of the decision.

### **6.3. Advantages and disadvantages of interpretive genetic counselling**

An advantage of interpretative genetic counselling appears in the feature that while the prevailing non-directive approach favours impartial information disclosure, the interpretive method, based on the true understanding of counselees, offers the possibility of tailoring information to the real needs of counselees. This new method also accepts that the counselees' expectations go beyond the limits of getting information and believes that the facilitation of counselees' decision-making and the provision of psychological support are essential parts of genetic counselling. By exploring the importance of the awareness of the counsellor's prejudices and values, the interpretive approach presents advantages to those initiations as well that intend to support the patient's deliberation by shared decision-making, however, fail to examine the effect of the physician's perspective on that activity [159].

The possibility that the counsellor's involvement in the decision-making might encourage the revival of paternalism means a disadvantage for the interpretive approach. However, if clear distinctions are made between the interpretive method and medical paternalism, the respect for the reproductive liberty of counselees and the active support of their decision-making could be simultaneously ensured. The Gadamerian thought implies the warnings that the physician cannot surely know what the patient's perception about her best interest is, and therefore, it is inadmissible to force a solution on the patient. Regarding the positive features of the interpretive approach, I argue that the communication training of genetic counsellors should be completed with the discussion of the role of interpretation in the counselling interaction. Although interpretation is a genuine intellectual activity, which cannot be adequately performed by following a simplified algorithm, I'm convinced that the four-openness model of interpretation can work as a useful guideline for counsellors to engage in open dialogues with counselees and to understand them.



## 7. Ethical importance of discussing publicly the pre- and postnatal issues of Down's syndrome

In our country, where, despite the fact that different screening methods are offered to pregnant women by university clinics and by other public or private health care institutions [160-168], a nationally organised prenatal Down's syndrome screening programme has not yet been introduced, discussing publicly the issues of prenatal screening is much more demanding than in countries with well-organised screening programmes. In those countries where prenatal Down's syndrome screening looks back to a three-decade long history, professionals of medical genetics have recognised that they should take responsibility for both the women who participate in screening and the patients who were born with the screened disorder [42]. In their views, this responsibility involves the acknowledgement of those social and ethical implications that are induced by the implementation of prenatal genetic screening and that influence the self-esteem and social status of individuals with disability. In Hungary, in the lack of a national prenatal screening policy on Down's syndrome, the fulfilment of these tasks has remained to relevant scientific conferences, of which the most regularly held and the most comprehensive is the Down Syndrome Symposium of Szeged, organised by the local university's Department of Medical Genetics. From its beginning in 1998, the Symposium has provided a nationwide forum for medical and non-medical professionals, for experts of prenatal and postnatal care, and for the parents having children with Down's syndrome to present their activities and to acquire knowledge on the activities of the other parties (Table 9). In this chapter, the ethical impacts of the Symposium are examined from the viewpoints of prenatal screening and postnatal care.

**Table 9:** Thematic division of presentations performed at Down Syndrome Symposiums of Szeged, 1998-2008

Topics	1st symp.	2nd symp.	3rd symp.	4th symp.	5th symp.	6th symp.	7th symp.	Total n (%)
1 Prenatal screening for DS	8	3	3	6	3	5	9	37 (31)
2 Biomedical description of DS	6	1	1	4	1	2	-	15 (13)
3 Ethical issues	3	3	2	1	2	2	1	14 (12)
4 Children's early intervention, education	2	2	3	1	2	2	-	12 (10)
5 Postnatal medical problems	3	1	2	1	2	1	1	11 (9)
6 Parent support groups' activities	3	2	3	-	-	-	1	8 (7)
7 Genetic diagnostic methods of DS	2	2	-	-	2	-	1	7 (6)
8 Epidemiology of DS	2	-	1	2	1	-	1	7 (6)
9 Self-reports of parents, of adults with DS	2	1	-	3	-	-	1	7 (6)

DS = Down's syndrome

### 7.1. Impacts of public discourse on prenatal Down's syndrome screening

The topics of prenatal screening and diagnosis were extensively represented in the programmes of the subsequent symposiums (Table 9), which was owed to the professional orientations of the organisers and to the current state of prenatal Down's syndrome screening in Hungary, which was aimed to be modified by the contributions of the screening professionals attending the meetings.

### ***7.1.1. Gaining public support for the introduction of a formal prenatal Down's syndrome screening programme***

One of the most significant issues where prenatal screening professionals have needed the support of health care policy makers and the wide public refers to the introduction of a universal prenatal Down's syndrome screening programme. At the Symposium, the need to change the official screening concept, which was developed in the 1980s and has included only the offer of amniocentesis for pregnant women aged 35 years or more, was justified by the reason that a scientifically outdated and ethically unjust screening policy should be stopped [43,44]. It was contended that only a formal screening programme could ensure the provision of screening to every pregnant woman, which was required by epidemiological data showing that in Hungary, between 2000-2004, among women giving birth to a Down's syndrome baby, the proportion of those being younger than 35 years was around 60% [11]. Universal screening could also fulfil the requirement of justice by providing equal access of care for those women, who, because of their lower education and social status, could hardly access to prenatal screening. Practitioners also warned that for the introduction of a prenatal screening programme, a wide range of conditions should be guaranteed, e.g. technical equipments for screening and diagnosis; software programmes for risk calculation; training for practitioners; availability of genetic counselling, chorion villus sampling, amniocentesis, and pregnancy termination; and appropriate budget to finance the programme. Besides urging the implementation of a national screening programme, the Symposium has also intended to function as a post-graduate training that informs practitioners about the latest screening technologies and about the ethical, psychological, and communication implications of prenatal Down's syndrome screening, which should be addressed by a formal screening programme as well.

Obviously, Down Syndrome Symposium has not been the only forum supporting the change of the official screening concept. For instance, in 2004, the College of Clinical Geneticists recommended performing 11-13 weeks ultrasound scan as a part of prenatal Down's syndrome screening protocol, and offered maintaining the practice of second trimester ultrasound scan as well [12]. This measure has increased the spread of first trimester nuchal translucency screening, although, with significant regional differences [11]. Therefore, introducing a well-organised, national screening programme has remained a prominent task of health care policy, since by this programme, the use of outdated screening methods can be avoided, every pregnant woman can access to screening, and ethical, psychological and communication issues can be adequately addressed.

### ***7.1.2. Building social consensus on the application of prenatal screening***

Discussing the topics of prenatal screening before an audience that involved parents, teachers, and physicians caring for children with Down's syndrome might seem controversial, however, the vivid ethical debate emerging on the goal of prenatal screening among participants with different expertise and interest showed the benefit of the common forum. Eventually, in the lack of a national screening policy, the task of setting off a social debate on the application of Down's syndrome screening was undertaken by the Symposium. Including the public in the discussion of the ethical and social impacts of medical genetics is internationally encouraged [169], and this process is especially important in Hungary, where the tradition of debating health care issues publicly is young. Since many professionals confronted with the ethical and social issues of prenatal screening only at the Symposium, the forum has played a pivotal role in the acknowledgement of these contentious topics.

Through the series of debates, some elements of a consensus on the implementation of prenatal Down's syndrome screening were outlined. The parents' sharp criticism on prevention as the aim of screening revealed that during the application of prenatal screening, not only the interests of pregnant women wanting to avoid the birth of a disabled child should be taken into consideration but also the interests and sensitivity of people living with disability [170]. The notes of the parents and paediatricians reminded screening professionals that because of their social responsibility towards disabled people, they should abstain from justifying screening by its potentials for decreasing the prevalence of Down's syndrome or for sparing finances when its cost is compared with the cost of care of disabled patients. The right of parents for using prenatal screening technologies has been accepted even by families having children with Down's syndrome [171]. However, the parents have expressed their wishes about using a language in public discourse that lacks discriminatory meaning against disabled people and does not include the assumption that the birth of a child with Down's syndrome constitutes a tragedy.

The personal experience that practitioners gained at the meetings played a decisive role in the modification of their opinions on some ethical questions. For example, a growing number of physicians have accepted that giving information about the genetic risk of the foetus should be considered as the aim of screening. Even though the aim of screening has remained a matter of debate, the initiation of the sixth symposium about omitting the phrase of prevention from public communication of prenatal screening issues gained wide support among providers of prenatal screening [172].

## **7.2. Impacts of public discourse on postnatal care for people with Down's syndrome**

By including the discussion of the medical, educational, and social questions of postnatal care of individuals with Down's syndrome in the programme of the Symposium, the organisers have expressed the notion that the provision of prenatal screening and the improvement of the quality of life of people with disability are equally fundamental. Emphasising the importance of improved care and social inclusion of people with Down's syndrome has of ethical importance as prenatal screening is frequently criticised for its features of strengthening negative attitudes towards disabled people and jeopardising the results of the disability rights movement [16]. Inviting paediatricians, public nurses, teachers, therapists, parents and adults with Down's syndrome to present the issues of postnatal care can convey the message that screening professionals are aware of the possible negative impacts of prenatal screening and they want to avert them.

### ***7.2.1. Increasing public awareness about lives of people with Down's syndrome***

Because of the scope of the audience, which was much wider than that of a traditional conference discussing separately the pre- and postnatal issues, the Symposium has fulfilled a special function in the increase of the community's awareness about the day-to-day lives of people with Down's syndrome. Giving a realistic account on this issue is paramount, as the lives of people with Down's syndrome is hardly known by the wide public, and even by biomedical professionals who meet disabled people only in the medical setting [173]. Stemming from these reasons, prenatal screening professionals especially welcomed the lectures presenting the lived experience of people with disability as they could get first-hand information on the recent prospect of disabled people about which they rarely had any practical experience. Nonetheless, being well-informed about the educational and social opportunities provided for disabled people belongs to the professional requirements of screening practitioners, since they are expected to give pregnant women balanced information on Down's syndrome. By making medical professionals aware of the non-medical characteristics of Down's syndrome and of the support services available to disabled people, the Symposium has helped physicians to avoid giving parents biased information, which focuses mainly on the medical problems.

At the meetings, the audience recognised with surprise how well-organised and strong the parent self-support groups were in Hungary, and how wide range of services were offered by them to promote families having a disabled child to control their lives. The capability of many people with Down's syndrome to lead an assisted independent life and

the parents' efforts for ensuring this kind of life for their children were almost unknown by the majority of the participant. The importance of changing the attitude of the public towards disabled people was illuminated by the experiences of the parents, which revealed that despite to legal guarantees, to find a supportive school remained difficult and disabled people had hardships in getting jobs after school years [174-176]. Although, for lay participants performing a lecture at a scientific meeting means a considerable challenge, their regular contributions have proved indispensable in reducing negative attitude towards disabled people and in modifying stereotypes associated with disability.

### ***7.2.2. Improving the quality of life of people with Down's syndrome***

Besides giving forum to relevant lectures, the Symposium also supported the improvement of the lives of people with Down's syndrome by giving opportunity for the parents and medical professionals to make contact and develop co-operation. The parents grasped this chance and addressed some fields where the two parties could mutually help each other. One of them referred to the issue of disclosing information about the affectedness of a newborn baby, and in order to prevent inappropriate practices, the parent support groups offered their help for obstetricians to develop a method of non-delayed, emphatic, and detailed information giving. The proposal was embraced and as a first step, a survey was conducted about the parents' expectations on information by the National Institute of Epidemiology in order to help obstetricians to construct protocols on informing parents about the birth of a disabled child [177]. Another occasion for co-operation emerged in the field of psychological support of new parents, and obstetric units were requested to make the information available that families could get support from fellow parents if they asked for it. The parents initiated the implementation of Down's syndrome clinics as well, since this outpatient care had been available only in the capital, and by the collaboration of paediatricians, a new clinic was established in the Department of Paediatrics, University of Szeged [178]. The familial climate of the Symposium, which was in great part generated by the parents who were accompanied by their children, promoted the frank dialogue and the co-operation of professional and lay participants.

The ten-year long history of Down Syndrome Symposium of Szeged has shown that conferences discussing the pre- and postnatal aspects jointly can reconcile the claims of antenatal screening and the interests of people with disability. These meetings offer the chance to preserve important social and ethical values, e.g. justice, respect for reproductive autonomy, equal moral value of disabled people. Considering the implementation of this type of symposium would be especially useful in those countries where the introduction of a prenatal Down's syndrome screening programme is in preparation.

## 8. Conclusions

In Hungary, the study of prenatal Down's syndrome screening has represented the first example of a practice where a medical ethicist was deliberately involved in the work of a medical department in order to explore the ethical issues of this procedure. For me, as a medical ethicist, this co-operation offered a unique opportunity to combine medical ethics theories and the screening experiences of professionals and pregnant women. Using this approach, the following new results have been yielded by the PhD thesis:

**1) Ethical acceptability of prenatal Down's syndrome screening:** In the Thesis, I have challenged the popular idea that the goal of prenatal Down's syndrome screening is disease prevention. I have asserted that this goal violates essential ethical norms, i.e. interferes with the reproductive autonomy of parents and includes eugenic and discriminatory hints against people with disability. I have contended that this medical procedure can be accepted on ethical grounds only if its goal is defined as enhancing the reproductive autonomy of parents and if the voluntary participation of women is ensured.

**2) Implications of an ethically justifiable aim on medical practice:** I have stated that the goal of enhancing reproductive autonomy has an effect on the overall process of screening, especially on the applied screening technology, patient information, doctor-patient relationship, and method of genetic counselling.

*Screening methods:* On this issue, I have asserted that the great number of methods, concerns about reliability of data on efficacy of tests, changes of professional attitudes to ultrasound scan, wider spread of first trimester policies, and uncertainty about the role of maternal age in screening have led to confusion among practitioners and pregnant women. I have contended that the confusion on screening technology would be dissipated by professional guidelines, which have to include some basic requirements of tests, such as individual risk assessment, high sensitivity (85% or more), and low false positive rate (5% or less). Since not only one but some methods can meet these expectations, other needs deriving from specific medical, ethical or economic considerations can also be satisfied. Thus, I have proposed that professionals should clearly define what technology could fulfil best the given needs and these data should be disclosed to pregnant women.

*Information:* I have emphasised that giving women pre- and post-test information should be considered an inherent part of screening. As, in my view, not only detailed but also personalised information is necessary to facilitate women's decision, I have interpreted the professionals' and the pregnant women's attitudes to screening and have constructed a guideline to help professionals provide verbal or written pre-test information. I have suggested that an information leaflet should be given to all women prior to screening and communication training should be included in the post-graduate education of screening professionals, where the data of screening tests and the process of risk communication are taught.

*Doctor-patient relationship:* To get insight into the genetic counselling relationship, the views of counsellors on the goal of genetic counselling were explored, and a research was conducted on the genetic counselling expectations of pregnant women. Based on our research data, I have revealed that, besides detailed information and psychological support, women expected help with decision-making as well, and the fulfilments of expectations affected decisively the success of decision-making. Since, of the divergent genetic counselling methods promoted by counsellors, the interpretive method promised fulfilment of counselees' expectations, I have proposed a shift from recent counselling methods to interpretive genetic counselling. The results and proposals of our research inspired the editors-in chief of EJOG & RB to launch a debate on genetic counselling in the journal.

*Method of genetic counselling:* In order to get knowledge about the counselees' expectations and to help their decision-making, I have asserted that counsellors should understand the counselees' situation. To promote the understanding process, I and a colleague have elaborated a methodological framework of interpretive genetic counselling that helps clarify the four most important factors of genetic counselling: the counsellor's attitude; the counselee's values and needs; the medical, social, and moral impacts of the disease; and the social context. I think, if the interpretive method is properly applied, counsellors can give counselees personalised information and emotional support, and can facilitate their careful deliberation without exerting paternalistic influence on them.

**3) Wider social impacts of prenatal Down's syndrome screening:** To emphasise the importance of taking into account both the needs of pregnant women and the interests of families having an affected child during the implementation of prenatal screening, I analysed the ten-year work of Down Symposium of Szeged that discussed jointly the issues of pre- and postnatal care. I have assessed the practice of the Symposium as worthy to follow, since it supported simultaneously the spread of prenatal screening and the improvement of the quality of life of people with disability. Concerning Down's syndrome screening in Hungary, I have promoted the introduction of universal screening, since only a formal programme can prevent the use of outdated methods, can ensure every pregnant woman access to care, and can adequately address ethical and communication issues.

In summary, the results of the Thesis can be applied as ethical guidelines by prenatal screening practitioners, relevant professional bodies, and health care policy makers. The data on the counselling expectations of women and the method of interpretive genetic counselling can be utilised by genetic counsellors and can be included in their post-graduate training. In academic education, the Thesis can serve as an example to present how medical ethics theories and day-to-day medical experiences can be combined. For medical ethicist, the Thesis can function as an encouragement to conduct descriptive ethical researches by which the effects of the specific Hungarian health care context on the operation of widely accepted ethical norms could be explored.

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# APPENDICES

# **Appendix 1**

**Adél Tóth, Tibor Nyári, János Szabó:**

**Changing views on the goal of reproductive genetic counselling in Hungary.**

*European Journal of Obstetrics and Gynecology & Reproductive Biology* 2008;137(1):3-9.

## **Appendix 2**

**Adél Tóth, Péter Szeverényi:**

**Interpretation in reproductive genetic counseling: a methodological  
framework.**

*Journal of Psychosomatic Obstetrics and Gynecology* 2007;28(3):141-145.

## **Appendix 3**

**Tóth Adél, Szabó János dr.:**

**A tájékoztatás legfontosabb etikai vonatkozásai a Down-szindróma praenatalis szűrése során.**

*Orvosi Hetilap* 2005;146(6):259-265.

## **Appendix 4**

**Tóth Adél, Szabó János dr.:**

**Miért nem lehetséges a semleges jellegű genetikai tanácsadás?**

*Magyar Nőorvosok Lapja 2005;68(0):113-120.*

## **Appendix 5**

**Adél Tóth:**

**The role of values in genetic counselling.**

*Studia Bioetica* Volume 2, p. 1-7, 2005

<http://utopia.duth.gr/~xirot/BIOETHICS> - accessed 10/05/2009

## **Appendix 6**

**Tóth Adél, Szabó János dr.:**

**A Down-szindróma praenatalis szűrésének néhány etikai vonatkozása.**

*Orvosi Hetilap* 2000;141(42):2293-2298.

## **Appendix 7**

**Tóth Adél:**

**Az értelmező orvosi etika szerepe a Down-szindrómás gyermeket nevelő családok és az orvosaik közötti kapcsolat kialakításában.**

*Pediáter* 2000;9(4):297-305.



## **Appendix 8**

**Tóth Adél:**

**A Down-szindróma prenatális diagnózisát követő terhességmegszakítás etikai kérdései.**

*Pediáter* 1999;8(4):311-316.

## **Appendix 9**

**Adél Tóth:**

### **The birth of bioethics and its basic principles.**

In: Bettina Pikó (ed.): Introduction to medicine. Basic principles of behavioral sciences and preventive medicine. University of Szeged, Szeged, 2004. p. 133-145.