Fundamental Problems and Solutions Concerning Genetic Testing (2nd part)

Tierney Bennet, L.C.

3. Genetic testing, risks and results

3.1.The weight of the risk factor in decision-making

One of the determining factors for many when the time comes for a serious personal decision is precisely that of risk, which also has its moral implications. Although levels of risk tend to be easily expressed in hard mathematical terms, what such numbers mean for individuals is highly variable and subjective, and depends upon their situation. Probabilities that are not 0 or 100% cannot predict certainty. Few people are aware that there is at least a 3% risk for significant birth defects or genetic disorders in the general population. Even if they were aware, it would probably not make much difference to their decisions to have or not have children, since there is little one can do to reduce that risk. On the other hand, a 3% chance of dying following a surgical procedure would probably seem quite high, and one might ask if the operation was really necessary, and what the alternatives were. Individuals differ in their attitudes to risk; some are naturally conservative, and would like to limit as many risks in their lives to as close to zero as possible. Others are prepared to accept, or even actively take risks if the possible benefits seem to be enough to justify them. Such decisions are especially difficult when the risk is a genetic one, with potential lifetime consequences not only for them, but also for another person, namely, their child. Sometimes a parent's previous experience with the disease causes it to be seen as a greater burden than when nei-

Alpha Omega, V, n. 3, 2002 - pp. 473-497

ther parent has had prior experience. For example, a women who is a carrier of Duchenne muscular dystrophy is more likely to be determined about reducing her risk of having an affected son if she has personally helped to care for her affected brother than if the family history is more distant.

Moreover, because of a lack of intercommunication, the gap between science and the public becomes greater, the public understanding of genetics becomes less, and a certain mistrust in science increases. In spite of all that is said, the fact is that for most genetic diseases gene therapy appears only as a remote possibility. In 1994, a Gallup poll in the UK showed an almost threefold increase from autumn 1993 to September 1994 in public support for the use of genetic techniques to enhance desirable genetic traits in children; however, with no more than 20% in any case the overall figures remain relatively small.[1]

One of the main reasons why so many woman are scared stiff of the natural process of childbirth and develop the phobia of compulsively having to know what is going on in their womb, are the profit seeking organizations and institutions that capitalize on women's natural curiosity and fears by willfully exaggerating the risks involved in childbirth. Many women run to the clinics at the first sign of a pregnancy, and many undergo prenatal diagnosis just in case something may go wrong. And when it does, abortion is all too frequently considered "the solution."

The risks of childbearing and the chilling specter of grossly malformed infants are often used in campaigns to keep abortion legal. They normally concentrate exclusively on the extreme hard cases, such as infants with anencephaly, multiple gross physical malformations, and the most severe imaginable cases of Down syndrome. It is argued that these babies have such a low "quality of life" that they would not have a meaningful existence; it is considered that caring for such a child would decrease a couple's own quality of life and the meaningfulness of their existences.[2]

---

[1] Cf. Verrall M.: *Demand Grows for Positive Gene Therapy*, Nature, Volume 371, Issue 6694, 15 September 1994, page 193.

[2] Cf. Singer P.: *Sanctity of life or quality of life?* Pediatrics, Volume 72, Issue 1, July1983, pages 128-129. The comparisons of this author are very graphic indeed: "If we compare a severely defective human infant with a dog or a pig ... we will often find the nonhuman to have superior capacities ... Only the fact that the defective infant is a member of the species Homo Sapiens leads it to be treated differently from the dog or pig. But species membership alone is not relevant ... If we can put aside the obsolete and erroneous notion of

Some risks are overrated, while other risks are silenced. The role of the media and some sectors of the medical community tend to convince the public that birth defects are rampant in certain classes of people, for example sickle cell anemia among Afro-Americans, or women over 35 years old.[3] As regards the latter, the natural tendency of the public is to overestimate the probability of rare but disastrous consequence; the average person is lead to believe that nearly half of all children born by women after the age of 40 have serious birth defects, which is by no means true, but yet the medical community does not do a great deal to help clear up this severe degree of misunderstanding; on the contrary, frequently all pregnant women over the age of 35 are automatically classifying as "high risk."[4]

Consequently, even though early disease diagnosis could be routine in the near future according to some scientists, currently there are also a number of practical limitations.

Retroviruses, for example, can enter the human body and change cells *in vivo*, but it is obvious that the use of such viruses causes severe biosafety problems. The HIV has achieved to pass the nuclear membrane, making it a model for future approaches to gene therapy. But legislation, in face of psychological problems, will presumably not permit the use of HIV-like viruses. However, other carriers like liposomes or the use of naked DNA have proved less effective. Presumably, none of these will be the method finally applied because all have their problems and disadvantages.

In spite of this, women still take the risks and physicians push them to do so regardless of the cost. The matter of cost is not indifferent and health care reform and cost-savings are at the top of the medical agenda of most European countries. The costs of a single PCR analysis, based on urine specimens, amounts to at least $25 a test; in cases of simple tests and a high percentage of carriers, that would be as nothing if it permitted diagnosis sufficiently early to result in definitive cure of disease.[5] However, in the case of more complex diseases, high number of mutations and/or low frequency of the genes,

---

the sanctity of all human life, we may start to look at human life as it really is: at the quality of life that each human being has or can attain."

[3] Cf. Handler D: *More from Hymie Gordon on Genetics*, National Right to Life News, July 1978, page 6. (The author quotes Hymie Gordon, M.D., Chairman, Department of Medical Genetics, The Mayo Clinic).

[4] Cf. Ibid.

[5] Cf. Culliton B.J.: *Hubert Humphrey's bladder cancer*, Nature, Volume 369, Issue 6475, 5 May 1994, page 13.

the costs to identify carriers can be extremely high and nearly economically impossible. One of the big risks that will probably ensue as more progress is made, is that the combination of commercial and scientific interests will push for rapid applications of genetic engineering in order to generate profits and new knowledge.

Another practical issue is reliability. Surely this should be taken into account if risks are to be considered worthwhile, that is to say, proportionate and beneficial. As with pathology, there is still a measure of art in the application of PCR to human tissue. More has to be learned regarding the natural history of some mutations, like those in bladder cancer predisposition, before diagnosis can be certain. When considering risks there is the important question of the therapeutic benefits of screening. The early detection of predisposition to more frequent but also more complex genetic diseases (cancer, diabetes, hypertension) is possible without pathology. Treatment can begin at an early stage with better prospects for healing. The gene variants contributing to complex diseases are likely to be common polymorphisms which, in the absence of the other risk factors, do not cause disease.

On the other hand, germ line gene therapy by injecting genes into fertilized eggs poses other risks not only for the individual but also for oncoming generations, as some fraction would carry the altered genes. With regard to the future, those surveyed in the course of a study issued by the Office of Technology Assessment (OTA) disagreed markedly about whether gene therapy would emerge as a major treatment modality over the next 10 to 20 years.[6] Some saw it as the coming wave of therapeutics, others as something yet remote from everyday life. Gene therapy might be useful for cancer therapy, fatal genetic diseases of childhood, and other extremely serious conditions lacking better therapy. The original notion of human gene therapy that was to treat single gene defects has given way to seeing it as a way to introduce genes into cells that can act as drug delivery devices. This opens a far more diverse set of possibilities. Cells treated with inserted genes could conceivably be used to treat AIDS, heart attacks, diabetes, as well as cancer. Several recent protocols approved or in preparation already illustrate the broader possibilities.[7] The range of disorders that can be approached by gene therapy will presumably not increase appreciably until better cell targeting and chromosome-site targeting are

---

[6] Cf. United States Congress, Office of Technology Assessment: *Pharmaceutical R&D: Costs, Risks and Rewards*, U.S. Government Printing Office, Washington DC, Febuary 1993.

[7] Cf. Ibid.

possible, and these may well take at least a decade of research to develop, if they develop at all. This is an important factor regarding the diagnosis for genetic defects: the point is that the risks involved would be of no avail if there were no possible therapy to cure them.

3.2. The justification of risk

It is only right that special attention should be reserved for the moral evaluation of tests that permit the discovery of eventual abnormalities, for example, in embryos. When such techniques are free of disproportionate risks for the child and the mother, when they respect the integral health and life of both, and when the aim is to render possible an early therapy or favor a serene acceptance of the child, then these techniques can be morally licit.[8]

However, given that these methods still have very few therapeutic possibilities, all too often they are put at the service of an eugenic mentality that accepts selective abortion as a means to inhibit the birth of children with defects. Such a mentality is to be reprimanded because it pretends to measure the value of a human life only according to parameters of normality and physical wellness, thus opening the way for infanticide and euthanasia.[9] Curiously, there are people who will fight for more handicapped parking slots at supermarkets, but stand silently by while handicapped newborns are starved to death in nurseries. Shelters are set up for battered and abused women, but the same people will then approve of and facilitate sex-selection abortions that are directed almost exclusively towards exterminating those pre-born children whose only birth defect might be that they arhe female.

The history reminiscent of a legal Nazi mentality replete with examples of forced contraception, abortion, euthanasia and even genocide, all committed in the name of purifying the race, should never be tolerated by society again. History also shows us what results from such programs. They always begin as the provision of a service; examined closely and declared beneficial to society, they soon become compulsory. Whether the State legally kills 500 retarded or useless citizens, or a husband and wife kill their less than perfect pre-born child, the philosophy, the motivation and the results are basically the same. The philosophy is that there is life not worth living, and that it is best eliminated. The motivation is to eliminate those who are “burden-

---

[8] Cf. John Paul II: *Evangelium Vitae*, Acta Apostolicae Sedis, Libreria Editrice Vaticana, Città del Vaticano, 2 May 1995, number 63.

[9] Cf. Ibid.

some" and who are not "cost-effective." The result is not only the deaths of the handicapped, but the deaths of consciences as well. In other words, whoever pushes abortion for birth defects has a mentality that can be uniquely identified as classically nazi-like. This is not stereotyping or slander; it is an easily proven and logical conclusion.

Anyway, to talk about breeding out genetic diseases is nonsense. Seriously affected persons are unlikely to marry and have children; the genes are usually passed along by carriers. For instance, there are 40 carriers for every person with sickle cell anemia; if every victim of this disease were eliminated, it would require 750 years just to cut the incidence in half; to stamp it out altogether would require 200,000 abortions for every 500,000 couples; and because each "normal" person is the carrier of three or four bad genes, the only way to eliminate genetic diseases would be to sterilize or abort everybody.[10] This sounds ridiculous, and yet there actually are people who advocate for it. Besides, the total risk of a serious fetal defect for a woman of 40 is less than one percent, and not the 50 percent perceived by the public, thanks to statistic manipulation. The extremely low percentage of birth defects are confirmed by a number of surveys performed in the United States and other countries. These studies were based upon legally mandated record keeping by abortion clinics.[11]

Of course a strictly therapeutic intervention in which the objective is the cure of disease, like those that regard genetic deficiencies, is clearly auspicious.[12] But to overcome risks at the moment there are several practical issues that require resolution, apart from a better understanding of the inheritance of diseases, the lack of treatment in most cases and ethical and social problems.

Somatic gene therapy seemingly poses less moral problems as long as benefits and risks are taken into account, human dignity respected and the patient gives his or her informed consent. It is considered ethical in most countries, but it benefits only the recipient and not future generations. Some cell types are more attractive than others to act as recipients for genes. The haematopoietic system is an obvious candidate because pluripotent stem cells are easy to remove from the body, and the engineered cells would be easy to replace into the body.

---

[10] Cf. Handler D: *More from Hymie Gordon on Genetics*, National Right to Life News, July 1978, page 6.

[11] Cf. Ibid.

[12] Cf. John Paul II: *Arbitraria e ingiusta la manipolazione genetica*, All'associazione medica mondiale, Insegnamenti di Giovanni Paolo II, Libreria Editrice Vaticana, Volume 6, Issue 2, 29 October 1983, number 5.

Somatic gene therapy has been analyzed by many European medical research councils and also by major religious, governmental, and public policy bodies, including the Roman Catholic Church, the World Council of Churches, the US Presidential Commission for the Study of Ethical Problems in Medicine and Biomedical Research, and the Parliamentary Assembly of the Council of Europe; they have concluded that the concept is ethically acceptable.[13] It is seen by most in the same light as any other form of organ transplantation. In practice, there are still safety problems associated with somatic gene therapy,

---

[13] Cf. Anonymous: *Gene therapy in man. Recommendations of European Medical Research Councils*, Lancet, Volume 1, Issue 8597, 4 Jun 1988, pages 1271-1272. Basically the article gives five recommendations: 1. The purpose of gene therapy currently under consideration is the correction of genetic defects; attempts to enhance general human characteristics should not be contemplated. Only somatic cell gene therapy, resulting in non-heritable changes to particular body tissues, should be contemplated. Germline therapy, for introduction of heritable genetic modifications, is not acceptable. Further technical improvements in the expression of transferred genes in somatic cells will be necessary before successful gene therapy can be achieved even in animal models; in the meantime trials in man are not justified. 2. The most appropriate "candidate" genetic diseases for early investigation of treatment by gene therapy are single-gene disorders for which the affected gene and its regulation have been characterized. 3. In the near future, it is likely that success in the introduction of normal genes into human cells will be achieved through the use of disabled retrovirus vectors, although other techniques may advance rapidly. Much further work is required in the development of safe species-specific and tissue-specific retrovirus vectors. The methods of gene introduction should not result in the spread of gene or vector to other tissues within the body or to people in contact with the patient. The possibility of a significant increase in the predisposition of the patient to cancer should be evaluated in considering the risks and benefits of the treatment. In addition, the expression and regulation of the gene inserted should be stable and sufficient to ensure a therapeutic effect. 4. General ethical considerations applicable to any new clinical treatment apply to human gene therapy and, in the first instance, will require assessment in individual cases. In the near future it is likely that such therapy will be clinically justified in particular patients with invariably fatal or life-threatening diseases, provided informed consent is obtained and no alternative treatment is available. 5. A national body should consider all proposals for human gene therapy and ensure the application of agreed national guidelines. Early trials should be monitored by a central body.

Also cf. Anderson W.F.: *Human gene therapy: scientific and ethical considerations*, Journal of Medicine and Philosophy, Volume 10, Issue 3, August 1985, pages 275-291. The term "gene therapy" encompasses at least four types of application of genetic engineering for the insertion of genes into humans. Somatic cell gene therapy is technically the simplest and ethically the least controversial. Germ line gene therapy will require major advances in our present knowledge and it raises ethical issues that are now being debated. In order to provide guidelines for determining when germ line gene therapy would be ethical, Anderson presents three criteria which should be satisfied prior to the time that a clinical protocol is attempted in humans. Enhancement genetic engineering presents significant, and troubling, ethical concerns. Except where this type of therapy can be justified on the grounds of preventive medicine, enhancement engineering should not be performed. The fourth type, eugenic genetic engineering, is impossible at present and will probably remain so for the foreseeable future, despite the widespread media attention it has received.

since viruses are generally used as shuttles for the genes, and there are fears of the virus infecting previously healthy individuals.

The first approved human gene therapy experiment was conducted in 1989 by Rosenberg, Anderson, and Blaese for the US National Institute of Health; they used tumor infiltrating Iymphocytes to carry a marker gene for neomycin resistance into tumors of patients with terminal melanoma; in 1990, the same people transfused a little girl with Iymphocytes bearing the adenosine deaminase gene via a retroviral vector;[14] the preliminary findings are that the technique worked.[15]

Safe germ-like therapy is not yet technically possible, since the site of integration of a gene cannot be controlled. Although a gene could technically be introduced into the germ-line, it might knock out another gene, or it might land in a region of the chromosome which is not expressed. For these reasons, in the United Kingdom, the Clothier Committee on the Ethics of Gene Therapy has ruled that germ-line gene therapy is presently unethical.[16] In fact, in most countries this type of gene therapy is at present forbidden when applied to human beings. In the many discussions among scientists, philosophers and theologians, the more acceptable is "corrective" gene therapy applied to embryos having some hereditary disease. However, the procedures give little hope as regards a justifiable moral solution, as most imply the artificial fertilization of an egg, the creation of an embryo, followed by genetic treatment and then the implantation in the woman.

Up to now treatment in most complex diseases has been limited to amelioration of the symptoms rather than tackling the underlying causes. Identification of the new disease-related genes may suggest new therapeutic and preventive strategies and enable environmental influences to be assessed. In the absence of genetic differences predisposing to the disease, environmental differences affecting the development of the disease can be pinpointed, so individuals at risk would have the option of modifying their exposure to the environmental risk factors.

Several aspects of genome analysis may yield a positive impact on patient care. First, for some genetic disorders presymptomatic

---

[14] Adenosine deaminase deficiency causes severe combined immunodeficiency.

[15] Cf. Morgan R.A.- Anderson W.F.: *Human gene therapy*, Annual Review of Biochemistry, Volume 62, 1993, pages 191-217.

[16] Cf. Brown P.: *Britain dithers over gene therapy*, New Scientist, Volume 136, Issue 1851, 12 December 1992, page 4.

knowledge of an inherited defect can provide opportunities for the use of preventive measures that minimize the morbid effects of a mutation. Second, the improved capacities to define the precise molecular defects causing a disease should markedly advance the understanding of its pathological bases, facilitating the design of rational treatments. Apart from conventional interventions, this will increasingly be the exogenous synthesis and delivery of a missing gene product or the introduction of the normal form of a gene into an affected patient-gene therapy.

While recognizing the progress during the past thirty years, for instance since the discovery of the connection between a well defined chromosomic reality and the Down syndrome, it is fitting to maintain the hope that science and medicine may one day be in a position to overcome the difficulties suffered by individuals in this condition.[17] Disease gene identification and localization, together with the advances in our understanding of the molecular biology of diseases poises gene therapy to provide selective genetic therapies.

In spite of all these hopes, however, and due to mainly technical problems, no patient has yet been completely cured exclusively by gene therapy. Many fetuses have nonetheless been aborted because of the pretext that they had genetic defects. In counseling a patient, the abortion mentality should be considered as a major risk. To the personalistic mind that considers that the fetus is a human individual and thus a human person, no amount of genetic testing, no amount of genetic screening, and no amount of promises of a future genetic cure for humanity, can justify the designed loss of one human being. It's not just a case of numbers, statistics or sociology; it's a case of ethics, of right and wrong. The range of diseases that can be attacked with current methods seems very narrow. There are many drawbacks concerning selectivity, specificity, sensitivity and safety of gene transfer. The major issue for the next several years is how to effectively deliver therapeutic genes so that they can act to cure or ameliorate disease symptoms, and all this obviously has many ethical implications. Before gene therapy can be used as a standard treatment modality without risk, the range of cells that can be targeted for gene insertion must be expanded considerably and might particularly include "stem cells" that divide continually and do not die off over a period of months as

---

[17] Cf. John Paul II: *La Chiesa chiama a promuovere i valori cristiani all'interno delle strutture sociali e sanitarie*, Ai partecipanti a un simposio sulla sindrome di Down, Insegnamenti di Giovanni Paolo II, Libreria Editrice Vaticana, Volume 12, Issue 1, 23 May 1989, number 4.

the type of cells currently used. For many applications it will be necessary to aim the gene insertion at specific organs or tissues. Current approaches to replace "bad" DNA with "good" ones would have to be much more reliable.[18]

4. The problem of genetic information, autonomy and consent

4.1.The limits of informed consent and privacy

The use of predictive medicine, developed thanks to the sequential treatment of the human genome, presents us with the problem of the mature consent needed for genetic research and also of the privacy of those elements that are known as a result and which regard not only one subject, but also his descendants. The communication of data which evidences defects, actual or recessive, is an especially delicate question.[19] One of the basic manifestations of respect of human dignity is the respect of autonomy and consent. Yet there are many laws that restrict individual freedoms for the benefit of society, for example, laws against impaired driving. But are there situations in which the principle of individual autonomy in reproductive choice should be overridden or restricted?[20] One that is already legislated in many countries is the ban on marriage or sexual activity between closely related individuals. This is perhaps not very logical from the genetic point of view. The risk for a significant problem occurring in the offspring of the most closely related, a brother and sister, is about 30%. This is

---

[18] Cf. United States Congress, Office of Technology Assessment: *Pharmaceutical R&D: Costs, Risks and Rewards*, U.S. Government Printing Office, Washington DC, Febuary 1993. The Office of Technological Assessment, of the Congress of the United States, closed on September 29, 1995. During its 23 year history, OTA provided congressional members and committees with authoritative analysis of the complex scientific and technical issues of the late 20th. century.

[19] Cf. John Paul II: *Utilizzare l'embrione come puro oggetto di sperimentazione significa attentare alla dignità della persona e del genere umano*, Udienza al Gruppo di lavoro sul genoma umano promosso dalla Pontificia Accademia delle Scienze, Insegnamenti di Giovanni Paolo II, Libreria Editrice Vaticana, Volume 16, Issue 2, 20 November 1993, number 7.

[20] Cf. Council of Europe, Steering Committee on Bioethics (CDBI): *Draft Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine: Bioethics Convention and Explanatory Report*, Strasbourg, France: Council of Europe, Directorate of Legal Affairs, July 1994, Article 5, which deals with the general rule as regards consent: "An intervention in the health field may only be carried out after the person concerned has given free and informed consent to it. This person shall beforehand be given appropriate information as to the purpose and nature of the intervention as well as on its consequences and risks. The person concerned may freely withdraw consent at any time."

quite similar to the 25% faced by a couple who are both carriers of an autosomal recessive genetic condition. The difference is that in the latter case we would not attempt to influence the couple's decision, even though we are aware of the potential costs to society. Of course there are numerous other reasons why incest is prohibited, but it remains a restriction on individual autonomy.

Another example that is based on guidelines rather than legislation is that of invasive prenatal diagnosis: for example, amniocentesis and chorionic villus sampling, currently offered to women who have an increased risk of having a child with a genetic abnormality. There are three main reasons for restricting access to these tests: the first is because there are risks of inducing miscarriage; the second is economic, in that the tests are expensive and probably not cost-effective to society at lower risk levels; the third involves the individual's intentionality: if a defect is diagnosed, it might or might not eventually lead to selective abortion.

In some countries, like Canada, these tests cannot be bought privately. This certainly restricts the autonomy of younger women who feel that they would like to undergo these tests at any risk level. So, what is the criterion that must be used in order to undergo a test? Is it necessity, or curiosity, a simple preference or choice? What can the counselor say? Many moralists conclude that there should indeed be limits on reproductive freedoms. But how should this be monitored and reviewed? Who monitors and reviews? The medical team? The state? The couple or the individual who undergoes the test? Who monitors and reviews for the fetus? Should this determination be in tune with the dignity of the human person, and above all with the value of life, and can it be backed by the force of law when necessary? Are the criteria to be based on or in tune with societal values, and in some cases backed by the law? Nazi Germany followed precisely these criteria, and we all know what that led to.

So who decides what activities related to genetic counseling should be sustained or prohibited even by law? What should be included? Sex selection for non-medical reasons? Research involving genetic alteration of germ cells or zygotes? Prenatal diagnosis for "susceptibility" genes? Should there be an attempt to enhance normal human traits?

In the present context, respect for persons has two aspects: respecting the autonomy of individuals by requiring their informed consent if they are competent and, if they are incompetent, (as in the case of a fetus, an infant, or a mentally handicapped person), providing

protection by requiring the consent of those who act on their behalf. In a way, most medical staff generally treat patients as incompetent, not out of disrespect, but because they rely on the family to determine the patient's best interests.[21] The patient's "right to know" at times can be in conflict with the medical staff's "discretion not to divulge." Results indicating a genetic abnormality are normally available to the patient or testee, but can the discretion not to divulge this information be invoked by medical staff under any circumstances?[22] Information with regard to gene abnormality may also indicate the future likelihood of certain genetic diseases. To be made aware of such information without recourse to any effective means of prevention or treatment would not be of any use to the testee but might in fact serve only to increase anxiety, confusion and even despair. In this regard, the question is how can the patient's decision to elect not to know be respected? There might be circumstances when a counselor is convinced that the patient has to know for his own good.

---

[21] Cf. Yesley M.S.: *Who Decides?: Bioethics in the United States of America*, in *Human Genome Research and Society: Proceedings of the Second International Bioethics Seminar in Fukui, 20-21 March 1992,* (Editors: Fujiki N.- Macer D.R.J.), Eubios Ethics Institute, Christchurch, New Zealand 1992, pages 34-45.

[22] Michaud J.: *Explanatory Report to the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Amplification of Biology and Medicine,* Directorate of Legal Affairs (for the Council of Europe), Strasbourg, May 1997, numbers 63 to 70, the explanation of Article 10: " Individuals are entitled to know any information collected about their health, if they wish to know... The right to know goes hand in hand with the "right not to know.".. Patients may have their own reasons for not wishing to know about certain aspects of their health... (This) is not regarded as an impediment to the validity of his consent to an intervention; for example, he can validly consent to the removal of a cyst despite not wishing to know its nature... In some circumstances, the right to know or not to know may be restricted in the patient's own interest or... in order to protect the rights of a third party or of society... It is for domestic law, taking account of the social and cultural background, to solve this conflict. Domestic law may justify, where appropriate under judicial control, the doctor in sometimes withholding part of the information or, at all events, disclosing it with circumspection ("therapeutic necessity")... Furthermore, it may be of vital importance for patients to know certain facts about their health, even though they have expressed the wish not to know them. For example, the knowledge that they have a predisposition to a disease might be the only way to enable them to take potentially effective (preventive) measures... (Also, there is a) particular condition when there is a risk not only to that person but also to others. Here too it will be for domestic law to indicate whether the doctor, in the light of the circumstances of the particular case, may make an exception to the right not to know. At the same time, certain facts concerning the health of a person who has expressed a wish not to be told about them may be of special interest to a third party, as in the case of a disease or a particular condition transmittable to others... In any case, the right not to know of the person concerned may be opposed to the interest to be informed of another person and the interests of these two persons should be balanced by internal law."

It is generally accepted that all human beings are free, and, as such, autonomous. Any sort of genetic counseling affects people, legislation limits their autonomy, and especially that of a fetus who is always a ward. So, how directive can genetic counseling really be? How much can legislation do to regulate "genetic problems"? Should the law protect all human lives, including that of a fetus, or has it the right not to protect a fetus in the name of the autonomy of another human being? The public understanding of what genetic counseling involves is sometimes inaccurate, even to the extent that some people are reluctant to attend such counseling. However unjustified, feelings of guilt or shame are common in families that have been afflicted with birth defects or genetic disease. Many people at first expect that they will be told whether or not the counselor thinks they should have children, and where to apportion "blame" if something goes wrong. In fact, as was shown in an international survey of geneticists' attitudes,[23] the truth is quite the reverse. The ethical principle of autonomy has become paramount in genetic counseling, to an extent probably not equaled in other fields of medicine.

Counseling is very often considered as non-directive and non-judgmental: that is to say, it supposedly does not tell people what they should do, nor make value judgments about their choices. The majority of genetic counselors on hand to advise parents during the testing process pride themselves on being "non-directive." They see their goal as providing information and helping patients sort out their feelings. But, can a counselor really be "neutral"?

Frequently there is pressure, especially on the woman, to abort. Numerous counselors use the phrase "non-routine decision" to refer to a couple's choice to discontinue a pregnancy after the diagnosis of a fetal defect, and booklets that some prenatal testing units hand out to couples who have just received a positive diagnosis, treat as inevitable the grief that will accompany the decision to abort a defective fetus, and, by implication, as inevitable the fact that parents will choose to abort. Meanwhile, parents are warned to shield themselves from those who will simply make moral judgments, and the mourning process following an abortion after prenatal diagnosis is carefully likened to the loss of a child through miscarriage or accidental death.

But the fact is that parents are responsible for ending the pregnancy, and their reactions to the decision, and to the abortion itself,

---

[23] Cf. Wertz D.C.- Fletcher J.C. (Editors): *Ethics and Human Genetics: A Cross-Cultural Perspective*. New York: Springer-Verlag, 1989.

are all the more intense for that. The medical community has only recently turned its attention to the emotional issues surrounding abortion in these circumstances, and it seems that the experience is more traumatic than had been expected. So exactly what did the counselor achieve?

The code of ethics of the Canadian College of Medical Geneticists states that supportive rather than directive advice should be given to patients in regard to reproductive decisions. However, the Royal Commission was very "disturbed" when it was discovered that counseling offered by physicians outside genetics centers quite often tended to be paternalistic as well as directive.[24]

Moral dilemmas are also faced by those who provide genetic counseling and these frequently place a profound stress on their autonomy. Since there is such a high frequency today of individuals choosing means to address the problems associated with genetic disorders such as tubal ligations or contraceptives to avoid pregnancy or the choice of abortion to destroy a defective fetus, many Catholics have judged it better to avoid genetic screening and counseling altogether. However, this does not appear to be a legitimate response to the risk of being involved in other people's immoral choices. Besides, in many countries, for example in the United States, where a couple is legally free to obtain an abortion, if the counselor refuses to make the information resulting from genetic tests available out of fear that the persons would make an immoral choice, he could even be accused of hindering their moral choice.

Also, quite frequently, individual privacy rights clash with interests of insurance companies, employers, family members, potential adoption parents and banks to gain information allowing the assessment of financial or personal risks. Moreover, there are cases where fundamental rights and interests of other persons might seriously be affected. This can imply both the safety of others being at high risk on account of professional incapability (for example, a pilot with a high risk of heart failure) or the birth of children with a defect that could be avoided or helped in the case that the information were known to a team of specialists. In the future, it is generally assumed that personal genetic data may be less and less private, making it impossible to keep the results confidential. This may happen either by requests for genetic testing or by persons who provide data on their own genome.

---

[24] Cf. Royal Commission on New Reproductive Technologies: *Proceed with Care*, Minister of Government Services, Ottawa Canada 1993, page 770.

However, some argue that the more tests are available and the more people are tested, the discrimination of carriers is likely to decrease because finally everybody will probably have to be considered in some way as genetically burdened.

Difficulties in the disclosure of genetic information prove that there is likewise a problem with solidarity. Discussion has shifted from confidentiality and privacy towards solidarity.[25] Therefore, an effective legislative approach to avoiding discrimination might probably have to focus more on the use of genetic information as well as its privacy.[26] Most of the major problems in the future will probably imply the exclusion from coverage by insurance companies, the discrimination by employers, or inclusively the discrimination by society or parents who want to unburden themselves of a defective child that costs too much.

Access to private DNA for genetic research is another issue. The question of whether DNA for family studies should be available, whether anonymous or not, is an ever more acute question. In some countries, such as France, blood samples collected for research can only be used for research; no individual data are given out. The situation varies in other European countries. Screening for non-curable diseases is extremely problematic if the resulting information is personalized.

With the right to know the risks and screening for curable and incurable diseases arises the question of when screening should be indicated by the counselor. In cases of incurable genetic diseases without any potential means to improve the situation through early health care, for example Huntington's disease, many think that genetic testing should be left until a time when the person can make his own decision on such a test. A positive result would likely lead to discrimination and a loss of autonomy for the individual. Many institutions all over the world are currently taking such an attitude. However, there are cases where knowledge of the disease can help in rearing a child. In Great Britain, for example, babies are universally screened for two

---

[25] Cf. Chadwick R.F.: *What counts as success in genetic counseling?* Journal of Medical Ethics, March 1993, Volume 19, Issue 1, pages 43-46.

[26] Cf. Council of Europe, Steering Committee on Bioethics (CDBI): *Draft Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine: Bioethics Convention and Explanatory Report*, Strasbourg, France: Council of Europe, Directorate of Legal Affairs, July 1994. Although the right to privacy is clearly stated in Article 10, numbers 1 and 2, number 3 of the same Article allows for "exceptional cases," restrictions which may be placed by law in the exercise of the rights. Such might be a test to identify the author of a crime, or to determine a filiation link.

genetic diseases, phenylketonuria and congenital hypothyroidism, both cases in which appropriate action in the first few weeks of life can prevent serious and permanent damage.

While many pregnant women welcome the choices prenatal testing has given them, others are ambivalent, have misgivings, or simply have not given the matter much thought. Yet the pressures to be tested are powerful. The most obvious pressure comes from the context in which tests are offered. Women who have reservations about screening - and who consider it as a kind of quality control of the fetus - find it difficult to decline tests when their counselors or obstetricians suggest them. In the doctor's office and in the many popular books available on pregnancy and childbirth, there is an assumption that reasonable and enlightened women will naturally want to make use of new screening technologies.[27] Politicians and pollsters have long known that the words "information" and "choice" are powerful ones, especially for women. If there is information to be had, and decisions to be made, the value lies in actively seeking the information and consciously making the decision. To do otherwise is considered to "let things happen to you," or not to "take control of your life." Women who reject screening are often regarded as turning away from the value of choice, and even more profoundly, turning away from the value of information.

Doctors and counselors, however, don't have to live with the anxiety generated by testing and the gathering of information; patients do. Yet physicians and women's health advocates repeatedly insist that the best reason for women to undergo prenatal screening is for "the reassurance it almost always brings." This is a strange assertion. Certainly, worrying is a natural part of any pregnancy. Can my body do all the things necessary to carry the baby to term? Will the baby be healthy? Will I be a good parent? Such free-floating concerns have always plagued women. But in the past few decades, the normal anxieties of pregnancy have been inflamed by a highly specific set of specters, prompted less by genuine health threats than by the promotion of certain tests.

---

[27] The 1983 President's Commission on genetic screening is typical in describing prenatal testing and carrier screening (the testing of couples before conception to determine whether they carry a genetic defect) as enhancing a woman's choices. Genetic screening and counseling are considered as medical procedures that may be chosen by an individual who desires information as an aid in making personal medical and reproductive choices, and where professionals should generally promote and protect patient's choices to undergo genetic screening and counseling.

Because there is a test for Down's syndrome, for example, women over the age of thirty have been bombarded with all sorts of propaganda. It's the same stunt as behind most publicity: women have been trained to concentrate their anxieties on Down's syndrome for the simple reason that they are offered tests for it. But they are offered tests for Down's, not because the risk is personally high for them, but because the public health sector has a powerful interest in reducing the number of citizens who may end up requiring government support.

Displaced anxiety can lead to artificial peace of mind. In the current climate of testing it is all too easy for prospective parents to forget that illness can befall a baby at any time during pregnancy and delivery, or after birth, and that the majority of birth defects are undetectable and unavoidable. Yet, as counselors will be the first to admit, many women who receive a negative result on a prenatal test seem to feel that they are in the clear. This false sense of security can make an undiagnosed birth defect or subsequent childhood illness all the more difficult to handle.

Pressures to undergo testing, at times from the counselors themselves, are frequently followed by subtle pressures to abort in the event of a positive diagnosis. In one study, twenty-five out of forty fetuses so diagnosed were aborted.[28]

Such complex and important decisions cannot be made by anyone other than those who will have to live out the rest of their lives with the consequences. People will often ask for direction, and counselors may have personal bias as to what they might do in a given situation. Some counselors, for instance, are in favor of abortion, and it can therefore be clearly perceived what their counseling may in all probability imply, especially where some genetic defect has been diagnosed.

4.2. Genetic counseling and respect for human dignity

Genetic counseling translates scientific knowledge into practical information and acquaints people about genes and chromosomes, and how these may affect the lives of individuals and those of their offspring. For this reason human genetic research must start to evaluate the services that will be needed at the time when tools for gene therapy are available. Reliable statistics about the frequency of genes, coding for genetic diseases or predisposition to cancers, hypertension

[28] Cf. Kristol E.: *Perfect Picture: The Politics of Prenatal Testing*, First Things, April 1993, Number 32, pages 17-24.

etc. will be necessary to orient public and private research and development in such a way that a maximum number of patients can be helped as soon as possible, when and if therapy becomes available.

A genetic counselor works with a person or family that may be at risk for inherited disease or an abnormal pregnancy outcome, discussing their chances of having children who are affected. It is the genetic counselor who advises whether or not a genetic screening should be performed, and the type of screening which might be more appropriate in a particular case. Thus, providers of genetic counseling should be individuals who have followed a specific educational curriculum and are certified as genetic counselors, and also doctors or nurses with special training in the subject. These health care professionals should be experienced in helping families understand birth defects and how inheritance works, given that they are responsible for providing information that helps families make personal decisions about pregnancy and child care as well as other important issues.

In order for the patient or the family to make the decision, three elements of informed consent are necessary: sufficient information must be provided, the information must be understood by the patient or family, and the decision whether to participate in the research or course of treatment must be voluntary. This is where the use of genetic counseling becomes clear. The fundamental guideline in genetic counseling must be the welfare of each and every human individual, and never only that of one individual's autonomy in prejudice of another. The principle of beneficence may also be expressed as the traditional "do no harm" as well as a utilitarian ethic of maximizing possible benefits and minimizing possible harms. Justice also requires fairness in the distribution of health care and in the selection of research subjects. Those who share the risks should also share the benefits. However, although the principle of autonomy is very important, it is not absolute, since at some points conflict between collective rights and individual rights may arise. When they do, the limits must be set in reference to the nature and the dignity of the human person, and not only to societal norms and conventions.

Included in the right to be informed, is the right not to be informed.[29] Gene diagnosis and therapy is beginning to enter the practi-

---

[29] Cf. Council of Europe, Steering Committee on Bioethics (CDBI): *Draft Convention for the Protection of Human Rights and Dignity of the Human Being with Regard to the Application of Biology and Medicine: Bioethics Convention and Explanatory Report*, Strasbourg, France: Council of Europe, Directorate of Legal Affairs, July 1994, Chapter 3 on Private life and right to information, Article 10, Private life and right to information, numbers

cal application stage. Because the genes contain the entire genetic code including information as to the likelihood of future development of genetic diseases, society must solve the ethical problems involving the individual's privacy of the acquired information prior to the practical application of these techniques. It should emphasize strongly in this regard - and clearly legislate - that information referring to genetic abnormality is essentially the property of the patient-testee and not of the medical authorities, and that protection of human rights should be of primordial importance.

At the time of genetic testing the medical staff should give sufficient information about the tests and what information is likely to come to light as a result, and the testee should be able to invoke the "right to know" or the "right to not know"; with few exceptions the medical staff does not have the right to withhold information. Further, to prevent any unauthorized access to genetic information, it is necessary to have a confidential information management system independent of the general information system in the hospital or clinic. As for gene therapy, in addition to the central evaluation system and in-hospital ethics committees, it is desirable to have a practical in-hospital monitoring and guidance system. In addition, because of the serious ethical problems in the application of prenatal or pregestational gene diagnosis and the handling of these results, it is imperative to promote gene diagnosis and therapy that is not only effective but also ethical.[30]

As regards the right to know and the right not to know, it would be necessary to prepare a written informed consent for signature at the time of genetic testing, except in the case of most infectious diseases and perhaps in some cancers. The medical staff in charge should explain the purposes of the test and what will become known as a result. They should be required to have enough knowledge to be competent in explaining these specifics in detail. Thus the testee at this time could choose whether to undergo the test or not, and also whether to be informed of the test results or not.

---

1 and 2 state: "1. Everyone has the right to respect for private life in relation to information about his or her health. 2. Everyone is entitled to know any information collected about his or her health. However, the wishes of individuals not to be so informed shall be observed."

[30] Cf. John Paul II: *Il progresso scientifico non può prescindere dalla dignità del trascendente destino dell'uomo*, Ai partecipanti al convegno del «Movimento per la vita», Insegnamenti di Giovanni Paolo II, Libreria Editrice Vaticana, Volume 5, Issue 3, 3 Dicembre 1982, number 2.

It should be strictly prohibited - as contrary to the patient's right of privacy - for the medical authorities to perform genetic test simply because of their perceived need to know. However, information concerning the HIV, HTLV, HCV and MRSA status of hospitalized patients is very important in protecting the health of the medical staff and in preventing intra-hospital transmission. These particular diseases can be tested for by microbiology also, but likewise in this case medical facilities should not perform these tests without notifying the patient or without obtaining his or her informed consent. Patients should be given the right to refuse; if the patient invokes this right, the medical staff can always suppose that the patient is indeed infected, and inform him or her that he or she will be treated accordingly. Of course, the necessity for patient management in this fashion would be quite expensive but unavoidable.

As regards the nature of genetic counseling, it is difficult to perceive how it can be neutral or non-directive. An offer of prenatal diagnosis, for example, implies a recommendation to accept that offer, which in turn might entail a tacit recommendation to terminate a pregnancy if it is found to show any abnormality, or to go ahead with the pregnancy in spite of what is revealed. This sequence can be present irrespective of the counselor's wishes, thoughts, or feelings, because it also arises from the social context as well as from the personalities involved. Within much medical literature there is a clear assumption that counselors are there, in effect, to help patients through the difficult process of agreeing to be tested and agreeing to abort in the event of a diagnosed defect. This is certainly not a neutral position.

The objective of genetic counseling is not only to inform but also to fully educate individuals or couples about the nature and extent of the genetic risks they face, together with the possible options open to them.[31] To educate also implies a moral education together with the

---

[31] Cf. Michaud J.: *Explanatory Report to the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Amplification of Biology and Medicine,* Directorate of Legal Affairs (for the Council of Europe), Strasbourg, May 1997, numbers 34 to 40, explanation of Article 5: "The patient's consent is considered to be free and informed if it is given on the basis of objective information from the responsible health care professional as to the nature and the potential consequences of the planned intervention or of its alternatives, in the absence of any pressure from anyone... In order for their consent to be valid the persons in question must have been informed about the relevant facts regarding the intervention being contemplated. This information must include the purpose, nature and consequences of the intervention and the risks involved. Information on the risks involved in the intervention or in alternative courses of action must cover not only the risks inherent in the type of intervention contemplated, but also any risks related to the individual characteristics of each patient, such as age or the existence of other pathologies... Moreover, this information

complete knowledge of the facts, indications and consequences; it is not merely general information that entails the nature and the extent of the genetic risks involved, together with the possible options open to the costs and the benefits, and even, perhaps, of selective abortion if things go wrong.

The final informed choice may be made solely by the individual, but the counselor will have given an authoritative recommendation that is not indifferent. Except in the case of certain disabilities,[32] the final informed choice is ultimately made by the individual, but the counselor obviously influences the choice made through recommendations that are explicit or implicit. There can be no neutrality. Even silence speaks.

Certainly reproduction is one of the most personal and spiritual of human activities, but even though a conscious decision to have children rests on many subjective factors including the relationship of the partners, their individual and combined desire for children, and their economic and social well-being, the decision also implies objective factors that transcend the individual decisions or those of the couple, or even those of society as a whole. The life of the fetus, for instance, transcends the decision-making of the couple. The individuals concerned must weigh these factors as well as the possible genetic risks or benefits. So there can be a "right" answer, and not only a "right" that may seem right for each individual involved. The moral duty is to

---

must be sufficiently clear and suitably worded for the person who is to undergo the intervention. The patient must be put in a position, through the use of terms he or she can understand, to weigh up the necessity or usefulness - of the aim and methods of the intervention against its risks and the discomfort or pain it will cause... Consent may take various forms. It may be express or implied. Express consent may be either verbal or written... Freedom of consent implies that consent may be withdrawn at any time and that the decision of the person concerned shall be respected once he or she has been fully informed of the consequences... Information is the patient's right, but ... the patient's possible wish not to be informed must be observed. This does not, however, obviate the need to seek consent to the intervention proposed to the patient."

[32] Cf. Ibid., numbers 41 to 49, explanation of Article 6: "Some individuals may not be able to give full and valid consent to an intervention due to either their age (minors) or their mental incapacity. It is therefore necessary to specify the conditions under which an intervention may be carried out on these people in order to ensure their protection." In any case, the purpose should always be to protect persons who are not able to give their consent; such are the cases of accidents, states of coma, reduced mental capacity etc. The intervention should always be for the direct benefit of the person. In the case of minors, Michaud continues: "In certain situations which take account of the nature and seriousness of the intervention as well as the minor's age and ability to understand, the minor's opinion should increasingly carry more weight in the final decision. This could even lead to the conclusion that the consent of a minor should be necessary, or at least sufficient for some interventions."

search for that right answer - the truth - and abide by it in practice. If this were not so, it would signify the denial of any objective truth, not only moral but also scientific. If there can be wrong answers, there can also be right answers, and these are the answers that counselors in particular are bound in conscience to give. This can only be done if the counselors themselves have a truthful, truth-seeking, well-informed conscience.

Authentic counseling seeks the good of all patients and especially those who are weakest. Through their specific action counselors enlightened with an anthropology that is coherent with the Christian vision of the person, of the couple and of sexuality, will be capable of giving a priceless contribution that can help people discover the meaning of love and of life, and of sustaining families in their mission as "sanctuaries of life."[33] And where the criteria of the person seeking counseling is not in conformity with objective good, for example, when he or she seeks to harm or destroy life, it is surely the counselors duty to clearly inform concerning the integral truth of what is involved, and the responsibility that all human beings have to justly use their free will in favor of life and good of themselves and others, especially if the latter are wards entrusted to their care and responsibility. Practically speaking, all this requires a deep cultural change rooted in the formation of moral conscience regarding the immense and inviolable value of every human life.[34] For this reason it is so important that the link between life and liberty be rediscovered. These are indivisible goods: where one is harmed, so too is the other. As John Paul II asserts, where life is not cherished and loved, there can be no liberty; nor can there be plenitude of life without liberty.[35]

Genetic counselors cannot always be held morally responsible for the moral judgments of those whom they counsel. But it certainly would not violate the standards of their profession if they indicated to those who seek their counsel their own convictions about the immorality of abortion. However, in the present state of the art in some countries, if they tried to interfere with the decision of those whom they were counseling they could well lose their license and could, according to some forms of legislation, be prosecuted. Of course the patient must be protected, which is the supposed finality of such legislation,

---

[33] Cf. John Paul II: *Evangelium Vitae*, Acta Apostolicae Sedis, Libreria Editrice Vaticana, Città del Vaticano, 2 May 1995, number 88.

[34] Cf. Ibid., number 96.

[35] Cf. Ibid., number 96.

but it must be remembered that the counselor too is a person with a conscience that ought to be respected.

Therapeutic interventions are probably the least problematic of gene manipulations. In 1983 Pope John Paul II addressed the World Medical Association Convention and said that "strictly therapeutic intervention" can be considered in principle as desirable.[36] Accepted professional and moral norms would have to be applied in such situations: the informed consent of the patient or his proxy if he were incompetent, the maintaining of confidentiality, a weighing of the proportion of risk and benefit for the patient, the avoidance of any procedure which would do direct harm to the patient without carrying any therapeutic benefit. That last consideration must be mentioned since there are actually those who advocate the direct killing of those suffering from a genetic disorder for the victim's benefit. The positive ethical qualification of any research must result in the ethical guarantees offered by the experiments as regards both the risk factors and the necessary consent of the person involved; this should also be extended to the applications of the discoveries and of the results.[37]

Many suggest that genetic tests which would not be beneficial for the individual in all cases, should not be carried out.[38] This especially holds true when that individual is a fetus which has no way of defending itself from aggression. Furthermore, there is broad support for the position that individual genetic information should always remain private, including the genetic information of a fetus, unless there is a certainty that a real benefit can be attained or an effective intervention

---

[36] John Paul II: *Arbitraria e ingiusta la manipolazione genetica*, All'associazione medica mondiale, Insegnamenti di Giovanni Paolo II, Libreria Editrice Vaticana, Volume 6, Issue 2, 29 October 1983, pages 917-923.

[37] Cf. John Paul II: *Non possiamo nascondere il pericolo che la scienza subisca la tentazione del potere demiurgico, dell'interesse economico e delle ideologie utilitariste*, Udienza: Ai membri della Pontificia Accademia per la Vita riuniti in Assemblea Generale, 20 Novembre 1995, in La Traccia, Number 11, 1995, number 4.

[38] Cf. Michaud J.: *Explanatory Report to the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Amplification of Biology and Medicine*, Directorate of Legal Affairs (for the Council of Europe), Strasbourg, May 1997, in numbers 80, the explanation of Article 12: "Tests which are predictive of certain genetic diseases may offer considerable benefits to an individual's health by allowing timely preventive treatment to be instituted or by offering opportunities to diminish the risks through modifications in behavior, lifestyle or environment. This, however, is not possible at present in many genetically determined disorders. The right to know as well as the right not to know and proper informed consent are, therefore, of particular importance in this field since problems may clearly arise for the individual arising from tests predictive of genetic disease for which there is currently no effective treatment."

can be carried out in his or her favor to improve a genetically defective condition.[39]

In cases where privacy rights clash with a social right to private information, this ought to be made known only to those who can do something to remedy the situation and who can objectively insure the favor of the sick individual, and never to the general public, which includes insurance companies and companies that seek profits on such knowledge and at the individual's expense.[40]

People will often ask for direction, and counselors may have personal bias as to what they might do in a given situation. But the decision to deprive a human being of his or her very existence cannot be made by anyone, not even by those who will have to live out the consequences the rest of their lives. The temptation that inclines towards a decision that kills an innocent person must be resisted since, in effect, the responsibility to protect life and health is the duty that must be ensured by all, but particularly by those responsible for helping others to make personal decisions, namely, counselors. Society tends all too often to quantify risk solely in economic terms, more than individuals do when assessing risk for themselves. While it is tempting to hope that genetic counseling might have some effect in reducing the incidence, costs and suffering due to genetic disease, this can never be a primary goal. The primary goal must always be the integral welfare of each and every human being, regardless of his of her actual state of health or social condition. Any other type of counseling would be discrimination.

---

[39] Cf. UNESCO, International Bioethics Committee: *Preliminary draft of a Universal Declaration on the Human Genome and Human Rights*, Politics and the Life Sciences, Volume 15, Issue 2, September 1996, number 8 which states: "Genetic data associated with a named person and stored or processed for the purposed of research or any other purpose must be held confidential and protected against disclosure to third parties."

[40] Cf. Michaud J.: *Explanatory Report to the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Amplification of Biology and Medicine*, Directorate of Legal Affairs (for the Council of Europe), Strasbourg, May 1997, numbers 135 and 136, the explanation of Article 22 concerning the disposal of a removed part of the human body: "Much information on the individual may be derived from any part of his body, however small (eg. blood, hair, bone, skin, organ). Even when the sample is anonymous the analysis may yield information about identity... Parts of the body which have been removed during an intervention for a specified purpose must not be stored or used for a different purpose unless the relevant conditions governing information and consent have been observed."

Sommario: *L'articolo si suddivide in due parti. Nella prima, viene affrontata la questione dell'embrione, della sua ricerca e legislazione: vengono affermati con forza la dignità personale dell'embione umano e, di conseguenza, i suoi diritti umani. Dopo avere affrontato il tema della dignità personale dell'embrione umano, nella seconda parte l'autore prende in considerazione la validità morale sia della diagnosi genetica prima dell'annidamento, sia della diagnosi genetica prenatale.*

Parole chiave: Statuto dell'embrione, diagnosi genetica prima dell'annidamento, diagnosi genetica prenatale, test genetico

Key words: Embryo status, genetic testing, preimplantory genetic diagnosis, prenatal genetic diagnosis