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Genetic Counselling

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This report on genetic counselling and its bioethical implications, is a natural extension of the 1994 Reports on genetic screening and testing, and on gene therapy[1, 2], because it addresses the impact that these fields of science have on the individual person (born or to-be-born) and sometimes on Society.

The increasing availability of genetic tests confers to the speciality of genetic counselling a fast growing place in medical practice. Genetic counselling provides the link between genetic technologies, several of which have been acquired through the Human Genome Project, and patient care. It can be defined as a communication process which involves diagnosis, explanation and options (Section II.1). This Report examines first the scope and practice of genetic counselling today, then bioethical issues related to scientific questions, to welfare of the persons receiving counselling (counsellees or consultands) and finally to moral issues for society at large.

I. The Scope of Genetic Counselling

The genetic tests listed in the 1994 Report[1] of the International Bioethics Committee of UNESCO (IBC) to which several new DNA tests can be added in 1995, indicate that it is possible:

- to establish a diagnosis of hereditary or congenital diseases in affected patients, with accrued certainty and precision (e.g. mutation involved);
- to predict the probability of development of a disease in individuals or families not yet affected (e.g. late-onset and susceptibility genes, carrier state);
- to take measures to alleviate the clinical expression of such disease, to decrease the risk of its development and possibly prevent it (e.g. monitoring, early clinical diagnosis, somatic gene therapy);
- to predict birth of an offspring with a genetic disease and allow decisions on the fate of the fetus (measures to take at birth, voluntary interruption of pregnancy);
- to offer means for avoiding conception or implantation of embryos with genetic diseases (e.g. pre-marital counselling, selective implantation, sterilization).

Genetic testing should as far as possible be accompanied by genetic counselling. The scope of genetic counselling is to communicate information and options of the above nature to patients, parents or family. This activity is to be considered as a medical act concerning a diseased condition for which a patient (including the mother of an unborn child) or a family seek help from a medical professional. It ought also to provide support in reaching decisions about options.

Considering that genetic testing will increase as new gene probes are added for the 3,000 known genetic diseases (may be 5% of all human genes), and that there could be a tendency for genetic screening, the extent of counselling may, however, progressively widen to:

- families with histories of diseases (e.g. cancers, miscarriages, psychiatric conditions);
- populations at risk due to reproductive age, environment (exposure to radiations or mutagens, lifestyle) or geographical considerations (areas with high prevalence of genetic disease, such as β-thalassemia in Cyprus, Sardinia, or such as Tay-Sachs disease in Jews of Eastern Europe origin);
- population at large (screening programs, "supermarket" genetic testing), with all the concern this may raise and which need to be weighed against health benefits.

While in many aspects genetic counselling is in the scope of medical professional practice (a predictive medicine), there are many issues which need to be regarded as being rather in the realm of scientific research since the knowledge about genes implicated in the aetiology of human diseases, about the limits of their normal variability and polymorphism,

about their interaction with environmental factors and with other genes, is still evolving. This ought to be reflected in the process of information communication which is part of genetic counselling. This process is delicate and requires sophistication in informing counsellees (Section III).

The way the practical options open to the counsellees are communicated are a most important part of genetic counselling and often touches upon moral, social or traditional preconceptions and stresses the ethical framework of counselling. It ought to convey the beneficial aspects of genetic testing to the counsellees: better assessment of genetic risk which often reassures, possibilities of being prepared or intervening, safer potential for healthy reproduction (Section IV). However, the moral questions, the socio-political concerns and pitfalls, should be constant preoccupation for genetic counsellors (Section V).

II. The Practice of Genetic Counselling: A Survey

This part of the Report is based on answers to a questionnaire aimed to obtain a current view of how genetic counselling is being done in different countries. Answers to the questionnaire were provided by members of IBC and leading professionals. (*United States of America*: Dr A. Robinson, National Jewish Center for Immunology and Respiratory Medicine, Denver, Colorado; Prof. V.B. Penchaszadeh, Medical Genetics, Beth Israel Medical Center, New York, for the Latino-Americans example; *United Kingdom*: Dr D. Shapiro, Nuffield Council on Bioethics; *Italy*: Prof. A. Bompiani, Italian Society of Medical Bioethics, National Committee of Bioethics; *Chile*: Dr R. Cruz-Coke, Unit of Genetics, J.J. Aguirre Hospital, University of Chile; *Mexico*: Dr R. Lisker Y.; *Argentina*: Dr L. Vidal-Rioja, IMBICE; *Japan*: Dr D. Macer, Institute of Biological Sciences, University of Tsukuba; *Israel*: Dr R. Carmi, Genetic Service, Soroka Hospital, Ben Gurion University, Beer-Sheba; *China*: Dr Qiu Renzong, Bioethics, Chinese Academy of Social Sciences, Beijing; *Zaire*: Rev. J.M. Mpendawatu, Vatican.) To highlight the opinions, excerpts of the answers are given while clearly the whole documents should be consulted.

II.1 Definitions of Genetic Counselling

The definitions given concur that it is a communication of information about diagnosed genetic conditions, in a way which allows to make a decision, as autonomous as possible, and safeguarding the emotional and ethical character of the person who asks for the consultation. While defined as based on a physician-patient relationship in many countries, the complexity of genetic counselling has led to a new profession of genetic counsellors who are not physicians, especially in North-America.

- UNITED STATES OF AMERICA:

A communication process which involves diagnosis, explanations and options (as in all medical consultation). In genetic counselling there is a stronger need for detail, especially in the explanations and options, for which empathetic and emotional support are an essential part. Counsellors are involved in the ethics of the "people's right to know".

- UNITED KINGDOM:

Counselling entails precision of diagnosis, the estimation of risks, and a supportive role to ensure that those who are given information are enable to benefit from it and from the interventions that are available.

- ITALY:

The objective, methods and indications of genetic consultation are:

Objective: to provide information to patients (and/or blood relations of a patient) at risk of contracting a disease that may be hereditary on:

- consequence of pathology in question
- probability of contracting and transmitting it
- possibility of keeping it in check and treating it

Methods: - construction and analysis of pedigree
- calculation of the risk of recurrence (Mendelian or empirical)
- estimation of the consanguinity coefficient
- more specific analysis

When is counselling indicated:

- known or presumed illness in patient or family
- congenital malformation
- mental retardation
- consanguinity
- recurrent miscarriage, infertility (more in Section II.3).

• CHILE:

A medical process of communication between a physician and a consultand (counsellee) where scientific knowledge, data and facts are exchanged in order to provide a framework to understand the genetic problem of the patient and the family.

• ARGENTINA:

Better called "genetic advising" - a useful tool in preventive medicine.

• ZAIRE:

Information on eventual pathology, not therapeutic but predictive.

II.2 Who does genetic counselling? Roughly how many counsellors are there? What are their professional qualifications? Are they licensed or certified in some other way?

Differences are apparent between countries in which genetic counselling is done by specialized physicians, or by primary care providers with more or less certification and training in medical genetics, and countries in which there is in addition a professional body of genetic counsellors who are not physicians. Psychologists and ethicists may be involved in some countries. The growing demand for genetic counsellors of all types is obvious in all countries. When given, the number of counsellors per million inhabitants vary: 6-4 (Israel, United States of America, Italy), 2.9 (United Kingdom), 1.1 (Chile), 0.3 (China).

• UNITED STATES OF AMERICA:

Several groups of qualified professionals:

- Medical geneticists: MDs, often pediatricians
- PhDs in medical genetics
- Counsellors: MSc level
- Some counselling by primary care physicians, such as obstetricians, some by nurses and social workers (these being not very well qualified at present).

The majority of well-trained and qualified counsellors (first three groups) work in clinics approved and certified by the American Board of Medical Genetics. Most professional counselling in the United States is performed by MSc level genetic counsellors, who have had 2 years of training in medical genetics after graduating from college. The training program must be certified by the American Board of Genetic Counselling (ABGC). The program includes all aspects of medical and human genetics, and the students must be supervised by board-certified clinical geneticists. They must present records of 50 cases that they have counselled. The course work includes: clinical genetics, molecular genetics, cytogenetics, counselling techniques, psychosocial factors influencing counselling, ethics and population genetics. To be qualified, genetic counsellors must pass an examination prepared by the ABGC. MSc level counsellors work under the supervision of a doctoral level medical geneticist, under whose name charges are made.

There are currently about 950 such qualified genetic counsellors in the United States and 60 training programs. There is a professional organization, the National Society of Genetic Counsellors (NSCG), which publishes a journal, the *Journal of Genetic Counselling*, and has an active continuing education program. The NSCG has a Code of Ethics to which all counsellor are asked to adhere, which stresses confidentiality, autonomy and privacy.

The demand for qualified genetic counsellors exceeds the available supply. The demand will increase more due to the Human Genome Project and to the current stress on identifying those at risk of genetic disease.

Due to the demand, the primary care providers will have to offer themselves commonly needed genetic services. Increasingly, several primary care physicians are sharing a qualified genetic counsellor, as part of their organization. Primary care physicians, in general, have insufficient knowledge of basic human genetics and medical genetics. A small number of nurses, especially those in maternal and child health, have been certified by ABMG, and there is a Society of Nurses in Genetics.

Suggestions for the future are:

- teach more medical genetics in Medical Schools;
- have more genetic counsellors;
- teach genetics in nursing schools.

The future will require of all who do genetic counselling a much more sophisticated knowledge of molecular genetics (to be aware of the various research laboratories that specialize in tests for specific diseases), so that they can understand and relate to the counsellee what is involved and obtain their informed consent. The special needs for ethnic groups in the United States are referred to in Section II.6.

• UNITED KINGDOM:

In 1990, there were 125 medical staff in British genetic centres: 48 consultant clinical geneticists, the other junior or other medical staff. There are non-medical genetic registrars (about 40) with educational approval by the Joint Committee on Higher Medical Training, who keep contact with high risk families. There were another 80 genetic co-workers including genetic nurses. A two-week course is offered by the Institute of Child Health, followed by a 6-month project work. Training of general practitioner and nurses is promoted by the voluntary Genetic Interest Group (GIG).

Most individuals and couples known to be at high risk are counselled by specialists according to recognized ethical and professional principles. This should be extended to community-based population-screening services. In the future, medical genetics should be part of the core education curriculum of doctors and nurses; screening and counselling should start at the level of primary health care, e.g. in family planning clinics; it may be integrated into general obstetric care, with help of obstetricians who can provide proper support for prenatal counselling, every unit having at least one midwife trained in genetic counselling. Simple genetic information could be incorporated into school curriculum.

• ITALY:

The genetic counsellor's (consultant) function is classified as a health service. There are no special regulations. There may be conventions (agreements) between hospitals and universities or research centers (to perform genetic tests).

There are 54 public centers affiliated to the Italian Society of Medical Genetics (ISMG), with about 3-5 counsellors by center (150-250 in total). There is an unestimated number of private centers. A center may comprise 3-5 professionals, typically:

- a medical geneticist or doctor with specialization in medical genetics;
- 1-2 biologists in cytogenetics and/or molecular biology;
- a psychologist;
- an ethicist.

There are also 1,500 free public family consulting rooms (dealing mainly with contraception) which should eventually be authorized to provide genetic counselling service, if only in the form of guidance.

The professional qualification of counsellors varies: generally there are heads of university centers or services for medical or human genetics. The vast majority would be MDs, teachers or specialists in human/medical genetics. Doctors and biologists have an initial post-graduate qualification in specialization schools in medical genetics, human genetics or cytogenetics. In order to acquire expertise in genetic counselling, it is necessary to complete a period of training, which has not yet been made official.

- CHILE:

Only physicians (medical geneticists and primary care physicians trained in medical genetics) are responsible for this specialized medical service. They have legal and ethical responsibilities.

- MEXICO:

Formal genetic counselling is performed by trained physicians who number around 100. Many counsellors have at least one year of previous training in Pediatrics. Most counsellors take a 2-year graduate course in medical genetics, which are available in several of the larger cities in Mexico. The counsellors are certified by a National Board of Medical Genetics.

Other physicians involved in management of patients with genetic diseases are obstetricians, pediatricians, surgeons; they may lack proper genetic training, but have the trust of families and answer questions which are in the area of genetic counselling. It has been difficult to regulate proper training.

- ARGENTINA:

Genetic advising is ordinarily performed by physicians (MDs) with medical genetic qualification. Since teaching of this specialization in Medical Schools is rare, most present geneticists got training in North-America or Europe. Their background and expertise were then examined by a board from the Argentine Society of Genetics, which awards the title of specialists. Recently, courses and residency in medical genetics were implemented at the National Institute of Medical Genetics, including theoretical and practical aspects of counselling.

Thus, genetic counselling is included in the patient care by medical geneticists and does not require other licensing or certification than that of medical genetics.

The number of services for clinical genetics, cytogenetics and associated areas, is not large and they are concentrated in big cities. The demand in distant places exceeds the supply. Plans by the State to increase care services for medical genetics, forensic and genome diversity, as well as teaching programs in genetics, are far from being implemented.

- JAPAN:

Genetic counselling is done by clinical physicians, especially obstetricians and gynecologists (OB&G), pediatricians. Of the 80 Medical Schools, only a handful have professors of genetics. There are no genetic counselling courses, but may be in the near future.

- ISRAEL:

Genetic counselling is done by both medical geneticists (MDs) and genetic counsellors (MSc, a few PhDs), the latter engaged in more "routine" cytogenetic tests. Medical genetics is a subspecialty requiring board certification in pediatrics, OB&G or internal medicine plus 2-3 years of residency in genetics followed by board certification examination. Genetic counsellors are not yet formally a profession but require an MSc in biology plus 2 years field experience in a recognized genetic institute before licensing by the Ministry of Health as clinical geneticists in genetic counselling. There are about 30 counsellors in Israel (for 5.3 millions) about half of them MDs. About 10% may be in private medical centers.

- CHINA:

Geneticists work in general hospitals (internal medicine, OB&G, pediatrics, psychiatric departments), institutes of maternal and child care, institutes of science and technology in family planning. They have a diploma of medical school, have been trained in genetics in medical schools in China, but mainly abroad. No formal course of medical genetics even in Peking, no special licensing.

About 400 geneticists work in China (1.2 billions), a tiny number.

- ZAIRE:

Absence of qualification or certification, which is urgently needed. The young generations should be sensitized through genetic education even in high-schools.

II.3 By what means do individuals or couples get <u>access</u> to genetic counselling? Is it government sponsored and/or undertaken by voluntary organizations? Is it associated only with research hospitals? Is reimbursement covered as part of the national medical treatment?

In most countries, access is by referral from primary care physicians. According to the general medical system of a country (state or private medicine), genetic counselling can be free, or partially reimbursed or dependent on the private health insurance of the patient. The need for making genetic counselling available to the needy (especially aliens, immigrants), or of having special programs, has been recognized by some countries. Some promote genetic screening and counselling at the national level.

- UNITED STATES OF AMERICA:

Couples have access to genetic counselling, in general, through referral by primary care physicians or by self-referral. Several primary care physicians may share the service of a qualified genetic counsellor. Some counselling is done in speciality clinics such as for neurofibromatosis, neurology, hemoglobinopathies, or for prenatal diagnosis.

- UNITED KINGDOM:

Screening and repeated counselling is encouraged actively, at the level of primary health care, including at preconception, i.e. these services should become part of family planning. Educational material should be provided by a national genetic information centre. A National Confidential Inquiry into genetic disorders done on behalf of the Department of Health (as part of its clinical audit programme) encourages greater awareness and understanding of the significance of genetics in clinical practice, beyond specialized clinical genetics.

- ITALY:

Access usually through general practitioner or specialist (OB&G, pediatrics), but also direct access on basis of public or medical information. There are 1,500 State-supported Family Consulting Rooms, dealing with family planning (e.g. contraception), which should become officially authorized to provide genetic consultation. There are special programs at regional levels (e.g. diagnosis of β-thalassemia, in Sardinia and Latium).

<u>Reimbursement</u>. It is possible to apply to the local health unit to cover expenses of genetic consultations, but there remains a problem with illegal immigrants who have no health coverage. Consultations can be in university/institute clinics, in public or private research centers. Consulting is often free of charges but laboratory analyses have to be paid and only in some cases there is partial reimbursement by the National Social Assistance Service. The State could intervene more effectively through the family consulting rooms which are free, and through programs in regions with prevalent genetic diseases such as β-thalassemia.

- CHILE:

Access through referral by physicians or self-referral. Genetic counselling is a regular medical service within hospitals and clinics where patients come to consult on their genetic problems.

- MEXICO:

Access is through government hospitals (not necessarily research hospitals) and private medicine. Referral to the geneticist-physician by other physicians who believe their patients need counselling.

- ARGENTINA:

Genetic advising is included in the care role of medical geneticists. There are no fees in hospital services sponsored by the government (State administrative departments or public universities). There is reimbursement for genetic and counselling expenses at semi-public and private centers, which is awarded by the health assistance system subscribed by patients.

- JAPAN:

Access is through compulsory health insurance, and patients can go directly to public or private clinics. Access to some university hospitals is easier by referral. Prenatal diagnosis is not directly covered. Some local city or prefecture taxes, that cover maternity costs, may be used to defer costs to the clients. But this is not part of the National health insurance (childbirth is not viewed as a "disease").

- ISRAEL:

Access is by referral from hospital pediatrics, neonatology wards, outpatient clinics as well as community "mother and child health" clinics. Many visits are by self-referral. Genetic counselling is in all major hospitals, which are all university-affiliated, but also some private medical centers or health insurance clinics. Cost is covered by the National Medical Insurance, although reimbursement of laboratory tests varies and is a matter of discussion.

- CHINA:

Only hospitals or institutes sponsored by the Ministry of Health, or bureaux for health of municipal/provincial governments, provide genetic counselling, which is free. But if the clinic is not the one, which has a contract with the working unit of the patient, payment is required. A reform of the health care program proposes that all clients pay a small part of the costs.

- ZAIRE:

Genetic counselling is not reimbursed by social security although some tests, e.g. karyotyping, are reimbursed.

II.4 <u>When</u> does genetic counselling characteristically get done? Is counselling directed to potential parents, couples, individuals? Are there limits on seeking genetic information about children at a time when no treatment is possible?

In most countries, counselling seems to be addressed to every situation when a genetic disease is diagnosed or even suspected. Most commonly, to couples who have an affected child and in the framework of prenatal testing. Pre-marital counselling, for consanguinity or for regional or ethnic reasons, is practised. Counselling to individuals with late onset diseases may be more delicate. Some people may not want to know of the probability of an untreatable disease. However, there seems to be no limitation on seeking information about children for untreatable disease, and the importance of clarifying who is a carrier of an untreatable genetic disease, for better research on such diseases and for later reference if a treatment becomes available, is recognized. Genetic information may often help in correctly labelling a disease, for example in cases of mental retardation or neurological, psychiatric disorders, and counselling is then important. Screening of newborns for genetic disease is still ill-defined in its extent and scope.

• UNITED STATES OF AMERICA:

Frequently, to couples with previous child having birth defect or mental retardation. Also, in cases of familial history of cancers, heart disease or psychiatric disorders. Other indications are: recurrent miscarriage, infertility, family history of genetic problems, or pregnancy over the age of 35 years. Also known, exposure to teratogenic drugs.

Screened newborns identified as affected by a genetic disease should receive counselling by genetic counsellors. This because the carrier state should be clarified (independently of treatment). This applies also to ethnic groups with an increased incidence of specific genetic diseases. Some genetic counselling is done to patients in speciality clinics (Section II.3 above).

• UNITED KINGDOM:

Genetic risks should be known and counselling given before a pregnancy occurs so that testing can be offered as a planned procedure (preconception stage, i.e. family planning). Prenatal diagnosis is the approach chosen by most informed couples at risk for severe genetic disease. Genetic counselling for prenatal diagnosis should be given as early as possible during pregnancy. It is essential for counsellors to keep in touch with couples and inform them of new medical developments, so that the high-risk couples can decide to use testing (e.g. chorionic villus sampling or DNA testing) for having healthy children. Community-based mass-screening can involve every pregnant woman in screening for fetal abnormalities, and the need for counselling is already large and will increase.

Prenatal diagnosis encourages reproducing: when test is not available, about half couples at risk counselled opt to stop reproducing. When offered prenatal diagnosis, 98% of at risk couples (e.g. thalassemia) use it to achieve a healthy family.

• ITALY:

Genetic consultation targets both individuals and couples or parents. The point of departure is a precise diagnosis set out in writing by a physician or a health unit. It is of fundamental importance to ascertain the parent's reason for requesting counselling, to ascertain their expectations which sometimes may vary between partners.

No restrictions are applied to the quest for genetic information concerning persons, mainly children, affected by incurable pathologies. With the consent of the person concerned or the person holding the necessary authority, the collection of data is essential in order to acquire greater knowledge of case surveyed and to further studies on the genetic/molecular basis of a disease. The ultimate aim is to improve therapeutic prospects.

• CHILE:

Counselling to individuals, potential parents, close relatives, couples and individuals with consanguinity risks. In fact, to every person and members of their family. The most frequent indications are:

- birth defects, recurrent miscarriages, infertility;
- prenatal diagnosis, teratogenic drug exposure, paternity testing;
- mental retardation, psychiatric disorders, sexual deviations.

Newborns are screened only for phenylketonuria (PKU) and hypothyroidism in urban areas.

• MEXICO:

Counselling is done mostly after the birth of a child with genetic disease or malformation. It is usually done in the outpatient clinic. Pregnant women afraid of having an affected baby, or consanguineous couples planning to marry, are counselled.

Newborn screening and parent counselling is done in a limited way, only for PKU and hypothyroidism. For diseases without treatment, most people would not want to obtain information.

- ARGENTINA:

Consensus is that counselling must be provided once the illness has been diagnosed and the prognosis properly assessed, but each particular case should be considered separately. Genetic advising (of couples) may be a useful tool in preventive medicine; however, it must never overrun the parent's decision.

Advice to adults at risk of developing a late onset disease should be given, although no rigid rules to follow in these cases. For children affected by untreatable disease or with short-lived prognosis, advice to parents should be provided, albeit each case is to be considered individually (more in Section II.5).

- JAPAN:

Genetic counselling is done only if requested, unless it is a disease in which a particular researcher is interested in.

- ISRAEL:

Done throughout life: after birth of a child with genetic disorder, in potential couples or parents with suspected family history, even now in premarital or preconceptional counselling even without suspicious family history. Counselling is used by the public regardless of whether the disorder is treatable or not. It is voluntary, not directed.

- CHINA:

Open to couples worrying about their baby being genetically normal, or who have prior defective child or affected relative, or have read in journals or books about inherited diseases and worry about a future or already born child. No limits even when no treatment.

- ZAIRE:

Open to those who worry of general risk in procreation, or for having a relative affected. Primarily for couples but now also for individuals. It would seem correct to abstain of genetic investigations in minors when the risk is only for procreation or when no treatment is possible.

II.5 What are the information ethics of genetic counsellors? Do they consider themselves as providing non-directed advice - simply information (however complex the idea of supplying information without advice may be), or do they think of themselves as serving as well social interests in public health?

There is a definite ambiguity in the non-directive aspect of counselling. Even just listing all the options, will have some directive influence. While non-physician counsellors or nurses will try to only explain the facts, the primary care physician counsel will most often be more directive. The ethics of counselling vary with who does the genetic counselling, but also with the nature of the case dealt with. A constant is that the welfare of the counsellee should always be above any consideration of public health and certainly of "society interest". This is in fact anchored in the European Bioethics Convention, approved by the Assembly of the Council of Europe on 2 February 1995, which states the *"recognition of the supremacy of the interest and well-being of human beings vis-à-vis the mere interest of society and science"*.

- UNITED STATES OF AMERICA:

Counselling by MSc-level (or Ph.D.) genetic counsellors is non-directive, whereas counselling by primary care physicians (especially obstetricians) is more likely to be directive. The genetic counsellors (MSc) do not try to serve social interests of public health: their main obligation is the welfare of counsellees.

While non-directiveness is a widely accepted ethical principle, experience has taught that Latino patients, for example, sometimes view non-directiveness as "detachment" on the part of the counsellor. The ethical challenge is to empower the patients to make their own decisions while at the same time to provide close emotional support.

- UNITED KINGDOM:

Counselling aims to be non-directive, putting in perspective the options and their possible outcomes, together with the benefits and disbenefits. However, this does not mean simply telling people the facts and leaving them to make their own decisions. Counselling involves actively helping couples to reach decisions in the context of their unique medical, moral and social situation.

- ITALY:

The rule is that of non-guidance. But, if due to the counsellor's personal convictions, it is felt necessary to take into account the social interests of public health, this is explained as part of the whole information given. Genetic counselling deals mainly with the problems of individuals (rather than society). The objective is not to provide a pre-established solution based on particular standards of individual or social well-being - but to explain every possible solution as objectively as possible, to allow a free and conscious choice. This may be hard to achieve; therefore, the psychologist in the counselling center plays a fundamental role as supervisor of the geneticist's work, both in substance and in form.

- CHILE:

Counselling is more likely to be directive, but in complex problems counselling is non-directive (for example in prenatal testing or in paternity determination). The information ethics of genetic counselling is based on classical medical ethics. It should respect the autonomy of the counsellee.

- MEXICO:

Most genetic counsellors view themselves as providing non-directive information. However, the evidence is that when faced with concrete clinical problems, their advice is indeed directive.

- ARGENTINA:

The details of counselling depends on the case: for example, Down's syndrome for an aged mother would be "simple" counselling. Genetic testing for a child with ambiguous genitalia would be more complex: the counsellor will predict the risk for future offspring, advise on preserving hydroelectrolitic balance, recommend genetic sex determination before civil registration of the child's gender. In prenatal counselling, advising should not overrun the parent's decision.

Counselling with intent to prevent spreading of recessive genes in the society seems to be of low relevance, because of the high number of recessive genes, and the high mutation frequency of certain genes.

Advice to adults at risk for late-onset disease should be given although there are no rigid rules to follow. Advice to parents of children affected by untreatable diseases, or short life expectancy, should be given, albeit each case must be considered individually.

Most important, counsellors must be cautious to avoid that counselling itself becomes iatrogenic, causing more harm. To protect a counsellee's welfare, the information given may be only partial. As an example, take a couple - well over reproductive age - with a son affected by an X-linked disease and who was about to die. The son's pedigree revealed that this was a fresh mutation, and the risk in other members of the family did not exceed the usual statistical probability. The mother was so deeply affected by the hopeless child that distressing her more seemed too cruel: therefore, she was not informed that she was herself the carrier of the X-linked disease, and information on the gamete in which the mutation had occurred was omitted.

- JAPAN:

Officially counselling is non-directive. However, given the unequal relationship of client and counsellor, it may be very difficult to assume it is perfectly non-directive. For example, some doctors may recommend and other may refuse to perform abortions for Down's syndrome.

- ISRAEL:

Basically non-directive, providing up-to-date, accurate, precise information in the most understandable fashion without any judgmental interventions or introduction of any

public health or social factors. Since Israel is an ethnic mixture with many new immigrants, counsellors face situations where counsellees need some guidance with regard to certain decisions that need to be made; however, it is always the interest of the counsellee and not of public health which is being served.

- CHINA:

Genetic counsellors seem in between providing non-directive information and serving social interests in public health. They should not make decisions based on their own values for clients, even when clients, because of ignorance or frustration, want counsellors to tell them what to do. It is a guiding role: often both sides share the same values. There is a feeling that these should not be against government policies, which include reducing quantity of population and raising its quality, meaning quality of life or prevention of seriously defective births. However, if the client insists to have this child, the counsellor cannot intervene. There is no governmental directive, but many counsellors do what they think is in the client's best interest as well as social interest.

- ZAIRE:

The frame is non-directive information including means to remediate to the situation. However, one should not only hold a "pregnancy-interruptive" reasoning. The counselling by two experts has to be on the severity of the pathology, and be well separated from the eventual decision by the mother.

II.6 Do genetic counsellors pay attention to the language difficulties and other, encountered in conveying information and/or advice, for instance to individuals from ethnic minorities?

Language communication is the key for insuring autonomy of the counsellee and free in his decisions. Difficulties in genetic counselling are numerous, and are often due to the difficulty to translate technical concepts, such as probability or heritability into words that can be understood in all what they encompass. This becomes increasingly true as the technical possibilities for gene diagnoses diversify rapidly.

There may be different sensibilities in ethnic groups in whom higher incidence of particular genetic diseases are noted. This incidence should be viewed as due to regional or ecological causes and not as ethno-racial traits. The language of counselling must not only convey information but harmonize with the cultural, social and religious environment. For example, support for the patient is more important in low socio-economic groups and non-directive counselling may be perceived as detachment, lack of interest on the part of the counsellor. Moreover, immigrants and aliens are often poorly covered by health-care insurance programs.

- UNITED STATES OF AMERICA:

Ethnic groups with increased incidence of specific genetic diseases should receive counselling. Carrier states, if diagnosed, should be clarified.

The example of genetic counselling of Latino-Americans is developed by Penchaszadeh[3]. The overall frequency of genetic disease is similar to other ethnic groups, with specific higher incidence of hemoglobinopathies and neural tube defects. But, almost 1/3 of Latinos in the United States have no form of health insurance. Preference for Spanish-speaking health providers contribute to a reduced access to health care, since there is a shortage of such professionals, especially genetic counsellors. The primary ethical concern in genetic counselling of Latinos is the deficient access to genetic services. Another problem is stereotyping: counsellors should be aware of the heterogeneity and beyond cultures, every patient should be addressed as an individual. Spiritual/religious beliefs must be taken into account for an ethically responsible genetic counselling. Individuals have a very personal interpretations of religious teachings, particularly for reproductive decisions, which are also influenced by economic concerns, limited family assistance and education. The ethics of non-directive counselling is challenged when, as frequently occurs among Latinos, the patient sees him/herself not as an autonomous individual but as a subordinate of the health professional.

Women may be too submissive to the will of their husbands. A high number of pregnant women are young, single and burdened by socio-economic troubles that make genetic risk appear ridiculously low in comparison. Disbelief of genetic risks and test results, because of superstitions or beliefs in supernatural explanations, require exquisite sensitivity and respect by the counsellors. Reliance on written educational materials require adjustments for literacy levels, cultural relevance. Tendency to rely on extended family for support and decision-making can lead to "unorthodox" counselling settings. View of non-directive counselling as "detachment" on the part of the counsellor makes an ethical challenge to empower patients to make their own decisions while at the same time to provide close emotional support. Cultural values and the wider social context of immigrant experience, poverty, and fear of added burden by genetic conditions, must be taken into account to avoid two ethical perils: one of paternalism, that can lead to authoritarianism and enforced compliance with "eugenics" or "public health" imperatives; the opposite peril being to fail to provide adequate support. Culturally appropriate and accessible services, and social support for individuals facing genetic risks for a child, as well as for those born with genetic disabilities, should be advocated since they are essential to make a reality of the right to decide.

Language difficulties exist in a broader sense in the US society: the possibilities in molecular diagnoses resulting from the Human Genome Project will increasingly require better-informed and more complex counselling to deal with situations such as for examples: Huntington's disease (variable mid-age onset but predictable outcome); Alzheimer disease (multigene, late onset, environment role); cancer susceptibility genes (multigene, probability calculation, option of early treatment if diagnosed early through frequent monitoring, preventive measures). There is an increasing ethical concern about screening tests for a variety of cancers, which may be premature but touch upon the ethics of the "people’s right to know".

- UNITED KINGDOM:

To meet the requirement for autonomy it is essential to communicate the diagnosis and its implications effectively. This can represent a major challenge since the language, culture and social level of those counselled cover such a wide range. A couple’s decision on whether to embark on a pregnancy at all, or proceed with prenatal diagnosis, may depend on how the information is transmitted.

- ITALY:

Difficulty to translate technical terms and concepts such as probability into words that can be understood by the layman. It is necessary for the counsellor to enter into the personality, culture, social and religious environment of the counsellee. It is particularly difficult with immigrants (whose background culture is less familiar to the counsellor).

- CHILE:

Ethnic minorities have no access to genetic counselling due to the absence of counsellors at the primary health care level provided by the National Health Service.

- MEXICO:

Attention to language difficulties in conveying information is given not only when patients belong to ethnic minorities, but also when the socio-economic and cultural levels are low.

- JAPAN:

Language difficulties in understanding doctors generally exist for all patients. Some physicians may use German or English names rather than the Japanese names to hide the nature of the disease or of a drug.

- ISRAEL:

Language barrier is one of the most important concerns in genetic counselling. Genetic counsellors are very aware of this and provide translation. Appropriate language and concepts have also to take into account that the society is a fusion of ethnic and cultural backgrounds.

- CHINA:

Genetic knowledge is based on the language of atom and molecule, but traditional Chinese are familiar with the language of *Yin and Yang.* Education will help. There is a cultural barrier also: disbelief or reliance on good fortune.

- ZAIRE:

Ambiguities in language and culture have to be realized to give information as precisely as possible. Certain physicians take too much liberty with this.

II.7 Is there any government involvement in decisions about what is said in the context of genetic counselling?

The States do not openly control what is said or decided in genetic counselling. However, some countries (e.g. France and Germany) have passed laws, which does involve the State in decisions and options related to genetic counselling.

- UNITED STATES OF AMERICA, JAPAN, ISRAEL:

No government involvement in decisions about what is said in the context of genetic counselling. Nevertheless, there have been attempts (such as for matters regarding abortion) to limit what physician can say when working in government institutions (for example the "gag" rule which was in effect for a while in the United States of America) (Section IV.2).

- UNITED KINGDOM:

No government involvement in what is being said, but the Department of Health takes interest in informing on beneficial uses of genetic testing or screening, and in keeping genetic counsellors in touch with families who may need them.

- ITALY:

Government or regional health authorities will only intervene by defining the levels of general and specialist health assistance. Laws currently in force, safeguarding the health of individuals, including the unborn, must be observed in every respect. However, there are currently no legal regulations disciplining medical practice applied to assisted procreation, which would bring law into line with technical developments.

- CHILE, MEXICO:

Government officials and the Ministry of Health are not involved. Genetic counselling is a private affair between persons; the State may be excluded from the process.

- ARGENTINA:

The State concern for genetic programs in public health care is limited, although it participates in public campaigns aimed at decreasing or preventing infectious, parasitic, nutritional or drug-abuse diseases.

- CHINA:

No government involvement so far. However, with the enforcement of the Law on Maternal and Child Health Care, officials may be more involved. But as officials in Ministry of Health say "the Law is a soft law in which many items are advice suggested to citizens".

II.8 Is information reported to public health authorities? Is information gained from one member of a couple provided to his or her spouse? Is it provided to other members of the family? Are these relationships handled by rules, by custom, or how?

Genetic information relating to patients counselled is not reported to public health authorities. There may be some interests to keep data bases on genetic diseases for research purposes, and care must be taken to respect anonymity. The problem of information leaking to insurance companies, or whether or not it is in the patient's interest to inform his insurance company could be important to discuss during counselling.

Communicating information to the spouse is in most cases subject to consent by the patient or the person who asks consultation. For the unborn or child, this will be most often the mother, and informing the husband is not automatic. Paternity testing would be a case were confidentiality to the spouse is obvious. Problems may arise in pre-marital testing. Informing other members of the family, including those at risk of having the genetic disease or being carriers, is also usually dependent on consent. At present, these decisions seem to be discussed case by case with the counsellor, who must be prepared to handle these delicate questions.

- UNITED STATES OF AMERICA:

Confidentiality is most important and, only on rare occasions, where other lives may be endangered and the counsellees are being uncooperative, is confidentiality broken.

Of increasing importance is the potential impact on privacy by insurance companies. The counsellors discuss this with the couples in detail, without telling them what to do, but stressing that the information is as private as they wish.

Information is not reported to public health authorities without the counsellee's permission. Information is usually provided to the spouse only with the consent of the spouse who has been counselled. The counsellor suggests that other members of the family, identified in the pedigree, be informed of their risk of genetic disease, if any, by appropriate counselling.

- UNITED KINGDOM:

The highest standard of confidentiality is preserved.

- ITALY:

Professional secrecy in medical practice applies to counselling. The reply must be given to the individual who asked for consultation, and only he/she can give consent to divulge the information. The geneticist and psychologist may, in individual cases, stress why it would be advisable to inform the spouse or relatives, but the consent to do so by the person counselled is in any event required.

- CHILE:

Confidentiality and privacy are by law, protected from the State and insurance companies. Genetic information is not reported to health authorities, only cases of infectious diseases. Information is provided to the spouse only with the consent of the counsellee, and anyhow restricted to the family.

- MEXICO:

The information is not reported to public health authorities. Counselling is mostly done to couples, but transferred to other members of the family only with consent. There are no set rules, and every case is negotiated with the patients.

- ARGENTINA:

Geneticists do not report to health authorities the knowledge gathered on the frequency and severity of genetic diseases affecting different populations. Nevertheless, each center records its activities in its own database. In addition, agreements towards having a National Registry of Genetic Diseases were recently signed between the Public Health Ministry and university institutes.

- JAPAN:

There is no central reporting or database for genetic disease. It is up to the spouse to tell the partner, unless there is a life threatening disease, in which case the spouse and family may be told before (or instead of) the patient. Whom to tell depends more on the doctor than on custom. Some information may be withheld even from the affected person, e.g. Huntington's disease.

- ISRAEL:

No report requested to public health authorities. Medical secrecy commits every physician and medical person. Patient consent is needed to forward any information to spouse or any family member. When appropriately identified and for reasonable purposes

(e.g. prenatal counselling where there is a concern of a genetic disease in the family and the diagnosis needs to be verified), a physician can request genetic information without the consent of the patient. This is an exception with no written rules.

- CHINA:

Information obtained in genetic counselling has never been reported to public health authorities, as obligatory for communicable diseases. If the genetic disease may affect any member of the family, the member should share the information. Generally speaking, consent should be obtained, except if keeping confidentiality of the information will harm the third party. If the client insists not to disclose, and the disclosure of information will do no harm to the client, the counsellors should balance the positive and negative consequences caused by disclosure (or keeping it confidential). These relationships are handled by custom as traditionally done; however, great efforts have been made to handle them by rule.

- ZAIRE:

Secrecy rules transfer of genetic information to spouse, family, even to the patient's physician, unless authorized by the person examined. The physician who does the genetic counselling has to bring the client to reveal himself the pathology or risk if this disclosure to others is useful. Secret can also be kept from one of the parents, except if the patient is a minor: then, his parents have a right to the truth. Confidentiality is also maintained toward health insurance and social security officials.

III. Scientific Issues in Genetic Counselling

III.1 Genetic Technologies

It is important to distinguish between "proof-tested" technologies and new or emerging technologies. This distinction applies both to the type of genetic test available to the counsellor, and to the type of human sample on which the test is being done. The new technologies multiply the number of options that the counsellor can point out to the counsellee, and often make these options more problematic than in the past. As a researcher stated: *"*(...) *with genetics, the technology far precedes the response to what we are going to do with the technology"* [(4)].

Types of genetic tests. Proof-tested technologies such as cytogenetics, karyotyping, enzyme tests, are being supplemented by numerous molecular gene probes. The gene probes can detect polymorphisms which have been associated with genetic diseases, and indicate who inherited such "pathologic" chromosome fragments and who has a non-pathologic DNA polymorphism. However, it becomes clear that many mutations can occur in genes related to a disease and that the significance of each such mutation may be different in terms of clinical disease. Detailed analysis by Polymerase Chain Reaction (PCR) sequencing techniques of such mutations or of other variable regions (short repeats) provide information which must be correctly interpreted for good counselling. For example, mutations in the gene for Gaucher's disease or in the BRCA-1 gene for familial breast cancer are numerous, and before the pathological outcome of each one is determined, prediction of disease is uncertain.

Time is needed to allow these recent research results to mature into "proof-tested" medical practices. Although these technologies are of diagnostic nature, they have a profound impact on life and quality of life, and may have to be regulated like new therapeutic drug approvals.

The many possible sources of human sample tested raise new questions. Testing blood or tissue swab samples from newborns, children or adults, can be prompted by a visible disease, a family history or can be screening programs of various extents. Prenatal testing can be *in utero*, aided by echography and using fetal cells from amniotic fluid, fetal blood or chorionic villus sampling (CVS) which allows first trimester testing. These techniques, even amniocentesis, present some low risks for the mother and fetus, and have often to be approved by several independent physicians. The time in pregnancy when the results are obtained may be critical for eventual intervention. The new technologies of genetic testing on pre-

implantation 4-cell embryos[5], through single embryonic cell DNA testing by CPR, can be used in the context of assisted procreation (IF). Their success has been shown in many cases and they allow in principle selective implantation of healthy tested embryos in high-risk couples (examples based on experience in the Unites States and in Israel would be Tay-Sashes, cystic fibrosis, female sex selection in X-linked disease, Rhesus incompatibilities). Even though IF is complex, lengthy and expensive, it expands rapidly in many countries. More recently, non-invasive tests on fetal cells circulating in the mother blood[6] offer new possibilities to transform prenatal diagnosis into routine procedures applicable to low-risk genetic diseases for which the invasive techniques (amniocentesis, IVF) are too risky or expensive. Pre-marital genetic testing aimed at detecting gene carriers, and counselling heterozygous couples not to have offspring together, is a form of pre-conception intervention[7, 8]. The advantages, accuracy and safety of each technology will have to be weighed carefully.

III.2 Gene Categories in Counselling

The public information about gene mutations related to diseases grows rapidly, mainly as a result of the Human Genome Project, but is not always presented in a way which allows to understand the meaning of the gene pathological function and even less of its normal function. The equation "gene = disease" is often perceived without qualification, so that it may be useful to classify genes in order to qualify their relation to clinical pathologies. A proposal is made to distinguish genetic alterations in at least 5 groups, each with differing meanings for genetic counselling.

Classification of genetic alterations relevant to counselling (with a few examples)	
I.	**Lethal in childhood or grave malformations** • Tay-Sachs (GM2 ganglioside, hexaminidase deficiency, life-span 4 years) • Mucopolysaccharidoses (Hurler, San Filipo, death in 2nd decade) • Gaucher Type II (betaglucosidase, lethal in childhood) • Cystic fibrosis (respiratory disease, median life-span 25 years) • Achondroplasia (nanism, malformations, FGF receptor I) • Trisomy 21 (Down's syndrome, non-hereditary)
II.	**Viable-if-treated hereditary diseases** • Phenylketonuria (low phenylalanin diet) • Galactosemia (exclusion of milk) • Hemophilia (X-linked, Factor VIII or IX replacement)
III.	**Late appearing genetic diseases** • Huntington's chorea (40 years; early if CAG repeats >> 32 in HD gene) • Myotonic dystrophy (onset in adult life, CTG repeats in DM-1 gene) • Familial hypercholesterolemia (onset 30-40 years, responds to treatment) • Alzheimer disease (at least 3 genes, Presenilin I, II, APO-E)
IV.	**Predisposition genes** • Spondylarthritis (HLA B27 - 600 higher chances) • Cancer: melanoma, bladder (environment factors), breast (BRCA-1, -2 in familial cancer which are 5% of all breast cancers, so role in most cases uncertain) • Schizophrenia (but in psychiatric diseases phenotype may be malleable)
V.	**Multifactorial diseases and their genes** • Diabetes 5% incidence but 6 genes (e.g. MODY = glucokinase gene) • Cardiovascular diseases 20% incidence but how many genes? (e.g. cholesterol receptors, angiotensin locus, coagulation factor V)

Remarks on gene classification:

The examples given could sometimes fit in more than one category. The classification does not aim at more than just stating: not all gene testing have the same meaning, and counselling should be appropriate.

In group I, the issues may be mostly concerned with prevention. Premarital examination and counselling is practised already for Tay-Sachs carriers in Jewish communities at risk[7, 8]; this could be the best approach because carriers are detected and encouraged not to marry, while preserving anonymity. Pre-implantation diagnosis of Tay-Sachs is available [6], but it obviously entices much more ethical and legal problems. Screening for cystic fibrosis carriers becomes widespread[9]. With certainty, the perception of whether the risk to have an affected child is too big or can be accepted will vary individually among couples and families. However, there will be little discussion that such genetic conditions are not compatible with the kind of life parents desire for their offspring. Hence preventing birth of such children can be understood by many as a justified behaviour.

Group II will hopefully grow in the coming years as more diseases become amenable to treatment. Thus, one may hope for a gene therapy for cystic fibrosis, through virus (Adeno- or others) based gene transfer to the lungs. However, some therapies may have their own problems: blood and platelet transfusions for hemophiliacs has taken a heavy levy of lives due to HIV blood contamination, and parents who have lost a child - not to the genetic disease but to the therapy - may not want to have another diseased boy. Pre-implantation sex determination may be an option in such cases.

Group III shows more difficulties in genetic counselling. Even if the genetic conditions have a high penetrance and disease is almost certain, as in Huntington's disease (HD), life may be normal until the age of 38-40. The abnormal length of the CAG repeats in the HD protein gene (probably leading to accumulation of polyglutamine) varies, and the length can predict the age of onset of the debilitation. Does that justify prevention? Other diseases appearing late in life, and attributable to a hereditary gene condition, could be added to this group, provided the probability of disease is high. Many, like Alzheimer disease, seem to involve multiple genes[10].

Group IV raises in addition the distinction between genetic disease and genetic susceptibility[11]. Counselling involves calculating probabilities of disease, probabilities which may also depend on the way of life and the environment. Inherited gene changes which increase susceptibility to certain cancers, or to certain psychiatric diseases, have come into the limelight, but counselling should take into account the many uncertainties.

An example to discuss are the growing number of cancer genes. A relative simple case are BRCA-1 and BRCA-2 dominant gene mutations which could account for 60% of familial breast cancers and 5% of all breast cancers[12]. Even though probability is high (maybe 85% lifetime risk of breast or ovarian cancer), many questions have to be asked regarding age of disease (40% of women with BRCA-1 will be cancer-free by age 50), possibilities of early diagnostic (annual mammography from age 20), in order to avoid labelling the finding of BRCA-1 mutations as a death sentence. Preventive mastectomy seems an extreme measure. The question is still open of whether BRCA-1 is involved in sporadic cancers and testing the gene may be best restricted to families with early onset breast cancer. The number of genes involved in various cancers is growing, but somatic mutations in the tumor cells may be quantitatively more important than heritable genes (e.g. colon cancer[13]). Excluding one heritable gene does not exclude the risk of cancer. Hence it is important to keep things in proportion and not to create a hysteria of gene testing for cancer[4]. Early detection of cancer by clinical screening is important but the recent emphasis on gene screening for a variety of cancers may not add much to cancer prevention.

The last group (V) are the increasing number of genes which are implicated in the diseases that are the most frequent in the human population such as cardiovascular diseases, diabetes and other nutritional conditions. These diseases are multifactorial and involve many

gene defects: six known for diabetes, probably more for heart disease[14]. Detection of one or several defects may be valuable in family studies but do not allow yet to evaluate the increase in risk of getting the disease in unselected individuals. Moreover, here as well the lifestyle and environment play a considerable role.

IV. Ethical Issues in Genetic Counselling

IV.1 Are All Options Open? Ethical Values of Counselling Options

The Code of Ethics of the NSGC (United States of America) states that counsellors *"strive to enable their clients to make informed independent decisions, free of coercion, by providing or illuminating the necessary facts and clarifying alternatives and anticipated consequences"* [15]. The alternatives or options to be explained often involve important ethical questions. Are all these options equally open to the counsellee? Are they all practically and morally justified and comparable?

Counselling for born individuals

The survey has indicated that there are no limits in obtaining genetic data in a child, or adult, even for untreatable diseases. What are the options available for counselling in such cases?

Risk evaluation is one option, that is telling a still healthy person that he/she has such and such probability of developing a disease at a given age. The psychological impact of such genetic predictive testing has been addressed recently in the context of screening for cancer genes[16]. While it could be feared that this may only raise anxiety, counselling had also beneficial effects because of a better assessment of risk. In a high-risk group with familial history of breast cancer, this United Kingdom study found that a quarter of the women were reassured because the risk calculation was lower than the one they perceived before counselling. In another quarter of women, who had underestimated their risk and learned that their risk was higher than thought, counselling did not seem to make them more anxious "suggesting that they would rather not confront the information". Even in single gene diseases for which gene testing can predict an untreatable disease with almost certainty (e.g. children of Huntington disease patients), most persons concerned take the test and knowledge of the test results seems to alleviate anxiety[17].

Preventive options may include advising frequent monitoring for early diagnosis of the clinical disease. For example, frequent mammographies for breast cancer would appear a better option than preventive mastectomy. In other genetic diseases, less dramatic preventive treatments may be possible (diet, drugs, psychological help). Examples are present screening of newborns for PKU, hypothyroidism, with immediate treatment.

Gene therapy may become an option in the not distant future, but will most likely be limited to supplying the normal gene into somatic cells (bone marrow, skin, mucosa cells), for example in ADA deficiency, hemophilia or cystic fibrosis (see 1994 IBC Report on Gene Therapy).

Testing all members of the family is an important process to clarify the carrier state for the diagnosed genetic pathologic trait and the counsellor will suggest to do so. Questions of breach of confidentiality may then arise[18]. Counsellees have in such cases reported deterioration in the relations with relatives (who feel threatened by the information) although genetic counselling appears beneficial for the nuclear family[19]. The right of a relative not to know of a genetic disease should be respected. Experience shows that too few individuals ask for the tests. There should be no coercion for family members to undergo a test or receive the results, although the beneficial aspects of genetic testing and counselling should be presented. The mutual responsibility should be stressed.

Confidentiality within the couple. Communicating genetic information to the spouse or partner should be done with consent of the counsellee as in other medical situations (Section II.8). This may be more delicate when a woman consults during or prior to

pregnancy, but the decision should be left to the mother. If relations within the couple respects the sanctity of their union, she will most likely give consent allowing prenatal counselling to be addressed to both member of the couple.

Prenatal counselling

Most subjects of reproductive age at risk of having an affected child with a severe single gene disorder surveyed in a United Kingdom (UK) study[19] express their intention to use prenatal testing. Methods include detecting genetic defects *in utero*, or prevention by selective embryo implantation or by pre-marital testing (Section III.1). When diagnosis is made *in utero*, the options open for counselling can include preparations for treatment at birth, if available, psychological preparation which will reduce the shock, planning ahead the future of the child, but will very often revolve around intervention to terminate pregnancy. As pointed out in the 1994 IBC Report on Genetic Screening and Testing, there are such divergences on the issue of abortion for severe genetic disorders that one can aspire only to the calm discussion of the ethical differences.

The option of pregnancy termination. Code of Ethics of the NSGC states that genetic counsellors strive to *"respect their client's beliefs, cultural traditions, inclinations, circumstances and feelings"* [15]. This is by no way easy to accomplish, as views on many issues varies. In particular, opinions and attitudes on abortion will vary with cultures and religions. In some, conception marks the point from which human life is holy and entitled to its complete rights (immediate animation in the Catholic Church) whereas, in others, the embryo will not be considered as full-fledged human being for different periods of times after conception (e.g. 40 days). This would be for example the position of Jewish law which also considers that if the fetus *in utero* endangers the mother's life, the mother should be saved by killing the fetus. While these considerations do not allow free abortion, they can include situations where knowledge of a severe congenital or genetic defect will endanger the mother well-being enough to accede to her will of interrupting pregnancy. This goes to show that not all traditional religions will have the same views, and *a fortiori* the "inclinations, circumstances and feelings" can be vastly different among those who do not refer to tradition or religion.

It is important to stress that genetic testing and counselling should not be considered as increasing the number of abortions and being suspect of "anti-life" activities. In fact, a Dutch study shows that when no test for a genetic disease (e.g. Huntington disease) was available, 50% of the couples at risk did not want to have children, whereas 85% of them want to have children once genetic testing and counselling is available[20]. For thalassemia, the rate of procreation increased from 50 to 98% when a prenatal test became available according to a UK Report[20]. The same applies to sterilization measures: women who had opted for sterilization because of a suspected carrier status for the fragile-X mental retardation, asked for reversal of sterilization following clarification of their carrier status which became possible after cloning of the gene in 1991, and many women of the different risk groups chose to have children[21]. Genetic testing and counselling tend, therefore, to increase reproduction by increasing safety and are to be seen in a "pro-life" light, which does not intend to increase the overall rate of abortions.

Selective implantation poses a new and interesting ethical issue in comparison to abortion. Is it preferable morally to select healthy freshly fertilized embryos for implantation than to abort a fetus at the month 3-4? Again the moral discussion may revolve around the definition of when full human life starts (see above), around analogies to "selections" in Nazi concentration camps, around whether a "mother" can be considered endangered by an embryo of which she is not yet pregnant. There are also many practical considerations related to the IVF procedure involved, the cost, the possibility of errors, but the method is being used and legal in several countries (Spain, Israel, United States of America). It is an option in counselling high-risk couples, but must be closely watched as it could technically make possible also germ-line gene therapy, trivial gender selection and eugenics (Section V).

Sex selection is still a valuable technology, e.g. for couples at high risk of X-linked diseases (Duchenne muscular dystrophy, fragile X mental retardation, hemophilia) and various methods may be employed (sperm fractionation, selective implantation, selective abortion). The non-medical arbitrary abuse of sex-selection is not thereby to be justified.

Pre-marital counselling is practised for carriers of genetic pathological traits, and may be applicable in regional or ethnic groups with relatively high incidence. Tay-Sachs among Ashkenazi Jews in Israel or in New York is an example: the method is suited for ultra-orthodox groups in which sex is limited to married life and weddings are prearranged. Interestingly, Rabbis are deeply involved in premarital genetic counselling[7, 8]. However, in some cultural environments, could love for a person not be seen as more ethically valuable than healthy reproduction? In Cyprus, where premarital screening for thalassemia is mandatory, 98% of couples in which both are carriers still marry according to a UK Report[20].

IV.2 Case-by-Case Individual Counselling versus Regulation, Laws

A consultand-centered ethics of genetic counselling means that it is initiated by a potential patient and aimed at enabling him to make informed independent decisions. Centering the ethics on the individual who consults and making it a duty to have is best welfare in mind, when explaining all the options available, would seem to demand a case-by-case approach rather than strictly following pre-established laws and regulations, that bind the counsellor by limiting the options available. The counsellee ought to be made aware of what is legal and illegal in the particular country. However, since laws vary with place and time, the counsellee should nevertheless be made aware of all possible options. For example, selective implantation could be mentioned as a possibility, if appropriate in this case, even though it may not be legal in the particular country while legal in a neighboring one. Selective implantation is prohibited in France and legal in Spain.

The wisdom of having strict laws limiting the medical options, being it in genetics or in assisted procreation, may be questioned. Medical ethics, and particular codes of ethics for genetic counselling, may be sufficient guidelines within which there is room for case-by-case advising respecting the spirit of the ethical codes. Problems posed by laws have been encountered previously, as illustrated by Parker[15]: *"Anyone attempting to establish a (government) policy focused on genetic testing itself and sought to prohibit testing to determine fetal sex should bear in mind recent experience with the so-called gag rule, which attempted to regulate what physicians in (government) funded clinics could advise their patients about abortion. Ultimately, this governmental regulation of the private interaction of health care providers was thought to interfere with the privacy of physician-patient interactions, to violate protection of freedom of speech and standards of care, and to leave physicians open to charges of malpractice, and the gag rule was struck down. For similar reasons, a policy prohibiting genetic testing to determine fetal sex for sex selection should probably not be imposed by bodies outside the genetic counselling profession"*. There may be justified reasons to fear from trivial sex selection, but such regulations could hinder medical sex selection in cases of X-linked diseases, such as the case of a couple who, having lost a hemophilic child, cannot bear the risk of another affected boy and ask for medical assistance in procreating a healthy girl. In a case by case approach, this assistance may be granted as ethical according to the circumstances.

Laws have to be just and consistent. As pointed out for the United States of America[15], if abortion is permitted through mid-second trimester without any statement of reasons, a law prohibiting abortion on the basis of genetic information on the fetal sex would be inconsistent. In Germany, there is self-regulating ethical rule not to provide information on sex determination prior to the end of the first trimester, the period of legally not penalized abortion[22]. If such rules are part of codes of ethics from professional bodies, it may be advisable to provide exceptions which can be dealt with case by case. Such decisions could be under the supervision of inter-professional ethical committees, like Helsinki committees, that could include lawyers, philosophers, religious and community leaders.

What is acceptable and bearable by couples and families in term of genetic alterations, being physical or mental, can be best evaluated in case-by-case decisions. This will be function of circumstances, beliefs and feelings. While counsellees should be encouraged (by counsellors, by Leagues for handicapped persons) to accept the maximum burden by correctly evaluating the consequences of a genetic condition, the options and possibilities offered by medical science should not be denied by laws, provided that the decision is made in the most autonomous way and with the best possible ethical counselling by physicians, geneticists and religious or laic ethicists.

The IBC may find it difficult to reach a universal ground of legal agreement on such numerous sensitive issues which vary among cultures and nations, but may be able to stand behind an individual decision-making case-by-case process that operates in the framework of comprehensive ethical guidelines.

IV.3 The Physician-Patient Relationship and Genetic Counselling

What will a counsellee do with the genetic facts presented to him, and how will he reach a decision, is an important question about the bioethics of genetic counselling. In this respect, it appears important to conserve the character of physician-patient relationship in genetic counselling. The sophistication of the topics covered by molecular medical genetics, as well as the psychological and ethical issues, may require and justify the creation of a new medical speciality. In some countries, as the United states, a specialized paramedical profession of genetic counsellors has emerged, who are not physicians. Nevertheless, all genetic counsellors should be directed to adhere to the bioethics of physician-patient relationship. The participation of other professionals, such as psychologists and ethicists, should be encouraged as their directives will help to reach a decision and to cope with it, but the physician should remain the pivot of the team.

Involving primary care physicians in genetic counselling is also a way to remediate to the current insufficient number of trained personnel to meet the demands for genetic services[23].

Physicians have an intent to treat, while it may be argued that the function of the genetic counsellors should only be to inform of the facts in a totally non-directive way. Indeed, studies have shown that the attitudes of obstetricians, clinical geneticists and genetic nurses differ in counselling following diagnosis of fetal abnormality[24]: up to 68% of physicians reported counselling in some directive way, versus 43% of the geneticists and 6% of the genetic nurses. However, the bioethics of doctor-patient relationship has evolved from the 70s to the 90s: it is no more a paternalistic ("priestly") model but a *"contractual model according to which, ethical authority and responsibility are shared by professional and client.* (...) *The relationship between professional genetic counsellors and their consultands reflects this shared decision-making process, which guarantees to consultands the authority to make choices reflecting their own values"* (quoted from Parker, Ref. 15). Sharing the decision, in a way which respects the patient's autonomy but takes from him some of the burden, would appear to best insure the moral obligations to help the patient, to maintain or restore his welfare through trust and confidence, obligations which characterize the doctor-patient relationship.

The question of non-directive, objective counselling deserves further scrutiny. As seen in the survey (Section II.5, United States of America), some persons consider non-directed counselling as detachment, lack of interest in their problem. In this respect, the following thoughts may be of value (Garver and Garver, Ref. 25, quoting from Friedman and Reichelt in the "Los Alamos Science" published by the Los Alamos Center for Human Genome Studies): *"The current standard for the [genetic counselling] profession is to present information in a non-directive, value-neutral way and in a manner that preserves client autonomy. Essentially that means that the counsellor should not project his or her values into the patient. But does this standard work in a practical sense? A patient with a high cholesterol level is not told by his doctor, 'Your cholesterol is 350. It could kill you, so gather some information on cholesterol and make whatever decision you want'. The doctor's advice will be much more directive; it is likely to include recommendations about treatment or lifestyle changes that can ameliorate the illness. Those in the genetic counselling profession, however, still cling to the non-directive counsellor and autonomous patient model - this model is increasingly untenable"*. Garver and Garver[25] continue: *"This is an inadequate analogy, because when a physician counsels about a high cholesterol level or about a sore throat or most other medical conditions, it does not involve a very important ethical or moral decision by the patient. On the other hand, many medical genetic decisions involve serious questions about prenatal diagnosis, abortion, and sterilization, which have different moral and ethical implications to most patients. These serious moral and ethical decisions have to be made by the patient, with the assistance of the physician, clinical geneticist, or genetic counsellor and sometimes with the advice of his or her rabbi, minister or priest"*.

The arguments are well presented, and stress the practical difficulties in non-directive counselling. One could add that often physicians do make important ethical decisions, with the patient's or family consent, as in the case of grafts, major surgery or terminal patient care. Why then do many feel that these situations are different from genetic counselling? This may stem from the fact that in genetic counselling there is usually no certain or immediate medical emergency involved and that, in the case of prenatal diagnosis, the relation to the unborn life is apprehended differently than the relation to an individual with a life-endangering illness. These appear to be secondary and subjective differences, which do not justify to take out genetic counselling from medical practice and to deprive the patient from the more directive support given by physicians.

Difficulties for physician in genetic counselling. Fully informing the consultand about genetic conditions may be difficult for physicians, certainly for primary care providers. The information is complex technically, and the ethical implications, the psychological impacts, may be beyond the physician experience. However, a fundamental bioethics principle is that of informed consent, which obligates "*to disclose the information in such a manner that a reasonable layperson can understand it and to answer the specific questions which the individual client or research subject may raise*" [(15)]. There is obligation of dialogue with the patient. Physicians have to comply with these obligations, which means providing their own time and effort, for counselling as well as for treating. Physicians need often more education in genetics, and should closely co-operate with more specialized professional geneticists and ethicists so that the obligations of communication and information are fulfilled (to enable consultands to make free and informed reproductive and health-care decisions), but without losing the medical intent of helping the patient to maintain or restore his welfare.

Counselling is not a one time affair and should be repeated as needed. The right to a second medical opinion may have to be extended to the right of a second information by another competent professional. This should be granted by the treating physicians in order to respect their client's own values, and help them reach the best possible decision about what to do. Giving moral support to these decisions is also a primary task of genetic counselling.

V. General Moral Issues Related to Genetic Counselling

The survey (Section II.7) indicates no involvement of States or governments in genetic counselling, while stressing its essential individual-centered bioethics. However, one should ignore neither the impacts that genetic predictive diagnosis will increasingly have on society nor the reactions of society to it. It is a sane aspect of "preventive bioethics" to anticipate society problems which may be easier to prevent than to cure[(15)]. There are practical and economic issues, but also moral and philosophical dilemmas which may be defused only through a committed educational endeavor toward the public and authorities.

V.1 Social, Economical, Political Issues

Voluntary aspects. Genetic counselling and testing should remain initiated by the individual or the person having legal responsibility (mother). There should not be systematic or compulsory programs imposed by society. Such obligatory screening programs may be prompted by genuine population health concerns (as in regions of high prevalence of a severe hereditary disease), but could too easily extend to behavioral traits with supposed genetic basis: alcoholism, homosexuality, crime[(26, 27)], creating serious concerns of reviving eugenics (Section V.2). Therefore, while society has a duty to make scientifically-proven gene testing available to the public, including prenatal or premarital counselling, it should never make it compulsory but only voluntary. Requirement for genetic testing in certain professions, where biochemical testing is usual (e.g. air pilots), may also be very problematic.

Confidentiality from employer. The predictive aspect of genetic diagnosis may prompt employers to dismiss still healthy employees by fear of eventual later disease. This would be unfair discrimination. On the other hand, knowing of a genetic predisposition may

avoid exposure to chemicals, environment that could increase the risk of disease. An employee may sue a company for not having tested such a genetic trait. Thoughts have to be given to such complex issues.

Confidentiality from insurance companies ought to be respected and upheld by proper legislation. This in order to prevent exclusion or higher premium for carriers of genetic traits that may be considered to increase the risk of disease (e.g. cancer genes, mental diseases). Such problems exist in other medical tests (e.g. high cholesterol), and may also be questionable since views on etiology of diseases (e.g. cardiovascular) still evolve with research. With genetic diagnosis, problems are more complex since the number of genes testable is large, testing may precede any symptom of disease or even gestation. People may be penalized for their genotype in a way not necessarily related to their phenotype. The other side of the coin would be persons who have received information about a certain genetic abnormality and may try to obtain insurance protection, for themselves or their offspring, without revealing the information, thereby taking unfair advantage from insurance companies.

Health insurance can certainly play a positive role by covering costs of genetic testing and counselling, or of early frequent monitoring of disease (e.g. frequent mammographies for BRCA-1) which would not be the case if the insurance covers only tests following a clinical symptom. The issues regarding insurance are, therefore, complex.

Confidentiality from State. There may be similar concerns regarding national health care systems as for private insurances. For cost-effectiveness, health-care rights of families who carry a gene pathology may be jeopardized, because of the costs of genetic testing or cost of hospitalization and care, or worse by encouraging them not to reproduce, if not to be sterilized[25]. This amounts to double-jeopardy, penalizing those who suffer from a natural variation outside their control (somewhat alike health care discrimination for age).

Data banks on genetic diseases may have values for health-care and research, especially for position mapping and identification of new genes on the basis of family studies. Such data bases should remain as far as possible anonymous, or coded, and not made available for any State activity outside justified ethical research programs, which are peer-reviewed.

Supervision and education of genetic counsellors. With the progress of the Human Genome Project, genetic counselling will very likely grow into a full medical specialization, if not a new professional area. Modern genetics should be taught and Medical Schools and in specialized courses. Advising, informing and educating the public about the meaning of genes, polymorphisms, mutations, certainties and uncertainties of genetic determination for health, behavior, performance, even intelligence, will be in great part in the hands of genetic counsellors. Involuntarily or unknowingly, the counsellors own values may influence their attitudes on these complex issues, some of which touch upon nature/nurture debates where both sides are partly right and which are never over[26] . These challenges will require guidance and education of counsellors beyond what is offered today by professional organizations and their codes of ethics. Who will provide guidance and whether inter-professional and international ethical bodies may be the answer should be discussed. Few would probably want politicians alone to make the rules, but controlling the counsellors has to be considered with lucidity.

Commercialization of genetic testing and counselling. New DNA probes and PCR or other diagnostic systems are being rapidly commercialized. Specialized diagnostic companies offer gene testing on a commercial basis and market them actively. This creates much debate, in particular for the group of cancer genes[4] whose value for the general public is quite unclear (Section III.1). Along with testing, there is a possibility that counselling will also soon be commercialized privately. Although these are diagnostic activities, and could be assimilated to sugar, cholesterol or blood pressure testing provided nowadays as "supermarket" services, such direct marketing of genetic tests[9] can lead to much more complex ethical problems. There are already advertisements for fetal sex determination and selection. Clearly, some thoughts about how to regulate private commercialization of genetic testing and counselling become urgent. Regulatory authorities should authorize the genetic tests as other therapeutic procedures.

Judicial responsibilities. Professional errors made by genetic counsellors have already led to lawsuits in several countries. Particularly revealing of the ethical problems are "wrongful life" lawsuits by defective newborns for faulty genetic counselling. As one example[28], an error led to exclude during pre-marital genetic counselling the risk of a woman for Hunter's disease, and an affected son was born. The granting of personal injury damage to the son implies that the error caused hampered existence while the alternative would have been not to be born at all. While such judgements are consistent with a social justice of compensation for damage caused by malpractice, they raise ethical questions of whether there is a "right not to be born". Conversely, persons afflicted by a genetic disease, but making the best of their handicapped lives, may thank their parents for not having gone to genetic counselling, because their alternative would have been not to exist. Such lawsuits should not be encouraged, as far as possible.

Justice, fairness and social equality principles should direct and prevail in all aspects of genetic counselling: its availability to all social, ethnic groups (enough appropriate counsellors), its initiation (free), its contents (not paternalistic), its options (the same for all social groups).

V.2 Moral, Religious and Philosophical Quandaries

Genetic hygiene: a hazardous concept. Eradicating diseases is a dream of mankind. Medicine has had already some limited victories against a few viral, bacterial, parasitic, environmental diseases. Many may think that, as the Human Genome Project develops, the increasing power to detect and predict genetically transmitted diseases, will allow their eradication by some "genetic hygiene". One should carefully distinguish between free decisions of individuals to take or not the risk of conceiving and ultimately the decision of bearing a genetically affected child, and between such actions or even recommendations on the part of society. The latter should be regarded as negative eugenics, a dangerous ideology which has not been accepted by Science. The former should be viewed as a private medical decision.

Eugenics concerns. Human applications of modern genetic technology do not have to lead inexorably to eugenic abuse. An essential distinction between genetics and eugenics is the importance ascribed to individual welfare rather than to society[29]. Only if genetic testing and counselling is not done as a private, confidential and free medical practice, with only the participation of genetic, psychology and ethic experts, is there a danger of social eugenics. Societies do not have to impose systematic or compulsory genetic screening to create the danger of eugenics. Indirect actions, such as limiting health care or life insurance to living patients with genetic disease, to couples about to reproduce or to families who have certain pathogenic genes, would amount to discriminatory measures leading to eugenics. Providing advantage to individuals who bring a "clean genetic map" of their own genome or of their children - such as lower insurance premium, certain jobs or other social bonuses - while eroding acceptance and care of genetically disabled, could be enough to raise the spectrum of eugenics[25, 30]. In fact, a simplistic vulgarization of the scientific achievements of human genetics, stressing genetic determinism or "genetic fatalism" [27] and neglecting the personal power of accepting, overcoming or compensating handicaps, would seed in the public's mind the seeds that already brought in this century eugenics and racial discriminations, under false scientific pretexts.

Objections to eugenics: scientifically there are no good and bad genes. Genes genecode proteins which have functions in the human body, its development and health. Genes are polymorphic, differing in some of their DNA sequence in different individuals according to heredity. There is no gene for cancer, or for muscular dystrophy or for Alzheimer disease, but some of the variated genes lose their function and cause or predispose to disease. Research has now established that these varieties of pathological genes are in fact often selected for some selective advantage due to certain life conditions or environment. Thus, in a certain region or human group, pathology-causing genes often show different types of mutations indicating that they do not result from one accident and a "founder" who would transmit it to all the affected patients. For example, in La Réunion, sufferers of limb-girdle muscular dystrophy have a number of different mutations in the same gene, indicating that

there must have been some selective advantage to have these changes[31]. In some cases, epidemiological and biochemical studies have revealed that the mutant gene, in heterozygous state, protected against other diseases: the sickle cell anemia mutations protected from malaria, the cystic fibrosis mutations protected from cholera, may be even the Tay-Sachs mutation protected from tuberculosis. In areas and times where these other diseases are no longer lethal, the mutations appear to us as bad genes, and their good side is forgotten. If it seems logical to use genetics to prevent birth of homozygous affected by disease, it would make no sense to eliminate the bad genes altogether. We do not know what other beneficial functions such pathologic genes may have, and certainly cannot predict what would happen to mankind were they eradicated. Even in cases where the mutations appear new and unlikely to have been selected, qualifying for being a "gene that went bad", the eventual advantage of eradicating the variant gene in heterozygotes should be weighed.

The human genetic heritage is imperfect. The frequency of carriers for pathogenic genes is high (1/20 for cystic fibrosis). With the number of genetic conditions, nobody could claim a 100% healthy genome. In the words of the Genethiq Group in Quebec [30]: *"All human beings are carriers of abnormal, variant genes which can cause some pathology or susceptibility to diseases; nobody can be qualified as genetically sane or genetically deficient. Discrimination on the basis of genetic make-up would make everyone a target. One has to conceive the individual human being in all its complexity and originality"*. These important thoughts point out well the moral attitude to fight eugenics or any attempt by society to make the individual responsible for "purifying" or correcting his genome to adhere to an illusory norm. The notion that the world and man are imperfect is also rooted in religions who care for the weak and diseased as a creature of God as opposed to "survival of the fittest". Bringing or not bringing a severely diseased child to this world should be an individual decision and not a way to straighten nature (the Jewish mystical tradition states that God made the world imperfect).

Human diversity precludes to define a normal prototype. Respect of human individuality also makes illusive the desire for a "perfect child" with no genetic defect. Eliminating or repairing some pathogenic genes, as justified as it may seem, must not be extended beyond the individual inability to cope with a severely diseased child. Society cannot define what is normal. The word "defect" presupposes that we have a perfect prototype, negating human diversity. Society cannot chose its own prototype, otherwise it may decide that being left-handed, having a certain character or a certain colour of eyes, hairs or skin, are genetic defects. Human diversity may be the secret of mankind's success. The possibilities offered by genetic testing to detect genes predisposing to mental illnesses, even including schizophrenia[31], have to be taken with great caution. Not only because of premature, erroneous or incomplete conclusions, but mainly because genetic traits do not necessarily equate to high heritability of a certain phenotype[27]. Using genetics to find a way to treat a phenotype is justified, eliminating a risk gene may be a useless and dangerous social exercise. *"Each individual has her or his individual heritage, including some special gifts and advantages and some disadvantages and risks* (...) *everyone of us is handicapped and challenged in one way or another."*[22] These mental genetic traits may insure human diversity, with its mix of artists, scientists, advocates of established order, as well as free-thinkers. Social environment and education make these genetic traits malleable, and this is certainly true for intelligence for which IQ is a poor measure[26, 27]. Every human being maybe entirely the product of his genes and entirely the product of the environment. Absolute free will and genetic fatalism[27] are two extreme views of human behavior, whose relative importance keep changing according to current philosophies, because both have part of the truth, but true life is a healthy balance between them, a synthesis of the two.

Scientism (not Science) can lead to genocide. If misunderstood, the much publicized achievements of the Human Genome Project and the growing recourse to genetic counselling have the inherent danger of nurturing a simplistic genetic fatalism, of the type which lead to the eugenic measures of sterilization for alcoholics and mentally deficient in several States in the United States of America and Scandinavia[25, 33]. Stefan Kuhl, in a recent book[34], researched the connection between these eugenics measures, which sounded based on a scientific genetic hygiene, and Hitler's "Mein Kampf" and the subsequent Nazi program of

eugenics, with sterilization and euthanasia for mentally retarded and chronic alcoholic. Another book by Lifton and Markusen[35] shows that the scientific prestige of the Nazi doctrine was based on the concept of applied biology which regarded racial revitalization as deriving from Darwinism's survival of the fittest (Francis Galton who invented eugenics in 1887 was Darwin's cousin). It shows how this concept served the objectives of the Nazi doctrine for cleaning out contaminating elements - first the mentally retarded and sick, later the Jews and other "inferior races".

With these and other recent historical precedents using pseudo-medical and psychiatric means to deny human rights, it is clear that great precaution must be taken not to make of science a scientism that ignores or overtakes the moral values of society. Preventive ethics is to foresee such possibilities of misunderstanding science. Genetic counsellors, as those who inform the public of the possibilities offered by the modern human genetic discoveries, have a major responsibility in presenting them in an absolute ethical context. Negative eugenics as well as positive eugenics (trivial selection or engineering of sex, eye colour, height or other enhancement genetics) are potential social calamities. Ignoring these social dangers could negate all the good which will come out of ethical medical genetic counselling for the individual.

VI. Conclusions

Genetic counselling, as practised today, adheres to the highest standards of the bioethics doctrine, respecting individual freedom, human rights, and cultural values. It provides an invaluable medical service and will increasingly allow patients in all countries to benefit from the most important scientific advances produced by the international Human Genome Project. A "preventive ethics" approach[15] implies to ensure that all these bioethical standards are maintained and to think ahead of the impacts of the new medical genetics on mankind. Preventive ethics implies to anticipate new ethical issues, with the aim of protecting the individual (born and to-be-born), his welfare, dignity and freedom, so that progress of scientific research does not infringe on them. Organizations, such as ELSI of the Human Genome Organization and the UNESCO IBC can play an immense role, not by imposing a universal order, but by harmonizing individual cultural values and science.

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ANNEX

INTERNATIONAL BIOETHICS COMMITTEE

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