Medical-ethical Guidelines for genetic investigations in humans

Preamble

The publication of these guidelines on pre- and postnatal genetic investigations is based on the following considerations:

Genetic examinations are constantly gaining in importance in medical research and practice. They are therefore being carried out in a growing range of indications. Thanks to genetic engineering it is possible to obtain increasingly more accurate information on genetic factors and hereditary defects in the persons investigated, and indirectly also of their relatives. Consultancy on genetic matters is thus greatly improved and broadened. In spite of genetic defects, persons seeking this advice can be protected against the consequences of serious illness, can have healthy children of their own or can be prepared, with adequate support, for the birth of handicapped children.

It is mainly the possibility of genetic engineering being used to investigate the key substance of heredity, desoxyribonucleic acid (DNA), that has aroused widespread concern in public opinion: it is feared that unnecessary or excessive genetic-diagnostic investigations could lead to discrimination against the individual. Also among doctors there is a need for more information on the conditions that should be fulfilled when arranging and carrying out genetic investigations.

The guidelines of the Swiss Academy of Medical Sciences are directed to the medical profession and can take into account only some of the problems associated with the medical application of genetic investigations. It is the task of the State to take appropriate measures to ensure that genetic investigations are used to the benefit of all those who need them for recognised medical indications, without putting at risk the individual's right of self-determination and without any misuse of the results of such investigations.

Genetic investigations must be undertaken under the responsibility of a physician who knows the indications for genetic tests and who is familiar with the informative value of the results obtained.

The cataloguing of hereditary diseases and handicaps in which genetic investigations could be helpful is not practicable, as this field of medecine is subject to constant change. Moreover, any such listing could give rise to undesirable preconceptions in society.

The comprehensive explanation and clarification of genetic investigation is becoming an increasingly important task for specialist medical associations and professional organisations, as well as for schools and the media.

Guidelines

1. Scope

These guidelines define the basic conditions for the procedures to be adopted by the physician in regard to medicogenetic investigations.

- They are restricted to investigations to detect or exclude hereditary factors responsible for disease.

- They are valid for all investigations carried out before or after birth and which allow conclusions to be drawn regarding such hereditary factors.

- They show how such investigations are to be carried out in the individual and in families, as well as for screening examinations.

2. Medical indications for genetic investigations

Genetic investigations are ethically justified if they serve the following purposes:

- Diagnosis and classification of an hereditary disease or handicap.

- Determination of a predisposition for an hereditary disease or handicap, with a view to appropriate planning for the life of the individual, and family planning.

- Detection of a predisposition for a particular disease at a time when symptoms are not yet evident, if effective measures can be taken to alleviate and prevent severe sequelae of the disease or if the result of the investigation is of immediate relevance for the planning for the life of the individual and to family planning.

- Counselling to individuals and couples concerning the risks to their children due to diseases or handicaps of genetic origin.

Such investigations are inappropriate if their aim is merely to determine the sex of the embryo or foetus or other factors that do not constitute a threat to health.

3. Recommendations for the performance of medical-genetic investigations

3.1 Conditions

Genetic investigations are permitted in persons legally of age, if they give their consent. In minors and legally incapacitated persons they may only be performed if the results are of immediate relevance for their health or for that of close blood relatives. Invasive prenatal investigations are to be undertaken only if there is a well-founded fear of a genetic risk.

3.2 Duty to inform

Patients with hereditary diseases or handicaps should be informed, in good time and in a proper way, of the existing possibilities for genetic investigation. The same applies for asymptomatic carriers of an assumed or proven pathological hereditary predisposition for a particular disease or handicap. If necessary, an appropriately specialised medical doctor should be consulted.

If for genetic reasons there is a possible need for the treatment of an individual's blood relative the doctor should take steps - with the consent of the person investigated or of his legal representative - to provide them with the relevant information (see Paragraph 3.7).

3.3 Genetic counselling in connection with the investigation

Genetic investigations must be accompanied by appropriate counselling before, during and after the investigation. Sufficient time must be allowed for the patient to consider the implications before the investigation is carried out. The advice to be given should comprise information that makes the significance and the consequences of a decision clear to the patient and enables him to fully understand this in the light of his own ethical concepts. It must not be given in a binding manner. The views of the doctor may only be expressed if the person seeking advice asks for them or if subsequent action to be taken do not conflict with the doctor's own conscience.

The information provided within the framework of this counselling must cover at least the following aspects: reason for, and nature and implications of the investigation and the possible risks associated with it, the possibility of an unsatisfactory or unexpected result, the significance of an abnormal finding and the measures to be taken to enable the patient to cope and live with his illness or handicap, information on self-help groups as well as alternative treatment. At the same time account must be taken of the patient's individual situation and relationships in regard to family, religion, education and psychological and social circumstances.

3.4 Obligatory consent and decision on disclosure of the results of the investigation

The decision to carry out, continue or stop the investigation rests exclusively with the patient, who will also decide whether and to what extent he wishes to be informed of, and to draw conclusions from the result of the investigation. The voluntary nature of participation in the investigation and the right not to be informed of the result must also be guaranteed for screening examinations.
In the case of persons incapable of judgment, the consent of their legal representative is required. Minors capable of judgment and legally incapacitated persons under guardianship have their own right of decision. They also determine whether the results of the investigation shall be disclosed to themselves or to their legal representatives.

The decision whether a prenatal genetic investigation should be carried out and what conclusions are to be drawn from the result belongs - within the framework of the legal dispositions - to the pregnant woman. It is desirable to involve the woman's partner in the decision-making process.

3.5 Long-term support of the person investigated

Persons subjected to genetic investigation must be supported and given access to long-term medical, psychiatric and social care, irrespective of any conclusions they may draw from the results of the investigation.

3.6 Quality assurance of the laboratory investigations

Genetic laboratory investigations must be undertaken by institutions that can show that their procedures and methods of working are impeccable and subject to external and internal quality control.

3.7 Professional secrecy and data protection

For results obtained in the course of genetic investigations, the same regulations governing professional medical secrecy and data protection apply as for other medical data.

The medical doctor may make the medico-genetic findings available to third parties only with the consent of the person investigated or of his legal representative, and only after the implications of such disclosure of information have been explained to them (see Paragraph 3.4)

3.8 Genetic investigations with a view to professional and non-professional activities

From the medical point of view, genetic investigations must not be carried out for the purpose of assessing the suitability of a person for certain activities or work, unless the investigation is performed in order to detect factors which, if present, would render a particular activity a considerable risk to the health of the individual or for other persons. Such investigations may be undertaken at the request of the person concerned or in accordance with a legal directive. The results are to be disclosed only to the person investigated.

3.9 Genetic investigations in relation to insurance

From the medical side, particular reservations are recommended when it can be seen that the results of a requested genetic investigation are to be used in connection with the taking out or the revision of an insurance policy. The results are to be communicated exclusively to the person investigated or his legal representative, after

the implications of the passing on of such information to third parties have been explained to them (see Paragraphs 3.4 and 3.5).

3.10 Research

The guidelines for research investigations in humans also apply for the collection and use of genetic data for scientific purposes. The results of genetic investigations carried out in the interests of a particular individual may also be used for genetic-epidemiological purposes without that person's direct consent, provided strict anonymity is observed. In the publication of the results of genetic research any possibility of identification with individuals must be avoided.

Commentary

Inherited diseases are often a great trial for the patients concerned and also for their families. For some conditions, medical help is today available which rarely leads to freedom from the symptoms (e.g. "cure" of hypothyroidism by continuous hormone substitution) but more often to alleviation, with prolonged life expectancy (diabetes, thalassaemia). However, in spite of effective but tiresome therapy other diseases still impose considerable restrictions on the patient (e.g. haemophilia). Many other hereditary diseases unfortunately still cannot be cured effectively despite intensive research efforts (e.g. myopathies). Here, too, the doctors have important tasks to fulfil, such as symptomatic treatment, adjustment of the patient's way of life, provision of mechanical aids and special training and counselling; many highly specialised self-help groups and organisations for the handicapped also provide valuable help. The contributions from legal disability insurance benefits provide the necessary financial support, as well as good, specialised counselling for the patients' families.

All those concerned need to be informed as accurately as possible about their illness. In order to be able to answer their many questions, a precise medical diagnosis is necessary. The available diagnostic armamentarium, which has become considerably more extensive and sophisticated over the past few decades, enables the doctor to collect the necessary data in increasingly better quality and with less trouble for the patient. As a result, his responsibility has also increased, because the answering of the patient's many questions on the future course of his illness calls for very precise knowledge of the particular disease and its genetic background; the same is true for the questions asked by the patient's blood relatives concerning risks for their children.

The families have the **right** to a thorough examination with the most modern methods available and to extensive specialist counselling following precise diagnosis of their case. On the other hand, **no one is obliged** to have diagnostic tests carried out on himself or his children: the **"right not to know"** must be fully respected.

These guidelines are intended to help doctors, in their everyday practice, to obtain an overwiew of the complex questions involved, in both the medical and non-medical fields, without great loss of time, in order to be able to avoid wrong decisions due to lack of knowledge on important aspects of the particular disease.

Far more intensive study of the specialist literature on the fundamentals of genetics and on individual technical, legal and ethical questions is essential for all those doctors who themselves wish to become actively involved in genetic investigation and counselling. All the other medical colleagues for whom such commitment is not possible may call on experienced specialists, either as counsellors or for specific tasks.

Prenatal diagnosis

Genetic counselling should today already begin before the birth of a possibly sick or handicapped child. This is possible because diagnostic methods are available that provide accurate information in the early stages of pregnancy. The vast majority of the prenatal tests that are carried out in fact give a **negative result** and thus enable the doctor to confirm the future parents' hope for a healthy child. This information of course concerns only the specific hereditary disease occurring in the particular family. On the other hand, it is obvious that no doctor will ever be in a position to give an overall "guarantee of a healthy baby", which opponents of these tests very often claim that doctors do; during the prenatal period, as generally is the rule in medicine, the **diagnosis** has to be based on a **specific medical question**, and the answer will be limited to this question.

If prenatal tests show, on the other hand, that a **foetus is affected by the hereditary disease being investigated**, then the doctor is faced with a number of new tasks:

- If the **disease can be treated effectively or can be cured after birth**, there is then ample time to make all the necessary preparations (for example: in the case of hereditary immune defects a cure is possible by means of a bone-marrow transplant; the chances of success are significantly better if the child is not exposed to infection at birth and if the treatment can be started early).

- If there is **no effective treatment** available the following possibilities have to be considered, in consultation with the parents concerned:

  - Decision **for** termination of the pregnancy in order to spare the child and the parents themselves the predictable suffering, in the sense of conscious family planning.
  - Decision **against** termination of the pregnancy, which is mostly taken for ideological reasons. In this case there are again two possibilities:

  1) **Avoidance of any prenatal test**, as the pregnancy will anyway be allowed to take its course. This option must therefore be discussed before the initiation of - in this case unnecessary - diagnostic measures (right not to know).

2) **No avoidance of prenatal tests**, but conscious acceptance of the possible future hereditary disease. The period of the pregnancy should be used for medical, psychological and material preparation for the birth and the life of a handicapped child, for which self-help groups and organisations for the handicapped provide great support and assistance.

All these decisions have to be made by the prospective parents. The doctor should help them in this, but only in an advisory manner, not making the decisions for them, and should draw their attention to all the available possibilities of support. If the stated or assumed wishes of the parents are not compatible with the personal moral views of the doctor, he may refer them to a colleague who does agree with parents' views.

Ethical scope

The aim of these guidelines is to define the limits of what is today considered as ethically justifiable; they take account of the fact that the current developments in this field are controversial.

The complex diagnostic methods available today often cause uncertainty or fear - mainly among people who are not involved - and as a result this sometimes leads to overall rejection. Partly responsible for this is insufficient knowledge, and partly **fear of misuse**. Great attention has been paid in these guidelines to the prevention of improper use of prenatal diagnoses (Example: the wish for termination of a pregnancy if the sex of a healthy foetus does not conform to the couple's wishes in regard to their "family planning").

There is very understandable and well-founded fear on the part of the handicapped and the organisations that care for them, of discrimination against these anyway already disadvantaged individuals, since they do not conform to today's image of the "ideal human being". The existence of this trend is indisputable. However, it has not come about as a result of the availability of the new technologies, but is rather just one aspect of the widespread materialistic attitude of society today and the belief, associated with it, that everything is possible. In recent times acceptance and understanding for the handicapped have not improved, but have rather tended to decline. There are justified fears that in

society accusations will be made against the parents of such children (e.g. children with trisomy 21).

Limitation, or even the prohibition of certain medical procedures that are known today would, however, do nothing to change these prejudices. As in the past, discrimination of the handicapped will still have to be combatted by providing information on their situation and by constant readiness to help them. Otherwise, it would not be ethically justified to try to deny other parents access to all the medical possibilities that are available today, provided they themselves feel able to accept moral responsibility for the consequences.

What is needed, therefore, is **more mutual tolerance and understanding** of the needs and aspirations of our fellow men, on the part of both the healthy and the sick and the handicapped.

Counselling

In these guidelines particular emphasis is placed on counselling. As is the case with diagnosis, only well informed doctors are in the position to provide well founded counselling. Doctors who are unable to meet this requirement will seek the advice of specialists or will refer their patients to such colleagues. This is equally true for technical questions relating to genetics, as also for specialised counselling centres of the disability insurance, for self-help groups and organisations for the handicapped or for legal and ethical/religious questions.

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LITERATURE

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