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SPECIAL FEATURE
Ethical issues connected with the privacy of persons found to have genetic defects

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ABSTRACT: Information on genetic defects constitutes knowledge of a very intimate nature and can change a person's life radically. Knowledge of one's own genetic defects can be a source of great anxiety even if this information is kept secret. When the confidentiality of this information is broken the person is often placed in a very vulnerable position and his/her basic rights are threatened. Very serious harm can be done to the person involved. On the other hand, genetic information pertaining to one person can have serious implications for others' welfare including spouses, children, and extended family members. How ought a genetic researcher/doctor deal with the information of genetic defects of his/her patients?

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Introduction

The methodology of modern medicine tends to be restricted to the empirical observations of diseases, the scientific research of their causes and the development of therapy to overcome or control these causes. A disease is often seen as a dysfunction of the human body and when this is discovered the disease is considered to be 'explained'. There is a tendency to forget the personal dimension of the disease. This aspect is surely a very important one. A disease, especially if it is serious, is primarily a personal reality. Persons experience a disease with their whole being. It often inflicts pain to patients who, in certain situations, may perceive it as a threat to their continued existence - not only to their lives, but also to their integrity as persons. The pain which a patient suffers is often not only physical but moral and psychological as well. This is especially so when the person feels abandoned by his or her loved ones who may project on the suffering sick person their fears of suffering and death[1].

1 Knowledge of one's own genetic defects ...

1.1 ... constitutes intimate knowledge:

The biological nature of every human is untouchable, in the sense that it is the constituent of the personal identity of the individual throughout the course of his history. Each person in his absolutely unique singularity, is constituted not only by his spirit, but by his body as well. Thus, in the body and through the body, one touches the person himself in his concrete reality. To respect the dignity of man consequently amounts to safeguarding this identity of the man ...[2]

Genetic defects may be seen as threats to one's body and therefore to one's personal identity and dignity. For this reason any information on genetic defects constitutes knowledge of a very intimate nature.

1.2 ... may radically change a person's life:

For example, if a twenty year old man knows that he has the Huntington's gene, he is suddenly faced with a stark reality which will accompany him for the rest of his life and have a strong influence on his life's important decisions. He knows that he will probably show symptoms for the disease in his forties and that he will die some time later. Will he get married? What will he tell his girlfriend?

1.3 ... may be the source of emotional stress:

Even if the person is "symptomatically" healthy, he or she could be suffering from the moral and psychological pain of a "future" disease through the mere knowledge of his or her genetic defects. And this knowledge would change his or her life radically.

Some studies indicate that as many as one in ten patients who test positive for the Huntington's disease mutation never make a full emotional recovery. In some cases they have been so severely depressed that they have had to be hospitalized. This is not surprising, given that there is currently no cure for the disease. Ponder and others[3] are concerned that positive tests for breast cancer could actually worsen a patient's chances of survival by triggering depression. For example, "do they become so frightened that they stop [breast] self-examination?" asks genetic counsellor Barbara Biesecker of the National Centre for Human Genome Research[4].

1.4 ... may stem from a false interpretation of genetic results

The 'discovery' of genes determining alcoholism, schizophrenia, manic depression and Alzheimer's disease have been peer-reviewed and published in leading journals. They were hailed in the media as breakthroughs - and then they were shown to be wrong. Benno Muller-Hill states " ... I have little faith in the notion that treatment of mental diseases will truly benefit from knowledge of the culprit genes and gene products. But I have no doubt that diagnosis will flourish. Cheap tests will be developed which will allow everyone to be tested for the variants of genes determining psychiatric ailments or psychic qualities outside the doctor's office..."[5]

2.3.3 Racial and Ethnic Discrimination

Few diseases are common enough in the general population to merit carrier screening. Many genetic diseases have an especially high incidence in a particular ethnic, racial, or religious group. Screening only members of such a group involves difficulty in determining who is a member of that group and risks charges of discrimination. Some of the screening programmes of sickle-cell screening carried out in the US in the early seventies were politically motivated and lacked sufficient expertise, confidentiality and provision for the counselling of subjects identified as positive. Many states passed laws requiring sickle testing at birth, at school entry or prior to marriage, laws leading to charges by blacks of attempted genocide. Positive individuals often suffered a decreased self-image. Positive children were often over-protected by parents. Individuals were sometimes discriminated against for purposes of marriage, employment or insurance[15].

2.3.4 Other Forms of Stigmatization

A revealing study of sickle screening programme was conducted in Orchomeno, a Greek village where marriages were frequently arranged by parents, a conceivably ideal arrangement to take into account genetic knowledge. Nevertheless carriers were stigmatized as undesirable marriage partners, not only for other carriers, but for everyone[16].

3 Combating Genetic Discrimination:

3.1 Education

Education of medical practitioners and the general public on genetics in order to eliminate discrimination based on fears and prejudices which are without foundation due to which some of the genetic discrimination occurs. For example, many people do not understand that a pre-symptomatic diagnosis showing the presence of a genetic defect is not the diagnosis of a disease and therefore should not be considered as such. Education must include efforts to promote values in Society which counter racism and promote human rights for all - particularly those suffering from some genetic defect[17].

3.2 Legislation to protect confidentiality

Laws are needed to protect confidentiality of genetic information. The price paid for services of omnipotent computers which store and process all sorts of data about individual persons and their affairs is the forfeiture of privacy. Medical records, like banking transactions and assets, purchases by credit cards, tax returns, are all vulnerable to inadvertent disclosure or investigation by public authorities[18].

3.3 Legislation to protect the genetically disadvantaged

Dr. Mark A. Rothstien, a legal expert, asserts that as the Human Genome Project progresses, the possibility of disclosure of confidential genetic information, coercive genetic screening, invasions of privacy, or genetic discrimination will arise. Legislators must be vigilant to ensure that the legal system, medical ethics, corporate culture and public policy definitely reject such incursions"[19].

The 1996 Treaty on Bioethics of the Council of Europe bans genetic testing for pre-symptomatic diagnosis except for medical or research purposes. This means that gene testing for insurance and employment purposes is banned[20].

3.4 Confidentiality in Cases of Unknown Paternity

Some very embarrassing, or even catastrophic consequences may follow the testing of a man's DNA to determine paternity ... When informed of the non-paternity of their husbands, some women are reported to have maintained a fretful silence rather than face the adverse consequences for their families. A woman may be put under a torturing strain as she decides whether to reveal the shame of infidelity or the shame of being a victim of rape to her husband. Depending on her own character as well as upon social factors, the DNA indetification provides the exact information on which a difficult choice may be made[21].

4 Implications for others

If the genetic information is initially known only to the diagnostician, who is then entitled to know it? The person in question? Is anyone else entitled to know? And if yes, who has the obligation of communicating this to them? The new practice of genetically-directed medicine brings more complexity to the answering of inescapable questions about diagnosis and prognosis. The doctor is challenged, not only to disclose true information to the patient for the sake of both the patient's health and the rule of honesty; he or she must also exercise prudence and discernment concerning how much of the known information to share, what to withhold, and with whom to share it. This caution is in order for two reasons. First, the doctor's primary concern for the patient's "best interest" may mandate a limitation on "the whole truth and nothing but the truth". Second, the serious concern for other members of the patient's family may impose restrictions. In other words, the medical dictum "Do no harm" may collide with the rule of telling all the facts of a case. However, this collision does not render the physician dumb. Words of information and counsel must be spoken despite the dilemma[22].

4.1 Some Guidelines:[23]

4.1.1 Informing the Patient:

The doctor is ethically obliged to give sufficient information to the patient so that s/he is in a position to give or to withhold consent in full awareness of the facts. The information given must be understandable and include the severity of the illness, the advantage and the risks of the proposed intervention, and the consequences in refusing it. On the other hand, each individual has the right not to be compelled to know about his/her own genetic defects.

4.1.2 Informing Others:

The communication of knowledge of genetic defects without the free and the informed consent of the individual/s concerned amounts to the breaking of a secret. The concentric circles of confidentiality need to be defined with respect to the effects on other members

of an extended family or offspring conceived out of wedlock. At times there may be legitimate cases in which the welfare of these persons may imply a strong obligation for the person with a serious gene defect to disclose his situation to these persons for their own good. For example, when a husband or wife discover that they have a gene defect which is transmissible to their own children, he/she has an obligation to tell his/her spouse, and their children, since these must also be involved in responsible decisions regarding their future progeny[24].

In those cases where the person with the genetic defect does not want to know his/her condition, it may be right for the medical practitioner to communicate the result with his/her next of kin and to explain the implications of these results on the family, after obtaining permission from the patient. Retrospective screening for other members of the family may be recommended if this would lead to their greater good. Care must be taken in all communications on these matters that they be done in a very delicate and sensitive way, taking into account the greater good of all involved.

4.1.3 Genetic Screening

Some push for legally mandated genetic screening. Indeed, in the US neonatal screening of certain diseases is mandatory. Arguments made for mandatory screening are higher compliance rates, lower cost, timely execution and facilitation of record-keeping of incidences and outcomes. However, this often goes against the informed consent and privacy of people and endangers their basic rights. Voluntary screening is more in line with informed consent and respects the freedom of the individual. It allows each person to face his/her own responsibility. Voluntary screening may also reduce the likelihood of adverse psychological effects if screening is preceded by appropriate education about the benefits and risks of testing and if consent for testing is truly informed[25].

Conclusion

It is shortsighted to view ethical recommendations as being in some way contrary to scientific research and progress. Real progress in medicine is one which helps individuals and humanity as a whole. Ethical considerations in medicine ensure that human dignity is protected and that medicine is a tool which fights disease and advances the integral good of all involved. Ensuring privacy in medicine, and in a special way ensuring it in genetically related diseases, is vital for this reason.

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Commentary on:

Ethical issues connected with the privacy of persons found to have genetic defects

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Fr. Edgar Busuttil's article highlights the ethical issues associated with some aspects of genetic screening and provides useful guidelines for those involved in handling genetic information. The guidelines provided stress the importance of obtaining the patient's consent before disclosure of such information to third parties and are consonant with the guidelines on informed consent recently issued by Malta's Bioethics Consultative Committee[1].

In their relationships with patients, health care professionals assume ethical obligations: to respect their autonomy, to act in their best interests (beneficence), to avoid doing them harm (non-maleficence) and to act with justice. These ethical principles, as outlined by Beauchamp and Childress[2], make it possible to ethically evaluate a vast array of clinical situations. The main ethical issues which are likely to arise in handling genetic information are those of confidentiality and consent, both of which emanate from the principle of respect for the patient's autonomy.

Genetic information is information of a very intimate nature and should, therefore, be handled with due discretion and sensitivity. That is because such information is not only a potential source of anxiety for the patient, but also because it may have serious implications for other members of the patient's family. From the viewpoint of professional ethics, a doctor's obligation to safeguard confidentiality in respect of such information stems from the general Hippocratic obligation not to divulge clinical information to third parties without the patient's consent.

Though universally acknowledged as an important ethical principle in the practice of Medicine, confidentiality is by no means an absolute obligation. There may be instances when this principle may conflict with others forcing the doctor to prioritise his ethical obligations. Faced with such a dilemma, the doctor may rightly decide that the patient's right to confidentiality should defer to the rights of other persons. Exceptions such as these must always be justified on the principle of non-maleficence and never by appeals to medical paternalism which, in this day and age, is regarded as an anachronism[3]. Respect for the patient's autonomy would still dictate that in such cases, every effort be made to obtain the patient's permission before genetic information is disclosed to others.

There is increasing concern that some insurance companies might, in future, insist on compulsory genetic testing for certain diseases before undertaking to provide health insurance cover. Information about genetic defects which is already known to a person before applying for insurance, would be expected to be revealed along with other relevant information of a medical nature[4]. It is likely, however, that demands for compulsory screening as a precondition for insurance will be resisted. There is obviously a need for guidelines to be drawn up to protect the interests of insurance applicants and to ensure ethical practice on the part of insurance companies.

With the increasing identification of genetic predisposition to certain diseases, there is also concern that prospective employees may, in future, be required to

undergo genetic screening in order to identify those most at risk of developing adverse reactions in their place of work. Compulsory genetic screening not only goes against the grain of personal autonomy but also violates the basic right to privacy. Here again, the need is felt for guidelines to bring appropriate control to this area[5]. Legislation may be necessary to protect the confidentiality of stored clinical information about individuals from inadvertent disclosure and access by unauthorized personnel. It is sincerely hoped that the government will deal with these important matters without delay.

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It is now universally agreed that good medical practice, and particularly that involving genetics, should respect the privacy of the individual and the confidentiality of data. There is increasing awareness of the importance of paying due attention to the psychological and social implications of disease conditions and of involving patients as much as possible in decision making through informed consent. The general medical ethical principles apply also to genetics, which, in addition, involve some situations that may present special ethical problems.

The main problems stem from the fact that genetic diseases affect families rather than individuals. In addition to the doctor-patient relationship, third parties are almost invariably involved. Each of the three parties has its own set of responsibilities, rights and limitations, which in fact constitute most of the ethical aspects of genetics. Clearly the ethical issues would be quite complex and it is important to distinguish clearly the ethical implications relating to the doctor, the affected individual and third parties.

Respect for the person - Confidentiality and informed consent

Medical practitioners are bound to maintain confidentiality and ensure their patients have been well informed prior to undertaking genetic investigations. Genetic disorders often cause great psychological distress because they directly affect the person's future prospects regarding marriage, childbearing, employment and other social aspects of life. Attention to these psychosocial implications is an integral part of genetic counselling that should precede most genetic diagnostic tests. In fact the counselling procedure prior to pre-symptomatic testing for genetic diseases such as Huntington's disease routinely involves a psychologist as well as a geneticist and extends over two or three sessions. These procedures ensure respect for the person who is undertaking genetic tests.

To whom should genetic information be given?

One of the main problems in genetics concerns giving information to third parties, whether relatives at risk, commercial insurance or employment companies. As a general rule, all information regarding the results of any investigation belong exclusively to the individual concerned and cannot be divulged to relatives or other third parties without his or her explicit consent.

Should information be given to relatives?

Problems arise if the genetic condition also imposes a risk on other family members. Is it the doctor's duty to inform relatives at risk? This is a very complex situation that cannot be generalized upon. At one extreme the patient may request that the condition of his test result is not divulged, even to close relatives. Such a request must be strictly honoured. However, in most cases individuals are willing to co-operate and to inform other members of their families including prospective husband/wife that they could be at risk. This is the duty of the individuals. The doctor is responsible for advising the individuals that their families might be at risk and that they can be offered genetic counselling and testing if they wish. In most cases it would be definitely unethical for a doctor to take it upon himself or herself to identify and inform relatives at risk. The main objection would be that this would be infringing on the privacy of the relatives by obtaining information about them without their prior consent, by telling them about a situation that they might have preferred not to know about. It might even cause undue anxiety, disputes and disruption within the family.

In contrast to the above are certain genetic conditions which impose a serious and immediate health risk. An example is familial ademomatous polyposis which predisposes to colon cancer, a disease which, in this particular situation, is entirely preventable by timely investigation. In such a situation the doctor, usually with the co-operation of relatives, can certainly put aside some of the classical constraints and go into great lengths to identify relatives at risk who could be saved from a life-threatening situation.

Should individuals with pre-mutations be informed?

A very delicate situation arises in the case of pre-mutations, which may be found in a certain type of genetic mutation known as trinucleotide repeat expansion. The most important example is familial mental retardation commonly known as the fragile X

syndrome. A pre-mutation is a gene which is in the process of undergoing expansion to a full, disease-causing mutation. Carriers of a pre-mutation would not be at risk of developing the disease; their children might also be spared but the disease might appear in the next and future generations. Should such individuals be informed? Undoubtedly this will be a source of great anxiety. These individuals would feel responsible for such a serious condition appearing in future generations. However, any person who wants to have a particular test is entitled to know the whole truth, however painful that might be. The problems that are likely to arise should be anticipated and dealt with in the counselling which leads to informed consent. Nevertheless the situation is a very delicate one that needs to be handled with great tact and understanding. A detailed discussion of possible approaches to the problem is beyond the scope of this brief commentary.

The problem of obligate carriers

A very difficult situation is that of obligate carriers. Some individual at risk for a dominant disease such as Huntington's disease might prefer not to know whether they carry the mutant gene or not. They might prefer to live in uncertainty which also provides a measure of hope. The problem arises if their children want to be tested. A positive test in one of their sons or daughters would make them obligate carriers. This would be clearly unethical, as it would go against their expressed wish not to know. This situation creates one of the most difficult ethical dilemmas that a geneticist can encounter.

Information to employers and insurance agencies

The problem of providing genetic information to employers or insurance agencies has been strongly debated. A person who gives consent to a medical examination for insurance or employment purposes is automatically giving consent to the results being revealed to the third party. A doctor cannot make a false declaration! In this case the ethical issues involve employers and insurance companies. The availability of genetic test results to such agencies would certainly be disadvantageous and discriminatory to affected individuals. Geneticists, doctors and patients may insist on a code of ethics setting guidelines regarding what information commercial agencies can reasonably request. The onus of responsibility to reach a consensus regarding what information can reasonably be asked for to avoid discrimination lies squarely on these agencies.

Paternity testing

In cases of paternity disputes, only the individuals who consent to participate in the testing procedure have a right to know the test result. The problems of embarrassment, shame and possibly even torturing strain do not arise because of genetic tests that may have been performed, but from the social situations which have led to the need for paternity testing. These are social problems. The moral and ethical issues, which may arise, are ones of inter-personal relationships. Rather than being the cause of embarrassment a paternity test result will resolve uncertainty and may help in re-establishing a degree of stability for the disputing parties and the child.

Genetic intervention

Perhaps the most important ethical issue relating to individuals with genetic disease is the extent to which medico-scientific advances should be made freely available for use on individuals. Interventions that involve procreation would raise the most serious ethical problems because they involve the basic values of society, the respect for human dignity and the rights of the embryo or foetus, which in this case is the third party without a voice to consent. It is already evident that interventions involving procreation would require a major re-evaluation of the classical social and legal concepts of paternity and maternity and families. Undoubtedly new major ethical problems are bound to crop up as the advance of genetics continues in its exponential trend. Undoubtedly they will raise controversy and dispute until the problems are resolved as they have been in the past. However, the big ethical problems will not stem from the human genome project or problems of confidentiality. The major ethical issue that is likely to become of crucial importance in the near future will centre on the modification or regulation of an unborn person's genetic constitution by genetic manipulation of the germ cells. The techniques of cloning and transgenic manipulation are already lurking in the background.