UNIT 4 GENETICS AND HUMAN ISSUES

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4.3 Summary

Further Reading and References

Sample Questions

Learning Objectives

Once you have studied this unit, you should be able to:

- describe how ethical issues in human genetics have emerged and why it is important to take care of them;
- understand about the various ethical principles and their importance; and
- discuss the emerging ethical, legal, social concerns in genetic testing and research.
4.1 INTRODUCTION

Hereditary conditions affect millions of families throughout the world. About 5% of all pregnancies result in the birth of a child with a significant genetic disorder, congenital malformation or disability. The burden of genetic disorders and birth defects is yet to be recognised. It has been estimated that there is a high prevalence of genetic disorders in India with about 495,000 infants with congenital malformations, 390,000 with G6PD deficiency, 21,400 with Down syndrome, 9000 with Beta thalassaemia, 5,200 with sickle cell disease and 9760 with amino acid disorders born each year (Verma and Bijarnia, 2002). A high prevalence of inborn errors of metabolism of 1 in 1000 live births has been reported from India (Rama Devi and Naushad, 2004). It is estimated that 8000-10000 children are born with thalassaemia every year and there are about 20 million carriers of b-thalassaemia in the country (Mohanty et al, 2002). Most non-infectious diseases, may have a genetic component. Therefore, continued efforts are required to develop effective treatments and make them available in India for diseases that are important to the health of communities as well as individuals and families.

4.1.1 Human Genome Project (HGP)

The human genome project (HGP) was an international research effort, with the aim to sequence the entire human DNA and also determine the location of all identified genes which was carried out from the year 1990 to 2003. This project was coordinated by the U.S. Department of Energy and the National Institutes of Health. The project was initiated with a hope to develop new ways to diagnose, treat, and someday prevent the thousands of disorders that affect us.

**Goals of Human Genome Project**

- **identify** all the approximately 20,000-25,000 genes in human DNA.
- **determine** the sequences of the 3 billion chemical base pairs that make up human DNA.
- **store** this information in databases.
- **improve** tools for data analysis.
- **transfer** related technologies to the private sector.
- **address** the ethical, legal, and social issues (ELSI) that may arise from the project.

Upon completion of this project enormous amount of genetic information has been made available. All adults have a right to know their genetic makeup and also implications for the health of their potential to next generation. HGP has raised some concerns in regard to issues such as access to genetic information, diagnostic services, privacy and confidentiality, disclosure to family members, freedom of reproductive choices, misuse of genetic information, stigmatization/discrimination of individuals based on their genetic makeup etc. Therefore though there is potential for advances through better diagnostic and preventive methods however on the other hand this knowledge has increased concerns about genetics in the public. It is therefore important to improve public knowledge and create better understanding about genetics. To remove the fear from the minds of people as well as help to protect families with genetic disabilities, it is important to understand societal and the ethical issues. The following are some examples of what could be the implications of genetic knowledge to various people:
- individuals and families – whether to participate in testing, with whom to share the results, implications for individual and other family members;
- health professionals – when to offer genetic testing, obtaining informed consent, how to ensure quality, how to interpret results, counsel patients, whom to disclose information;
- employers, insurers, the courts, other social institutions – the value of genetic information to the decision they must make about individuals;
- governments – how to regulate the use of genetic tests and information they provide and how to provide access to testing and counselling services for society; and
- society – how to improve public understanding of science and its social implications and increase participation of the public in science policy making.

The scientific community should address the above mentioned questions before use of this knowledge could be considered as ethically valid. As the challenges are conquered we will begin to harness genomics for the benefit of our populations (Hardy, 2008, Seguin, 2008).

4.1.2 What is Ethics?

Ethics, as a field in philosophy or religion, is concerned with systematic reflection on the moral life and its conflicts. “Ethics” is a generic term for various ways of understanding and examining the moral life and for resolving ethical problems (Beauchamp and Childress, 1994). Biomedical ethics (or bioethics) is an interdisciplinary field for the systematic study of ethical issues that arise in research, medicine and society. Medical ethics is a system of moral principles that apply values and judgments to the practice of medicine or its practical application in clinical settings. To be ethically correct these principles should be based on the community that is served and in view of their religious, social or cultural value systems so as to adequately protect their rights, welfare and safety. Under the principles of ethics there is an obligation to confer benefits, to prevent and remove harms, and to weigh and balance the possible goods against possible harms of an action.

4.1.3 Ethical Legal and Social Issues (ELSI) in Genetics

As you know during the last 1-2 decades there has been explosion in knowledge in human genetics, and several new technologies have been developed for example gene therapy, genetic engineering, stem cell therapy, cloning, prenatal diagnosis, preimplantation diagnosis, creating designer babies, genetic screening etc. In addition new issues have also emerged, for example those relating to patenting of DNA sequences, Intellectual Property Rights (IPR) issues and potential for bio-terrorism. In this rapidly evolving field there is a need to continuously monitor developments and respond to emerging ethical issues promptly and judiciously since improper use of these can have far reaching implications for human race.

You may be surprised to know that in no other area of biomedical research there has been a greater concern for ethical issues than in the field of human genetics. With the successful completion of Human Genome sequencing in April, 2003, number of guidelines were laid identifying the ethical, legal and social issues and measures that may be taken for improving awareness and understanding of human genetic disorders.
The integration of genetic technologies into the public health raises significant challenges. How can we assure the quality and accuracy? What impact will these new technologies have on existing programs? Issues of autonomy, privacy, confidentiality will require special consideration. Will it be possible to obtain fully informed consent from large populations? How to protect privacy in genetic registries and databases? How can group stigmatization and discrimination be prevented? None of these questions have easy answers, but they raise important challenges for the community. These are the various ethical, legal and social issues that address the societal concerns emerging from the new genomic information and technologies.

4.2 GENETIC TESTING AND RESEARCH

4.2.1 General Concerns

It is important for you to note that in human genetics the concerns may not only be to the individual but also the family, community or society from which s/he has been drawn. Kindly note some of these concerns.

What are the Concerns?

- Genetic information is sensitive and private and needs adequate protection. In genetic research the harm may not only be physical, but also psychosocial which may produce anxiety and depression or damage familial relationship.
- There is a likelihood of social stigmatization and discrimination in schooling, employment, health and general insurance, which requires much great care.
- When involving participants in research it is important to obtain written informed consent and maintain confidentiality of their personal or other details or data collected from them.
- There is great importance of spoken word in medical genetics, since genetic counselling is akin to therapy in other fields. Which means that the ‘word’ is equivalent to drug/intervention in other fields of medicine. Appropriate communication skills are necessary for genetic counselling.
- Genetic manipulations have consequences for the future, some of which are unknown and long term. Hence, greater care towards potential dangers is necessary.
- Any biomedical research conducted by student, or faculty on human participants should be reviewed by an independent committee known as the “Institutional Ethics Committees” or IEC to ensure protection of participants in research.

4.2.2 Ethical Principles in Genetics Research

There are three main ethical principles for all kinds of biomedical research including genetics research in human participants:

4.2.2.1 Autonomy or Respect for Persons

This principle refers to respecting the self-determination of individuals and protecting those persons with diminished autonomy. Informed consent is one
way of protecting this principle i.e., rights and voluntary decisions of individuals. For all biomedical research the informed consent of the individual, or legal guardian should be obtained. It protects the individual’s freedom of choice to voluntarily agree/disagree to participate in research or give samples for genetic testing. Adequate information about the testing is given in a simple and easily understandable language in a document known as the Informed Consent Form with Participant/Patient Information Sheet.

4.2.2.2 Beneficence and Non Maleficence

In simple terms it just means doing good and protection from harm. Beneficence involves giving importance to the welfare of persons and providing maximum benefits. On the other hand Non-maleficence refers to preventing from harm or trying to reduce harm and is derived from the traditional medical norm of “do no harm”. The principle of beneficence involves giving highest loyalty to the welfare of people and families with the goal to improve the health of population.

4.2.2.3 Justice

This principle requires treating persons with fairness. It involves distributing benefits and burdens of health care as fairly as possible in society. Therefore persons should be treated fairly, giving them what they deserve, or are entitled to. Distributing the benefits and the burdens of health care ought to be governed by ethically justified rules such as: to each according to need, to each according to an equal share or opportunity, etc.

Table 4.1: Examples highlighting use of ethical principles in genetic testing

- Involve voluntary approach in testing and treatment; avoid coercion (force) by society, or health professionals (autonomy).
- Respect for human diversity and for those whose views are in the minority and respect for people’s basic intelligence, regardless of their knowledge (autonomy).
- Education about genetics for the public, medical and other health professionals, teachers, and other persons (beneficence).
- Protecting privacy & confidentiality (autonomy)
- Prevention of unfair discrimination in employment, insurance, or schooling based on genetic information (non-maleficence).
- Teamwork with other professionals through a network of referrals to provide better tests, care and treatment (beneficence).
- Timely provision of indicated services or follow-up treatment (non-maleficence).
- Refraining from providing tests or procedures not medically required (non-maleficence).
- Providing good quality of services, including lab procedures (non-maleficence).
- Sharing results as well as benefits of research with communities (Beneficence, justice).
- Equitable selection of participants from community (justice).
- Protection of poor, vulnerable groups (justice).
4.2.3 Specific Ethical Issues in Genetic Studies

4.2.3.1 Informed Consent

A well informed consent and understood consent is essential part of all genetic testing. The central issue of human research in respect to the individual’s autonomy is achieved by obtaining “Informed consent” from adult individuals who are capable of giving a valid informed consent after they have been provided with complete information regarding the condition and the tests. Those persons who are tested are entitled to receive all information in a way that they can understand what is proposed to be done, they must be made aware of any risk, they must be given time to decide whether or not they would like to participate or withdraw from genetics testing/screening. In addition to this the details about the disorder to be screened and its inheritance pattern, reliability of the screening test and what will be done with the samples should also be explained. Information about the implications of a positive screening test (abnormal) should also be explained. In the case of an individual who is not capable of giving informed consent (for example individuals such as children, mentally challenged persons or prisoners etc), the consent of a legal guardian should be taken. Adequate information about the research given in a simple and easily understandable language in a document known as the Informed Consent Form with Participant/ Patient Information Sheet. Table 2 gives the details of elements that should be given in an informed consent form/ patient information sheet (ICMR Guidelines, 2006).

Table 4.2: Elements of Informed Consent Form

<table>
<thead>
<tr>
<th></th>
<th>Nature and purpose of study stating it as research.</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>Duration of participation with number of participants.</td>
</tr>
<tr>
<td>3</td>
<td>Procedures to be followed.</td>
</tr>
<tr>
<td>4</td>
<td>Investigations, if any, to be performed.</td>
</tr>
<tr>
<td>5</td>
<td>Foreseeable risks and discomforts adequately described and whether project involves more than minimal risk.</td>
</tr>
<tr>
<td>6</td>
<td>Benefits to participant, community or medical profession as may be applicable.</td>
</tr>
<tr>
<td>7</td>
<td>Policy on compensation.</td>
</tr>
<tr>
<td>8</td>
<td>Availability of medical treatment for such injuries or risk management.</td>
</tr>
<tr>
<td>9</td>
<td>Alternative treatments if available.</td>
</tr>
<tr>
<td>10</td>
<td>Steps taken for ensuring confidentiality.</td>
</tr>
<tr>
<td>11</td>
<td>No loss of benefits on withdrawal (from participation).</td>
</tr>
<tr>
<td>12</td>
<td>Benefit sharing in the event of commercialization.</td>
</tr>
<tr>
<td>13</td>
<td>Contact details of PI or local PI/Co-PI in multicentric studies for asking more information related to the research or in case of injury.</td>
</tr>
<tr>
<td>14</td>
<td>Contact details of Chairman of the IEC for appeal against violation of rights.</td>
</tr>
<tr>
<td>15</td>
<td>Voluntary participation.</td>
</tr>
<tr>
<td>16</td>
<td>If test for genetics and HIV is to be done, counseling for consent for testing must be given as per national guidelines.</td>
</tr>
<tr>
<td>17</td>
<td>Storage period of biological sample and related data with choice offered to participant regarding future use of sample, refusal for storage and receipt of its results.</td>
</tr>
</tbody>
</table>
A copy of the participant/patient information sheet should be given to the participant for her/his record. When the written consent (signature or thumb impression) is not possible due to sensitive nature of the project or the participant is unable to write, then oral consent can be taken after ensuring its documentation by an unrelated witness. However, the approval for oral consent has to be taken from an ethics committee beforehand. When human biological material or samples are collected, whether in a research or clinical setting, it is appropriate to ask persons for their consent to future storage and use of their samples.

### 4.2.3.2 Vulnerable Participants

Vulnerable individuals are those persons who are unable to give valid and understood consent for e.g., minors, mentally disadvantaged, prisoners, institutionalised individuals, terminally ill, students, subordinates and people who do not speak the language etc. Further there are influences of culture or customs which can make certain communities vulnerable. Even adult women may be incapable of giving informed consent without consulting family members like mother in law or husband. In community research where even if consent of the community leaders or head of family is taken, individual consent of every person is a must. In telling about test results (e.g., carrier status for a recessive disorder; which parent carries a balanced autosomal translocation that has caused a disorder in their child; incidental discovery of non-paternity) and in assisting couples to reach reproductive decisions, professionals should be careful to protect the interests of those who may be vulnerable to harm from a hostile environment. Similarly children, persons of diminished mental capacity, and some other groups may not be in a position to make a decision due to their limited capacities. Such people may be vulnerable to harm because of their position in society and need protection from any potential adverse effects of genetic testing. Minority groups are entitled to respect, to their beliefs, customs or way of thinking even if the medical geneticist disagrees with these views. They should be treated equally with persons whose views are in the majority. They should be provided with due information and be given opportunity to take their own decisions.

### 4.2.3.3 Genetic Counselling

Every individual should be provided with complete information and opportunity to make choices regarding one’s health. This is important to the person’s integrity and contributes to psychological well-being. All genetic testing should be accompanied with pre-test counselling (to tell about the nature of tests, what is expected etc) as well as a post test counselling (to explain the results of the testing and its implications). Pretest and post test counseling is an integral part of prenatal diagnosis (Kabra, 2003). In order to help a person to make a valid choice, he/she should be provided with more than one alternative. Only those persons who are qualified and experienced in communicating the meaning of genetic information should undertake genetic counselling.

<table>
<thead>
<tr>
<th>Purpose of genetic counselling</th>
</tr>
</thead>
<tbody>
<tr>
<td>• provide detailed information about the genetic disease in question.</td>
</tr>
<tr>
<td>• help individuals/couples understand options and present state of medical knowledge so they can make informed decisions.</td>
</tr>
<tr>
<td>• help individuals/couples adjust to and cope with their genetic problems.</td>
</tr>
<tr>
<td>• the removal or lessening of guilt or anxiety or fear that they may have.</td>
</tr>
<tr>
<td>• helping individuals/couples achieve their parenting goals.</td>
</tr>
</tbody>
</table>
4.2.3.4 Risks and Benefits

Any research procedure that exposes a person to any risk should be done only if there is proper justification for doing so and there should be expected benefits of the research. All expected or potential risks as well as benefits should be discussed in detail before participation of the person. Also it is important for you to note that in genetic research, the primary risk may not be physical but rather may be psychosocial and can have effects or implications on other members of the family as well. Genetic testing therefore should be offered when the probability of harm is less and the expected benefits are more. Adequate counseling should be given to participants on the meaning of genetic information they receive.

4.2.3.5 Pedigree Studies

Pedigrees involve obtaining history of other members of the family of the patient or the proband under investigation. It is an important tool in genetics however it may reveal information about the likelihood of other members of the family being either carriers of genetic deseases or being affected by the disease. Special privacy and confidentiality concerns arise in genetic family studies because of relationship between the participants. It should be kept in mind that within families each person is an individual who has the right to keep the information about himself or herself confidential. Family members are not entitled to know each other’s diagnosis. Other members are secondary subjects and they are not entitled to know about the genetic diseases of other members without appropriate informed consent. Before revealing medical or personal information about individuals to other family members, investigator must obtain consent of the individual to do so.

To explain this further I will give you an example:

In view of the cultural background of our country where woman is still a vulnerable and exploited participant, revealing information to the husband that his wife is the carrier of balanced chromosomal translocation (leading to recurrent abortions or a genetic syndrome in her child) or that she is a carrier of a single gene causing ‘X’ linked or recessive disease, may lead to grounds for a divorce despite the fact that the husband himself is a carrier of the autosomal recessive disorder. While general principles of counseling require presence of both the spouses, necessary care must be taken not to end up in breaking the families. In view of above concerns appropriate caution may be exercised. We must understand that revealing who else in the family has agreed to participate may lead to breach of confidentiality. Also if there is a patient, out of personal interest s/he may put undue pressure on relatives to enroll in the study which may not be acceptable to other members.

4.2.3.6 Privacy and Confidentiality

Secure safeguards should be in place for protecting private ‘identifying information’ of participants and maintaining confidentiality of research data. Identifying information comprises of all details like name, date of birth, contact details, and other personal information that can be used to identify an individual and therefore needs to be protected. Adequate care should be taken in keeping genetic records, however participants should be told beforehand regarding the limits to safeguard confidentiality and of the anticipated consequences of breach of confidentiality. If the result of the research is of benefit to the health of the
participant, then they should be communicated. Once study is over genetic data should be coded or delinked with clinical details to maintain confidentiality (anonymised). Adequate measures should be in place to restrict access of records to unauthorised persons. It needs to be emphasized that consent for screening or a subsequent confirmatory test does not imply consent to any specific treatment or termination of the pregnancy. Specific consent is required from the affected patient to share his/her genetic information with family members who may be benefited from it.

4.2.3.7 Inducement vs Compensation

Individuals who participate in research may need to be duly compensated for their participation. Provisions should be made for adequate compensation to reimburse extra travel expenses, loss of wages, compensation and medical care in case of any injury. However the amounts should be reasonable and should not become undue inducement, i.e., so huge that it becomes the only reason for participation as an extra income and motivation factor for a person to give samples for research. Inducement can be in cash as well as in kind. Making promises for extra services or facilities may induce a person to agree to participate in testing even if it is not of any direct benefit to the person. Such promises for extra payments in cash or kind and the amount of compensation must be seen reviewed and approved by the ethics committees before it is provided to individuals. In case of any injury happens due to participation of a persons in research it is the obligation of the researcher/physician to provide adequate treatment or compensation to cover the extra costs incurred due to this.

4.2.3.8 Genetic Screening

Genetic screening implies search in population or individuals who have, or are susceptible to have a serious genetic disease. It also includes persons who, though not at risk themselves, are carriers and therefore at risk of having children with the particular genetic disease. It is essential that screening must be done with specific purpose using validated tests. Genetic screening should be done for conditions if it can be ensured that a suitable treatment/management of the disease is possible. Depending on nature of the genetic defect that is identified and its pattern of inheritance, siblings and other blood relations as well as existing and future generation/offspring may be affected. Screening of newborns is permissible to detect those genetic diseases like phenylketonuria where serious effects of the disease could be prevented by early intervention such as special diet or treatment. It should not be done when there is no cure or for diseases which manifest later in life. Screening of children should be deferred till the time they are old enough to understand and are able to participate in the decision making process, unless the intervention based on result of the test is likely to be of direct benefit to them at a younger age. Anonymous screening may be conducted on general population in order to establish prevalence of genetic traits/diseases. Anonymous samples means all identifying information about person who has contributed sample is removed and there is no way to know whose sample is it. Left over anonymous blood spots collected for screening newborns for treatable disorders could also be used for this purpose.

4.2.3.9 DNA Testing

For all kinds of DNA diagnosis the general principles of informed consent, confidentiality and other criteria used for any investigation in genetics should be
followed. Since the knowledge in this field is new, and relatively complex, a DNA test must be preceded and followed by appropriate genetic counselling. The laboratories carrying out DNA diagnosis should make adequate provisions to explain fully the nature of testing, implications of results to families. Preferably they should prepare brochures in simple language which can be understood by the persons. Differentiating between clinical practice and research can be difficult in genetics as genetic investigations may extend to other individuals as well as families (Parker, 2004).

4.2.3.10 Prenatal Testing

It is aimed at detecting presence of abnormalities in the foetus. The foetal sample for examination may be obtained through amniocentesis, chorionic villi sampling, cordocentesis or other biopsies. Foetal cells in maternal circulation can also be used for prenatal testing. Non-invasive methods like ultrasound should be preferred whenever available. Prenatal diagnosis should be performed only for reasons relevant to the health of the foetus or the mother and not to select the sex of the child (in the absence of an X-linked disorder). Prenatal diagnosis can be used to prepare parents for the birth of a child with a disability or they have a choice to go for abortion of affected foetus before 22 weeks of pregnancy. Professionals should recognise the human and economic costs involved in prenatal diagnosis and should limit its use to situations where there is a clear benefit. Many of the centers in tertiary care hospitals or urban centers have now initiated carrier screening for some common genetic ailments. Thalassaemia screening an effective control program is being carried out at the Institute of Immunohaematology, Mumbai, India. More than 14,000 women were screened and carriers identified, followed up and offered prenatal diagnosis (Mohanty et al, 2002).

Sex selection, whether for male or female, is very harmful to society since it creates an imbalance in sex ratios. For example you must be aware about the change in the sex ratio in India as commonly pregnancies with girl foetuses are being terminated. The situation is very bad in several states of the country including Delhi, Punjab, Haryana, Rajasthan, Bihar where sex ratio is greatly altered. Concerned with the misuse of genetic tests, particularly for the pre-selection of sex, the Government of India has enacted a law known as “The Prenatal Diagnostic Techniques (Regulation & Prevention of Misuse) Act 1994” amended in 2003 to include the Preconceptual diagnostic techniques also. Pre-implantation DNA diagnosis is also a type of prenatal diagnosis. Same precautions and safeguards should be adopted for this purpose also.

4.2.3.11 Presymptomatic and Susceptibility Testing

Presymptomatic testing (e.g., for Huntington disease) identifies individuals who will develop a genetic disorder later in life. Susceptibility testing (often referred to as ‘predictive testing’) identifies persons who are at increased risk for developing common diseases, such as heart disease, but who may never develop the disease in question. In some cases, presymptomatic testing (e.g., for familial polyposis coli) can lead to prevention of the disorder’s most serious effects (e.g., by colon surgery to prevent cancer). Susceptibility testing can lead to preventive programmes for heart disease or regular examinations to make possible early diagnosis and treatment (e.g., for breast cancer). In other cases, where successful prevention or treatment are not possible, as in Huntington disease, the major benefit of presymptomatic testing is to provide information for planning one’s
life and for deciding whether or not to have children. Pre-morbid diagnosis (diagnosing before appearance of adverse symptoms) in children should not be done for which there is no available intervention. Pre-morbid diagnosis in adults may be carried out with informed consent and appropriate genetic counselling.

### 4.2.3.12 Gene Therapy

Somatic cell gene therapy is permissible for the purpose of preventing or treating a serious disease when it is the only therapeutic option. It should be restricted to alleviation of life threatening or seriously disabling genetic disease in individual patients and should not be permitted to change normal human traits. The guidelines and clearance for gene therapy is regulated by the National Bioethics Committee under Department of Biotechnology (DBT) and clearance from the local Institutional ethics committee should be obtained. Safety should be ensured especially because of the possibility of unpredicted consequences of gene insertion. All gene therapy trials should have the provision for long term surveillance. Germ Line Therapy is prohibited at present. Gene Therapy for enhancement of genetic characteristics (so called designer babies) should not be attempted, as long term effects are not understood regarding alterations to genetic machinery of humans. Similarly it would be unethical to use genetic engineering for improvement of intelligence, memory, physical abilities etc. even if specific gene/genes are identified in future. Eugenic Genetic Engineering for selection against personality, character, formation of body organs, fertility, intelligence and physical, mental and emotional characteristics is prohibited (ICMR Ethical Guidelines, 2006).

### 4.2.3.13 DNA and Cell-line Banking/ Repository

Human genetic material can be enormously precious and especially if it is regarding rare disorders or is derived from special communities or population groups. This material needs adequate preservation and protection for future use and from potential exploitation. Biobanks/Repositories collect, store, and distribute human biological materials for research purposes. Human biological samples in biobanks include organs, tissues, cells, body fluids or samples like serum, buffy coat, DNA, hair, nails, excreta, sweat, buccal scrapings etc. The samples may be anonymously stored with or without its corresponding clinical information in a database. Appropriate informed consent is needed for long term storage of samples for future testing and research. Issues related to use of samples, their control and ownership, and the benefit sharing to the individuals or community also need discussion. To prevent any exploitation and protect the rights of participants, the three main requirements are individual informed consent for future research, approval of the IEC and the Repository Ethics Committee, wherever applicable.

### 4.2.3.14 Cloning

Research using human stem cells to grow new tissues (in order to repair or replace those damaged by disease) holds potential promise. Some of this research may involve nuclear fusion of an adult individual’s cell with an enucleated egg, a first step toward potential human cloning. The possible benefits of research using nuclear fusion to produce tissues for the treatment of disease are recognized, provided that there would be no attempt to reproduce an entire human being. At the present time, “reproductive human cloning” is unsafe and should not be attempted.
4.2.3.15 Stem Cell Research and Therapy

The stem cell research holds a great promise for improving human health by control of degenerative diseases and restoration of damage to organs by various injuries. At the same time it also raises several ethical and social issues such as destruction of human embryos to create human embryonic stem (HES) cell lines, potential for commodification in human tissues and organs, possible use of technology for germ-line engineering and reproductive cloning etc. The research in this field, therefore, needs careful consideration. It is important to protect safety and rights of those donating gametes/blastocysts/somatic cells for derivation of stem cells; or fetal tissues/umbilical cord cells/adult tissue (or cells) for use as stem cells. Safeguards are also needed to protect research participants receiving stem cell transplants, and patients at large from unproven therapies/remedies. Most of the stem cell procedures are still in the research or experimental phase and long term effects are unknown therefore stem cell studies should be done as a clinical trial and not offered as proven therapy to patients.

4.2.4 Other Concerns in Genetics Research

4.2.4.1 Commercial Issues, Stigmatisation and Discriminations Based on Genetic Characteristics

When commercial companies are involved in research, it is necessary to protect researchers and participants from possible coercion or inducement to participate in the study. Academic institutions conducting research in alliance with industries or commercial companies require a strong review to probe possible conflicts of interest between scientific responsibilities of researchers and business interests (e.g. ownership or part-ownership of the investigator in the company developing a new product). Prospective participants in research should also be informed of the sponsorship, so that they can be aware of the potential for conflicts of interest and commercial aspects of the research.

The other concern is regarding insurance issues where knowing about certain types of genetic information can lead to discrimination by insurance companies who may refuse to provide insurance or charge higher premium. Similarly there is a potential threat that employers may not provide jobs to individuals with certain type of genotypes or school may not admit children based on their genetic diagnosis. There are also major concerns regarding stigmatisation of certain population groups or certain communities based on the genetic information. Many of these examples may sound theoretical today however with emerging new tools for genetic diagnosis there is a need to make provisions in advance for protecting populations or groups from such potential harm. It is need for us to be aware of these scenarios and protect our rich heritage.

4.2.4.2 Patenting

Biomedical research in human genetics can lead to the development of diagnostic and pharmaceutical products. Patents may be necessary to raise funding to develop such products commercially, but gene sequences without proven utility should not be granted patents. Patenting has the potential to impede international collaboration, especially between developing and developed countries, to the ultimate detriment of service delivery to those with genetic disorders. Genetics differs from many areas of research in that important new knowledge can come from a family, or an ethnic group, with a particular genetic variant. If this leads
to the development of a diagnostic test or new therapies, equity requires that the
donors, or the community generally, should receive some benefit.

4.2.4.3 Research in International Collaboration

India with its rich biodiversity and genetic resources, conserved gene pools,
indigenous population groups is often an attractive destination for researchers
from other countries. However it should not only be provider of samples but be
an equal collaborating partner in research to ensure development in science.
Such collaborative partnerships should lead to development of capacity within
the country for better genetic research rather than mere transfer of samples abroad
for diagnostic purposes. At present a lot of genetic research is carried out in
International collaboration. This requires adequate care so that the rights of the
communities/ persons from whom samples are derived from are adequately
protected if there is transfer of biological material across borders. On one hand,
collaboration in genetics helps build capacity and is useful for betterment of
scientific knowledge but on the other it should not give the impression of
experimentation on the population of one country by another. There is a Govt of
India notification on “Exchange of Human Biological Material for Biomedical
Research” issued on 19.11.97 by Ministry of Health and Family Welfare. Appropriate regulatory clearances are needed for international collaboration and
Ethics committee approval has to be taken before the initiation of research. Some
special concerns are given below in Table 4.3.

Table 4.3: Safeguards for Research in International Collaboration

- The collaborating investigators, institutions and countries can function as
equal partners by building appropriate safeguards.
- Careful consideration should be given to protect the dignity, safety and
welfare of the individuals if there is any possibility of exploitation of
participants/ populations.
- Choice of study populations for genetic study should be justified in scientific
and ethical terms.
- Nature, magnitude, and probability of all foreseeable harms resulting from
participation in a collaborative research programme should be explained to
participants.
- Research protocol should outline the benefits that persons / communities /
countries should experience as a result of their participation.
- The burden and the benefit should be equally borne by the collaborating
countries.
- Guidelines, rules, regulations and cultural sensitivities of countries
participating in collaborative research should be respected.
- Issues such as intellectual property rights (IPR), exchange of biological
materials, data transfer should be carried out as per signed Memorandum
of understanding (MoU).
- Collaborative research requires clearance from ethics committees as well
as from Health Ministry Screening Committee (HMSC) as per regulatory
requirements.
4.3 SUMMARY

The last decade has witnessed tremendous advances in science and technology and the prospects of genetic testing, genetic engineering, cloning, stem cell research, prenatal testing, DNA diagnostic etc. all have raised many ethical dilemmas in the minds of scientists and society. There are advantages of such new technologies but there is also the fear of the unknown distant possibilities which needs careful examination. Weighing of risk benefit ratio, protection of privacy and confidentiality and sharing of burdens and benefits are some of the main issues. Close monitoring of research in these areas is important. Education has the potential power to prevent stigmatization and discrimination by emphasizing that genetic disorders are not caused by the behaviour of affected persons or families but the fact that most people may carry some recessive lethal mutations and that our offspring or we are all at genetic risk. For all kinds of genetic testing and research, respect for persons is important and should include informed consent, right to referral, full disclosure, protection of confidentiality, and respect for minority groups, women and children. The following statements summarize the issues:

- Genetic services should be voluntary but should be made available to those interested in them.
- Genetic counselling should be non-directive to let the decision making be done by individuals without any force or coercion.
- All clinically relevant information that may affect the health of an individual or fetus should be disclosed and various options explained.
- Confidentiality of genetic information should be maintained. When there is a high risk of serious harm to family members at genetic risk, the information should be used to avert this harm.
- Individual privacy should be protected from third parties, such as employers, insurers, schools, commercial entities etc.
- Prenatal diagnosis should be performed only for reasons relevant to the health and only to detect genetic conditions or malformations.
- Choices about counselling, screening, testing and abortion following prenatal diagnosis should be available on a voluntary basis.
- Optimum support and education should be provided for children and families with genetic conditions.
- Research protocols should follow established procedures for ethical review and informed consent.

Further Reading and References

Ethical Guidelines for Biomedical Research on Human Participants, 2006.


Sample Questions

1) Write a short note on Human Genome Project and its implications.

2) What do you understand by Genetic Counselling and why is it important?

3) What are the ethical, legal and social issues? Discuss.

4) Write a note on the Ethical Principles in Genetic research.

5) What is informed consent in research? Give a few main elements of informed consent.

6) What do you understand by vulnerable persons/populations?

7) Why should there be adequate provisions to protect the privacy and confidentiality of persons in genetic research?

8) Describe any three specific concerns in genetics research.

9) Write a short note on biobanks.

10. What safeguards should be in place for genetic research in International Collaboration?