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Review Article

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Ethical issues in genetic counselling with special reference to haemoglobinopathies

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Genetic counselling is provided in places where genetic tests are carried out. The process involves pre-test counselling as well as post-test counselling to enable the individuals to face the situation and take appropriate decisions with the right frame of mind. Major ethical principles which govern the attitudes and actions of counsellors include: respect for patient autonomy, non-maleficence, beneficence, or taking action to help benefit others and prevent harm, both physical and mental, and justice, which requires that services be distributed fairly to those in need. Other moral issues include veracity, the duty to disclose information or to be truthful, and respect for patient confidentiality. Nondirective counselling, a hallmark of this profession, is in accordance with the principle of individual autonomy. High prevalence of haemoglobinopathies with availability of good and sensitive carrier detection tests and prenatal diagnostic techniques makes these good candidates for population screening of carriers along with genetic counselling for primary prevention of the disease. Screening of the extended family members of the affected child, high risk communities and general population screening including antenatal women are the main target groups for planning a Haemoglobinopathy control programme. A critical mass of trained genetic counsellors who have understanding of the ethical issues and its appropriate handling with the required sensitivity is needed in India.

Key words Carrier detection - ethical issues - genetic counselling - genetic counsellor - genetic disorders - haemoglobinopathies - prenatal diagnosis

With the advent of the human genome programme in 1990 and the ultimate release of the human genome map in 2000[1], tremendous advances have been made in the area of molecular genetics which have not only led to identification of large number of disease causing genes but have also opened the flood gate of DNA diagnostic tests which have found a firm place in clinical management of various diseases by way of genetic counselling, carrier detection, pre-symptomatic, pre- implantation and prenatal diagnosis followed by reproductive choices.

Genetic counselling is defined as a communication process involved in human problems associated with the occurrence and recurrence of a genetic disorder in a family. This process involves the expertise of a trained counsellor to guide individuals and/or a family to comprehend the medical facts related to diagnosis, prognosis and management of a genetic disorder, the role of heredity in such conditions and the probable impact on the other members of the family, preventive measures for further recurrence of such disorders in the family, choose the right course of action according

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to the ethical, moral and religious standards and adopt the best possible solution to the disorder in the affected individual or family. This also requires provision of adequate social and psychological support to the individuals and the affected family so that the patient and the family can cope up with the problem of genetic disorder. Thus genetic counselling provides the individuals and the family members all the necessary scientific information about the particular disease, its course, possible management, risk of recurrence and any provision of prenatal diagnosis or other preventive measures thereby reducing the psychological distress and improving the personal control of the specific situation. Trained counsellors are expected to provide this service in the genetic clinics where genetic tests are being carried out and the process involves pre-test as well as post-test counselling to enable the individuals to face the situation and take appropriate decisions with the right frame of mind.

Ethical issues in genetic counselling

Rapid growth in the area of medical genetics is providing a wealth of new options for dealing with genetic disease. These technologic advances often pose unique and significant ethical dilemmas which must be resolved by providers of genetic services and individuals who receive those services, as well as society in general. Reproductive options such as artificial insemination by donor, genetic screening, *in vitro* fertilization, sex selection via prenatal testing, surrogate motherhood, foetal tissue transplantation, and gene therapy have generated considerable ethical concerns. As genetic counsellors relay important genetic and reproductive information to families at risk and to the public, they often play an important role in the way the relevant ethical issues are understood and acted upon.

Major ethical principles[2-4] which govern the attitudes and actions of counsellors include *(i)* respect for patient autonomy, or the patient's right to information and his/her right to make his/her own decisions, non-maleficence, which is defined by Fletcher *et al*[5] as one's "duty to minimize or prevent the infliction of harm on individuals and families"; *(ii)* beneficence, or taking action to help benefit others and prevent harm, both physical and mental, and *(iii)* justice, which requires that services be distributed fairly to those in need.

Other moral issues include veracity, the duty to disclose information or to be truthful, and respect for patient's confidentiality. Nondirective counselling, a hallmark of the genetics profession, is largely in accordance with the principle of respect for patient autonomy and incorporates the other ethical principles as well.

Genetic counselling raises special ethical issues related to confidentiality and privacy protection. Information about the individual, family history, carrier status, risk of genetic disease to self or offspring can be stigmatising and hence needs to be kept confidential. Most of the time the client is concerned about the future reproductive or personal health risks. The counsellor has to make an assessment by obtaining family history and by analysing the pedigree chart to provide information regarding the treatment options, preventive strategies including reproductive options and the financial and social implications. The psychosocial impact of the carrier status is very crucial in our society. Hence, a thorough assessment of the socio-cultural issues is essential along with large scale awareness programmes including comprehension of the information amongst young adults, patients and their relations and the medical professionals themselves with the help of religious, social leaders, social organisations, patient support groups and effective mass communication systems like radio, TV, films, periodicals, *etc.* to create public awareness. This will facilitate the role of the genetic counsellors to a great extent.

Psychosocial aspects of genetic counselling pass through four different phases of coping process: *(i)* Initial shock and denial; *(ii)* Subsequent anger and guilt; *(iii)* Followed by anxiety and depression; and finally *(iv)* the phase of acceptance and adjustments.

The counsellor needs to handle the clients carefully through the different phases to bring them out of each phase.

The common indications for genetic counselling are occurrence of congenital malformations, mental retardation, neurogenetic disorders, myopathy, ambiguous genitalia, skeletal dysplasias, childhood deafness, Down's syndrome, haemoglobinopathies, haemophilia, Wilson's disease, mucopolysaccharidosis, familial cancers, consanguinous marriage, advanced maternal age, *etc*[6]. The above may be related to chromosomal disorders or monogenic disorders or multifactorial diseases. Genetic counselling is a multistep process involving clinical expertise and psycho-social impacts, hence specially trained clinicians can offer better counselling to such individuals and families. The

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steps involved include history taking along with drawing up the pedigree chart, clinical examination, diagnosis, management, advice on risk of recurrence and available preventive measures including prenatal diagnosis, wherever available. Advances in the post-genome era have transformed the genetic counselling based on Mendelian probabilities to the science of certainty based on antenatal diagnosis. A genetic counsellor needs to have excellent communication skills.

A primary care physician or paediatrician or many times an obstetrician are the ideal clinicians who need to undergo training in genetic counselling as these specialists can provide a complete fullest comprehension of the disease to the families at risk and help them through their problems and facilitate their decision making for adjustments as well as reproductive choices. Many a times specially trained individuals belonging to other specializations such as nursing or social sciences may take up the responsibility of genetic counsellor. Such counsellors should have the required medical knowledge, necessary skills for providing efficient and effective counselling and also proper attitude, behaviour and appropriate training.

Ethical problems faced by the counsellor

There cannot be a universal model for genetic counselling because counselling is an understanding of a set of facts according to the counsellors' frame of reference, background in the science of genetics, and previous training and experience in effectively communicating with the consultee. In order to communicate effectively, the counsellor must consider the educational background of the consultee, what to disclose and how to limit the ways in which he or she can communicate. It has been found that the principal obstacles to the effective use of genetic counselling are emotional conflicts, and lack of knowledge of genetics and biology.

An equally difficult assignment for the counsellor is presenting his/her knowledge in an unbiased manner[5]. It is difficult for a counsellor to impart unbiased information because of the consultee's personal and family history such as parental age, ethnic background, reproductive history, *i.e.* abortions, stillborn or dead siblings, and the age, sex and health of the living children. This may lead the counsellor to adopt a directive rather than a non-directive approach to genetic counselling. The major difference between directive and non-directive counselling is whether or not the counsellor actively participates or helps the consultees to make a decision. Directive counselling has a positive influence on the consultee's decision. The non-directive approach involves presentation of the facts in an unbiased manner, leaving the entire responsibility of decision with the consultee. Counsellors can be faced with dilemma in respect to certain inherited conditions because of improper measurements and observations and/or because of similar symptoms of many genetic diseases. However, the counsellor probably cannot completely disassociate himself/herself from his/her own values and present the information in such a way that the recipient is not completely free to make his/her own judgement. The counsellor may not change the truth but the tone, manner of speech and other facial and body gestures can influence the information transfer. In a case where a counsellor feels that a pregnancy might be best for a family he/she could say to Mr and Mrs X, 'there is only one chance in four that your child would be affected. Your chances for a healthy birth are very high, three chances out of four or 75 per cent'. For family Y with these same inherited defect, but a different social history, the counsellor may emphasize more on the problems that may arise because of the particular disorder.

During counselling, the counsellor may come across other findings, that may put him in a situation of ethical dilemma[7]. Some of these are foetal sex, findings of questionable or potentially harmful significance, false paternity, *etc.* Is there a need to disclose foetal sex to parents, if this finding is not related to any disease, except in X-linked disorders? Should physicians co-operate with the desire of the parents or individuals autonomy to know the foetal sex, especially when there may be reason to suspect that some parents will misuse the information and seek abortion elsewhere for undesired gender?

Occasionally, disputes arise about the significance of laboratory findings especially about the true vs. pseudo-mosaicism or by possibility of contamination by maternal cells. When genuine doubt exists and it is too late to do a repeat procedure, what should the parents be told? Should conflict about findings and interpretations between professionals be revealed to parents? Another example is when sonography suggests an irregularity of the foetal head but the amniotic fluid is normal for alpha-fetoprotein. The issue is whether the disclosure of a finding of probably small significance will result in severe parental anxiety leading to psychological problems. Another difficult conflict involves males and females with normal phenotypes who are discovered

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to have XX or XY chromosomal complement, respectively. Should they be told? Will a full biological explanation harm their self-esteem and damage them psychologically?

Medical geneticists learn many family secrets, such as previous abortions, previous abnormal births, and occasional false paternity. The findings can be made after PND (prenatal diagnosis) of a recessive disorder and testing the carrier parents or in the context of genetic screening after the birth of an affected child. The putative father believes that he must be a carrier, but tests are negative. The option left is partial or total deception. Should the family be protected from the disruption due to disclosure, with the risk of inappropriate decisions about future child bearing being based on false information? Should actual risk be revealed with no explanation? Should the mother be told the truth leaving the option to her to decide about further disclosure? These are some real dilemmas for the counsellors.

Genetic counselling for haemoglobinopathies

Thalassaemia and sickle cell anaemia are the commonest haemoglobinopathies among the single gene disorders in India and their prevalence varies from 3-10 per cent in different populations. Sickle cell disorders are much more frequent in specific population groups including some primitive tribes. High prevalence of these disorders, availability of good and sensitive carrier detection tests and prenatal diagnostic techniques make these conditions good candidates for population screening of carriers along with genetic counselling for primary prevention of the disease. Screening of the extended family members of the affected child, high risk communities and general population screening are the different target groups for planning a Haemoglobinopathy control programme as was done in countries like Cyprus, Greece and Sardinia.

It is necessary that the individuals being tested must know 'what is being tested' and 'why'. Counselling before the test should include complete information about the burden of the disease, and the implication of being detected as a thalassaemia carrier. It should be made clear to them that in majority of cases there are no health consequences to the carriers. The tests for carrier detection are available and it is possible to prevent the birth of an affected child if appropriate actions are taken at appropriate time by opting for prenatal diagnosis at the identified centres. The occurrence or recurrence in a family, the economic, social and psychological impacts, resources available to the family and the strategies for prevention have to be explained. The counsellor is expected to present all the information fairly and even-handedly without encouraging a particular course of action *i.e.*, a non-directive counselling and the client is encouraged to take autonomous decision on the future course of action after understanding the implications of the different strategies available.

Population screening is adapted as a national strategy in small countries to prevent genetic disorders specially haemoglobinopathies[8]. In a country like India it is a major challenge and antenatal diagnosis of pregnant woman can be a possible, feasible strategy to prevent birth of a thalassaemic child. It is possible for the genetic counsellors to approach this target group in antenatal clinics under the national family welfare programme. Diagnostic accuracy and safety for mother and foetus, abortion of the foetus diagnosed as homozygous for thalassaemia or sickle cell diseases, and access to prenatal diagnosis for those who cannot afford it are the current ethical debate although more instances of persons coming forward for antenatal diagnosis are coming to light.

The Indian Council of Medical Research conducted two multi-centric studies, one on Community Control of Thalassaemia in six States[9] and the other on Sickle cell disorders amongst fourteen ethnically different primitive tribal population in four States over a period of five years[10] from 2000 to 2005 for carrier detection, identifying index cases and screening of antenatal women for possibility of prenatal diagnosis along with genetic counselling. This was the first study in India reporting on the effects of genetic counselling in the primitive tribes. Many of the marriageable youths counselled came forward for premarital screening indicating an effective counselling. Similarly counselling high risk communities like sindhis, lohanas, banushallis, kutchis, *etc.* with targeted screening of these population groups for thalassaemia has been attempted with reasonable success with the help of religious and social leaders[11]. It is interesting to note that when the thalassaemia carrier screening was started in Assam and West Bengal the response was very poor. However, after intense awareness programme in the community and counselling on one to one basis the success in screening programme has increased tremendously in West Bengal and Assam. West Bengal Government has undertaken the Thalassaemia Control programme in the State. Counselling extended families of the affected children is the most rewarding strategy

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adapted to reduce the burden of thalassaemia and other haemoglobinopathies although the fear of stigma and financial burden has prevented many eligible couples to come forward for carrier detection and prenatal diagnosis. Unless all these groups are approached with sustained efforts by trained counsellors, no control programme can accomplish reduction in disease burden in the country.

Establishing testing centres alone without the necessary critical mass of trained counsellors and appropriate counselling strategies to understand and interpret the implication of the test results, will not be able to address the issues involved in genetic screening programmes. The need of the hour is to create a critical mass of trained genetic counsellors in the country which is grossly lacking at present. The only institution in the country at present to generate such human resource is Sanjay Gandhi Postgraduate Institute of Medical Sciences (SGPGIMS), Lucknow. More such centres need to be established in different regions of the country to face the challenge of controlling genetic disorders to reduce the morbidity.

Conclusion

Moral problems arise constantly in social life with the need to resolve conflicts between moral rules and principles to help, regulate and modify desires. When genetic risks are high, the desire to have a healthy child and to avoid danger to oneself, family and society are frequently in conflict. Although more than 90 per cent of the counselling sessions end well with no or very little chances for the occurrence of the disease, the remaining people in the high risk category are left with three options: *(i)* prenatal diagnosis and abortion if required, *(ii)* artificial insemination, and *(iii)* gene therapy Genetic counselling is a practical method of calculating risk figures, intended for information regarding the unborn, and we ought to use it in an efficient manner but in a direction, which our ethics and morality point to. The decision taken by the parents after the counselling session must leave them satisfied instead of placing them in a state of dilemma.

Application of science and scientific principles has two faces. To decide the correct use, man must deal with his conscious, individuals and social status and the ethics underlying the applications. Rapid technological progress in the field of genetics demands the moral and ethical attention of each medical geneticist and genetic counsellor. These professionals are in an ideal position to understand and influence the ethical impact of new technology on the individuals and society. It is important that genetic counsellors continue being patient advocates while ethical guidelines for their use with appropriate counselling procedures are developed after ensuring adequate human resource in this area. There is an urgent national requirement to generate more trained genetic counsellors who can approach the problem with the understanding of the ethical issues and its appropriate handling with the sensitivity required for such conditions. Policy makers and the professionals have equal responsibility in this regard. Public awareness is the need of the hour.

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