TABLE NO. 1 : INDICATIONS FOR GENETIC COUNSELLING

1. Know or suspected hereditary disease in a patient or family
2. Birth defects
3. Unexplained mental retardation
4. Advanced maternal age
5. Teratogen exposure
6. Consanguinity

THE INFORMATION CONVEYED DURING GENETIC COUNSELLING:

i) THE MAGNITUDE OF THE RISK:

To understand the magnitude of risk, it is essential to examine the affected patient whenever possible to obtain appropriate laboratory analysis and diagnosis. Genetic heterogeneity and variable expressivity create problems in diagnosis. In order to provide optimal assessment of risk, it is important to use all available information especially when the genotype of the individual being counselled is unknown.

ii) THE BURDEN OF THE DISEASE ON THE PATIENT AND FAMILY:

Conveying the magnitude of recurrence risk of a genetic disease is not the sum total of genetic counselling, but the first step. Understanding the burden of disease refers to the clinical, financial, social and emotional burden imposed upon the patient and/or the family. The burden of a given disease may vary with the severity of the disease itself, but will also vary with the individual and family affected.

iii) POSSIBILITY OF MODIFICATION OF EITHER THE BURDEN OR THE RISK:

For a number of genetic diseases, it is possible to modify either the burden or the risk. If the disease is treatable or preventable then the burden of disease although still significant is quite different from what it would be if treatment or prevention were not available.

PRACTICAL ASPECT AND EFFECTIVENESS OF GENETIC COUNSELLING:

Essential information to be collected prior to counselling are:

- Confirmed diagnosis in the index patient
- Mode of inheritance of the disorder
- Accurate pedigree of the family

The process of genetic counselling can be divided into 4 consecutive phases:

- Initial phase: information is gathered and diagnosis is made.
- Facts about disease, genetic implication and possible options are imparted to and discussed with the counsellee.
- Counsellee evaluates assimilates and learns to cope with the given information
- Decisions.

The first step in genetic counselling is to ascertain what question the counsellee (the individual seeking counselling) is really asking. Since it involves communication, one must assess the receptivity, emotional and intellectual, of the individual seeking counselling. Couples should be counselled together. Patients seeking genetic advice should be seen in a quiet room. There should be no feelings of hurry, as varied and intimate personal questions and problems often arise. The interview will often take as long as an hour or more.

There is a tendency to hide the knowledge of hereditary defects in one's family and is considered in the same category as moral offense. In addition, it is not only a feeling of shame which induces many people to keep genetic facts secret, but also the fear that the marriage prospects of their children may be decreased. There is, of course no reason for being ashamed of genetic constitutions which fate and not the individual has decreed. In circumstances like these, secrecy itself is culpable, for it may have more serious consequences than the truth.

The mode, in which this type of information in explained to counsellees, is of utmost importance and also greatly influences the interpretation of the facts. Discussion of the options open to a couple makes the counselling process more comprehensive and relevant.

Genetic counselling must not be directive, i.e., the counsellee should not be directed to whether to have children or not. Reproductive decisions are highly personal ones and should be left to individual couples rather than being made by their physicians and counselors.

The information must be conveyed in language that can be understood by the counsellee. It should be believed that candor and honesty are the best way to present such information. The information communicated is heavy with emotional content and often contains medical or scientific information that is difficult for individual to comprehend. The verbal counselling should be followed up by a letter, which enables them to read and reread the counselling and to share it with physicians, family members or other supportive individuals.

PSYCHOLOGICAL ASPECTS OF COUNSELLING:

The responsibility of a genetic counsellor does not end when he has made an accurate diagnosis. He must, in transmitting his information to the individual concerned, take into account the psychological effect of unfavourable prospects. It is not sufficient to tell the truth, but it is necessary to tell it humanely.

Lack of motivation, overt anxiety and even hostility can seriously disrupt the counseling relationships. The factor most commonly found by counselors to cause difficulties in effective communication is the educational background of their client and in particular the client's knowledge of biology. The environment in which the counselling takes place can also greatly influence the process.