EUGENICS AND GENETIC CONSELLING

The term eugenics (Gr. eugenes = well born) was coined by an English scientist Francis Galton in 1885. The science of eugenics can be defined as a science of well born, improving the inborn qualities of race and obtaining the better heritage by judicious breeding.

The betterment of human society can be achieved by following two inter-related methods:

- 1. By one of the method we can deal with the already existing human beings. The improvement of already existing human beings can be achieved by improving the environmental conditions, e.g., by subjecting them to better nutrition, better unpolluted ecological conditions, better education and sufficient amount of medical facilities. This type of method of improving the human race is known as euthenics.
- 2. By another method we can improve the future generations by improving the germplasm of existing individuals. This type of method is known as eugenics. Eugenics believes in artificial selection of physically and mentally sound individuals and discouragement of defective individuals for the inheritance of their defective germplasm to the future generations.

In other words, eugenics seeks the measures to preserve the best type of germplasm and to eliminate defective germplasm from the human society by applying the laws of inheritance to human beings.

The primary aim of many of the ancient systems of eugenics was to produce a race of physically perfect human beings. The Greeks had definite ideas regarding eugenics. In Sparta, a physically perfect manhood was the chief aim, whereas the Atheniens carried more for the intellectual achievements.

Following the doctrine of Greeks and until the 19th century there was little interest in the eugenics. Of the particular importance was the eugenics movement in England in the last part of the nineteenth century.

The movement, spearheaded by persons of outstanding intellect such as Francis Gallon and Karl Pearson, had its objective in the application of biologically sound principles to human populations. Since the biological basis of heredity was unknown, the first objective was to establish the nature of heredity.

Galton and Pearson chose to work with human beings and with what they considered important human traits such as intelligence, stature and special abilities. We now know that these characters are very complex traits and are under the control of many genes (polygenes) interacting with environmental variations.

So, quite naturally the early investigators made little progress. However, Francis Galton should be credited for being the real founder of the modem movements of eugenics. He defined the eugenics as the study of all the agencies under social control which may improve or impair the inborn qualities of fine generations of humans either physically or mentally.

Darwin also attached great importance to the eugenics and he compared it to a signpost with three directions. One of these indicates the influence of heredity on the fate of nations.

Another point to the rules is that an individual should try to carry out in regard to parenthood based on the law of human heredity. The third aim indicates the regulations to be adopted by the society to encourage racial progress.

For better type of development both good heredity and suitable environment are necessary.

When we consider the future welfare of the human race then the following two factors alarm us greatly:

- (i) The declining birth rate among the normal and superior people (those having best germplasm)
- (ii) A relative rapid increase of the abnormal and defective individuals (those having defective germplasm).

For the betterment of future generation, it is necessary to increase the population of outstanding people and to decrease the population of abnormal and defective people by applying the principle of eugenics.

The eugenics can be applicable by adopting following two methods:

- (A) By encouraging the marriages between desirable persons (constructive method or positive eugenics).
- (B) By discouraging the marriages between undesirable persons (restrictive method or negative eugenics).

A. Positive Eugenics:

The positive eugenics attempts to increase consistently better or desirable germplasm and, thus, to preserve best germplasm of the society.

The percentage of desirable traits can be increased by adopting following measures:

1. Early Marriage of those having Desirable Traits:

It is most commonly observed fact that the highly placed persons of the society often have great ambitions for the future life. In achieving their ambitious goals, they often devote the best part of their youth and they are able to marry in their mature age (e.g., 30 to 35 years).

The biological and psychological investigation have revealed that the aged persons often lack in necessary amount of emotional warmth for the sexual activities and moreover, their germplasm also loose its vigour.

Therefore, some laws should be formulated to prevent the late marriages of highly endowed persons by applying high taxation on them and at the same time the young persons having best hereditary traits should be encouraged for early marriage.

2. Subsidizing the Fit:

Because the highly endowed persons lead a well-planned life and to avoid unnecessary difficulties in nursing the children they often prefer to have small number of children. Therefore, the selected young men and women of best eugenic value should be encouraged to increase their birth rate.

3. Eutelegenesis or Germinal Choice:

H.J. Muller has suggested that eugenically sound persons not only should increase their family size but through artificial insemination the outstanding man can serve as father to many more children than would be otherwise possible. The artificial insemination is already widely practiced to permit those women whose husbands are sterile or have some serious hereditary afflictions to bear children.

The sperms and eggs of outstanding persons can be stored for future use by quick freezing and storing them in deep freeze. These germ cells, thus, can be stored for 100 or more years.

Very recently the scientists felt the urgent need of establishment of sperm and egg banks to protect these precious germ cells from the radiation. The germ cells could be collected during early adulthood and stored in lead lined containers in the deep freeze. In this state the germ cells would not be subjected to radiation exposure which might affect the donors.

In a modern technique, a woman's ovum is taken out and artificially fertilized in a test-tube. The zygote is then implanted into woman's uterus for the embryonic development. The resulting baby is called test-tube baby.

4. Education:

For the eugenically oriented reforms in the society, the people should be educated about the basic principles of human biology, human genetics, eugenics and sex. The children should be instructed about basic laws of health and they should be encouraged to develop a physically and mentally healthy body.

Moreover, sex should be free from the widespread confusion, narrow minded concepts and religious taboos and ethical bindings because that is a natural biological instinct. The children ignorant about the facts of sex may do more harm to society than otherwise.

5. By Avoiding Germinal Waste:

The wastage of best type of germplasm can be avoided by adopting following measures:

- (i) The selection of marriage partners should be made with intelligence.
- (ii) The social hindrance which do not allow the teachers, nuns and priests to get married, must be removed. By adopting such measures wastage of best type of germplasm due to lack of opportunities can be prevented.
- (iii) The wars must be avoided because in wars the best germplasm of the society is wasted.

6. Improvement of Environmental Conditions:

Both heredity and environment have interrelated role in the development of eugenically better persons. Therefore, every person should get better food, living conditions, education and medical guidance, etc., so his or her hereditary traits can undergo their best development.

7. Promotion of Genetic Research:

Our knowledge about the genetics is not sufficient enough because we still have little information's about various human diseases and metabolic disorders which are generally related with the genes. Therefore, the research in the field of cytogenetics should be increased so that we can learn more and more about the human beings.

8. Genetic Engineering:

During the late 1970s, the science of genetics entered a new era dominated by the use of recombinant DNA technology or genetic engineering (or biotechnology) to produce novel life forms not found in nature.

Through this technology, it has been possible to transfer genes from mammals into bacteria, causing the microbes to become tiny factories for making (in relatively large quantities) proteins of great economic importance such as hormones (including growth hormone and insulin) and interferon's (lymphocyte proteins that prevent replication of a wide variety of viruses).

These proteins are produced in such small quantities in humans that the cost of their extraction and purification from tissues has been very expensive, thus, limiting their medical use in prophylaxis (prevention) and therapeutics (treatment) of disease.

By genetic engineering, it has become possible to produce various blood clotting factors, complement proteins (part of immune system) and other substances for the correction of genetic deficiency diseases (euphenics).

Recently, experiments have been conducted in which human cells deficient in the synthesis of purines have been obtained from the patients with Lesch- Nyhan syndrome and grown in culture; these cells have been converted to normal cells by transformation with recombinant DNA.

The exciting potential of this technique lies in the possibility of correcting genetic defects-for example, restoring the ability of a diabetic individual to make insulin or correcting immunological deficiencies. This technique is called gene therapy.

B. Negative Eugenics:

The negative eugenics attempts to eliminate the defective germplasm of the society by adopting following measures:

1. Sexual Separation of the Defective:

The defective persons may have various sex-linked diseases such as night blindness, haemophilia, colour blindness, etc., and various other defective traits which may be regulated by dominant or recessive genes.

The increase of germplasm of the persons having such defective traits in the population can be checked by keeping them away and separated from the society. Different states have wisely adopted the restricted measures in segregation of the mental defectives from the society and to place them in mental hospitals.

2. Sterilization:

The sterilization is the best means to deprive an individual from his power of reproduction without interfering with any of his normal functions. The sterilization method is based on surgical operation of sperm duct or vas deferens in males and oviducts or fallopian tubes in females.

The former is known as vasectomy and the latter is tubectomy or salpingectomy. The family planning movement in India has adopted the sterilization as the tool for controlling the rate of rapidly increasing population and in that case the sterilization is euthenical in its application than eugenical.

3. Control of Immigration:

Through immigration there are enough chances that undesirable or defective genes of different races and nationalities may intermingle with the normal germplasm of the population. Therefore, the immigration rules must be strict and the persons with undesirable hereditary traits must not be allowed to migrate from one place to another.

4. Regulation of Marriage:

Presently most human societies are money-minded and for a marriages, relationship like the wealthy or highly placed persons who, however, may have several defective genetical traits, are preferred over those who have economically weak but having eugenically sound hereditary traits. Some rule must be enacted to encourage marriages among desirable mates.

5. Birth Control:

People possessing more of undesired, dysgenic traits should be encouraged to have small families if at all allowed to reproduce. Their foetuses possessing dysgenic traits may be destroyed by abortion.

6. Statutory Ban on Marriage among Close Relatives:

Marriages among close relatives are called consanguineous marriages. The hidden recessive deleterious (e.g., lethal) traits can appear in the progeny in full view if both the parents carry them. The chances of both the parents carrying the hidden traits are more if they are close relatives and have some common ancestors.

Therefore it is advisable not to marry a close relative. If family histories of a rare abnormality are collected and there is no dominance in them, we may generally expect that the anomaly is due to a double dose (i.e., homozygous state) of a recessive gene.

If the parents are blood relatives, there will be a reasonable chance that both will be carrying at least one of the deleterious recessives possessed by one or more of their common ancestors.

Thus, by marrying a relative, one tremendously increases the chances that a recessive, which has been carried concealed down through the generations, will become expressed in the effective double dose in children.

Many societies have some sort of taboo or restrictions on marriages between close relatives, which may have arisen as a result of the observation that such marriages often produced defective offspring.

In most countries, no man is allowed to legally marry his sister, mother, daughter, granddaughter, aunt or niece. Many societies have extended such restrictions to include prohibition of first cousin marriages also.

Consanguineous marriages has influenced world history:

Consanguineous marriages have been sanctioned by royalty. This is the reason for the spread of haemophilic gene to European dynasties by the offspring of Queen Victoria. The death of crown prince of Spain, Prince Alfonso, took place by profuse bleeding after a minor cut which would not have been deadly in a normal individual. This was due to X-linked recessive gene for haemophilia he inherited. This gene does not allow the blood to clot.

This condition was perpetuated in royal families of Europe because of the close intermarriages. Rasputin, "the mad monk of Russia", obtained strong hold of Russia because he was being considered successful in treating the profuse bleeding (i.e., bleeder's disease) attacks of Tsarevitch. This monk used his power and played a major role in the effectiveness of Russian revolution which changed the history of the world. Thus, we see how the inheritance of gene has influenced world history.

7. Genetic Counselling:

Negative eugenics is most widely acceptable when it is practiced through genetic counselling. A voluntary restriction of child bearing by couples with inherited genetic disorders (e.g., albinism, sickle cell anaemia, etc.) can be brought about through proper counselling by well qualified persons in the field.

The term genetic counselling is applied to service, typically available in medical settings, in which the prospective parents are provided with the estimates of the probability that they will produce children with genetically controlled defects. This vital service is intimately related to medical and diagnostic procedures, but the counselling itself does not involve their actual performance.

Medical genetics units are often attached with certain hospitals and medical centres and they provide services of genetic counselling. A genetic counsellor may be a medical doctor or well-trained professional human geneticist.

The most common situation under which people obtain a genetic counsellor's advice is one in which phenotypically normal couple produces a first child suffering from a major defect. Understandingly they wish to know what are the chances for subsequent children to be affected.

The genetic counsellor's first task, invariably, is to collect the relevant evidence by making a pedigree study of the man and the women concerned. He then proceeds to estimate the risk or probability of genetic defects among their progeny by applying established principles of inheritance, to the information already collected.

The counsellor, normally, cannot predict anything with certainty and is only in a position to explain the chances of occurrence of a trouble. The final decision is always left to the couple themselves. The role of genetic counsellor will become more important with the perfection of the technique of identification of genetic defects.

It is relatively easy for a trained clinician to identify people suffering from a genetic disease. But the identification of carriers (heterozygotes) for genetic diseases are readily identifiable when they have either a reduced level of the concerned enzyme (e.g., HGPRT enzyme in Lesch-Nyhan syndrome), or a protein/enzyme with an altered charge and, consequently, changed mobility when subjected to electrophoresis (e.g., HbS produced by persons heterozygous for the sickle cell gene). Analysis of family pedigree also provides information of the likelihood of an individual being carrier for a genetic disease.

Once genotype of both the prospective parents become known, it is a simple matter to work out the probability of their child inheriting the disease. If parents heterozygous for a genetic disease decide to produce a child, it is now possible, through appropriate tests done about 6-8 weeks after conception to advise them if their child has inherited the disease.

This is done by obtaining foetal cells from biopsies of trophoblastic villi, which form an external part of human embryo and later form a part of the placenta.

Foetal cells may also be obtained by amniocentesis done usually after 18 weeks of pregnancy. The cultured foetal cells may be used for determining their karyotype, levels of critical enzymes and restriction enzyme digestion patterns of DNA.

Such an antenatal (=prenatal) diagnosis is now attainable for more than 35 genetic diseases and for a variety of karyotypic defects. The sole purpose of such a diagnosis is premature termination of abnormal foetuses.

This approach has helped to reduce the incidence of thalassaemia from 30 to 2 per cent a year in the Cypriot (i.e., a native or inhabitant of Cyprus) community in England. However, antenatal diagnosis may sometimes be misused, e.g., it may be used to selectively abort the foetuses of one set.

Some newspaper reports show that some parents in India are using amniocentesis and ultrasound technique (another vital tool for antenatal diagnosis) to selectively abort female foetuses, obviously to save themselves from the hardships and sufferings they have to face to arrange the exceedingly high amounts of dowry for their daughters.

Some other Duties of Genetic Counsellor:

Apart from providing counselling services to high-risk individuals, a genetic counsellor must be able to provide advice to those groups or populations which have a high incidence of certain specific diseases. For example, African people have higher incidence of sickle-cell anaemia, those of Mediterranean descent are prone to thalassaemia, and East European Jews have high probability of carrying the Tay-Sach gene.

The genetic counsellor should also try to wash away the guilt feelings of the parents of the affected child by pointing out that genetic calamities can occur in any family, and the parents are not alone in their problem and no one can escape all the possible undesirable expressions of their genotype.

Genetic counselling and antenatal diagnosis provide definite relief to the possible parents and reduce the frequency of genetically defective individuals in the population. However, it is unlikely that these measures would eliminate the deleterious (=lethal) alleles from a population.

This is so because most genetic defects are recessive and they would persist in the population through the heterozygotes; therefore, even such an extreme selection would lead to only a slow decline in their frequency.

Further, it is not likely that all the couples in any society will willingly agree themselves, at least in foreseenable near future, to these procedures. However, genetic counselling has become a routine aspect of medical practice in most developed countries.