EDITORIAL

Role of Genetic Counselling in Modern Health Care

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Not so long ago medical genetics was perceived as a highly specialized branch of medicine, of relatively little importance to the general public, and many doctors considered training in genetics a luxury. Such attitudes have largely changed today, with the recognition that many common disorders have at least partial genetic origin, and with the rapid development of powerful diagnostic tests for both chromosomal and single gene disorders and human genome sequencing achieved. Knowledge of genetics is now considered essential today for any health professional.

It is important to know as to what type of disorders are genetic? " Genetic " does not necessarily mean " hereditary ". The first term implies simply that the genetic material, on a chromosomal or a gene level, contains one or more mutations, which are the cause of the disorder. Once a mutation is present in a patient, particularly if it is constitutional (and thus present in all cells), it can ofcourse be transmitted and thus becomes a hereditary disorder.

Four types of genetic disorders have been recognised: (i)Chromosomal disorders: These affect about 1/200 live-born children and about 1/500 adults (1). (ii) Monogenic ("Mendelian") inheritance: is the result of mutations in single genes, at specific gene "loci". We have about 20-30,000 individual genes, for several thousand of which monogenic disease has been described (2). (iii) Polygenic or "multifactorial" although this causation is not" as genetic" as are monogenic and chromosomal disorders, the majority of malformations and of common familial disorders have this type of cause. (iv) Mitochondrial disorders. In recoent years, a new type of inheritance has been proven, resulting from mutations in the mitochondrial genome. The mitochondrial chromosomes can be deleted or suffer other types of mutations which interfere with cellular production of ATP (3). In many cases the mutation is "de novo" in an affected individual, but hereditary transmission is purely maternal, since, a fertilized egg's mitochondria originate from the maternal germ cell only.

Genetic counselling may be described as the process through which individuals affected by, or at risk for a problem which may be genetic or hereditary, are informed of the consequences of the disorder, of the probability of suffering from or of transmitting it to their offspring, and of the potential means of treating or of avoiding the occurrence of the malformation or disease in question. Genetic counseling in common disorders is often given by the family doctor, the pediatrician or the obstetrician. However, with the recognition that thousands of problems have a major hereditary

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component, counseling is increasingly done in specialized centers which also provide the laboratory diagnostic tools. Four aspects are involved in giving genetic counseling:

- *Arriving at a specific diagnosis :* this is often the most difficult, trying and time consuming part of the process, for the health care professionals as well as for the family.
- *Estimation of risks* : to develop the disorder and/or to transmit it to offspring.
- *Practical aid :* this includes, for example, recommending doctors for specialized examinations or health care professionals for tests e.g., prenatal and other diagnostic tests.
- *Supportive role :* this aspect is atleast as important as the diagnostic question, for the great majority of genetic disorders cannot be cured or even satisfactorily treated. However, accepting and learning to live with a genetic diagnosis is particularly difficult when reproductive options are involved, and feelings of "guilt" may touch several generations and cause rifts in the family just at the time when solidarity is most needed.

Post Human genome era has opened up new vistas in the preventive as well as curative aspects of medical profession. With variant and mutant gene sequences, it is now possible to predict the phenotype of the organisms including man. It, therefore, has become very important to diagnose the diseases before the birth of a child. Genetic counselling has an important role to play in this direction. When families come for counselling, specific questions need to be addressed e.g.,

- What is the problem ? An accurate diagnosis is the sine qua non of genetic counselling.
- What does the disorder mean ?
- What caused the abnormality?
- Is it curable or treatable ? Possible therapy its success, burden, and expense need to be discussed.
- Will the problem recur in future pregnancies ?

Approximately 2-3% of infants are born with congenital malformations, the majority of which have atleast a partially genetic basis. And if one considers the causes of illness in persons up to the age of 25 years or so, at least 5% of all individuals will suffer by adulthood from a malformation or disease with a major genetic component. (4). Thus, the comprehension and control of genetic disorders is, particularly with regard to the decline in mortality and morbidity from infectious disease, a major public health concern.

In most genetic divisions, patients can either come self-referred or be recommended by a physician. The reasons for seeking genetic counselling can be divided into the following six categories:

- The individual (often hoping to be a parent) suffers himself from a genetic or potentially genetic disease.
- A close relative has a genetic disease, and the individual who consults is worried either about his/her own risk of developing the disease or the risk that his offspring will suffer from the disorder.
- The individual is at increased genetic risk for a specific genetic disorder given his or her particular ethnic origin
- The individual or couple is having reproductive problems, e.g. infertility, repeated miscarriage, etc
- The couple has already born a child or fetus with a malformation or genetic disorder.

- The couple requests counseling, concerning prenatal diagnosis, for such reasons as advanced parental ages.

To provide both diagnostic, counselling and follow-up services, close ties must be established with such hospital departments as pediatrics and obstetrics and access to following specialized diagnostic services is essential:

- Clinical diagnosis and genetic counselling.
- Chromosomal analysis: both postnatal and prenatal diagnostics.
- DNA extraction and banking.
- DNA analysis.
- Prenatal diagnostic services.

The process of genetic counselling has changed dramatically over the past 25 years. Instead of being based on purely clinical findings, the identity of many disorders can be proven because their genic or chromosomal basis is known. The availability of an ever-increasing number of laboratory tests allows more accurate diagnosis, and often gives the opportunity for asymptomatic or prenatal diagnosis to family members who prefer to use it.

The training of individuals competent to give genetic counseling has been formalized in a number of European countries, as was done in the United States and Canada a number of years ago through their respective Boards of Medical Genetics. Medical doctors with post-graduate training in medical genetics depend heavily on cytogeneticists and molecular geneticists for diagnosis.

References

1. Robinson A, Puck T. Genetic disorders in children and young adults–a population study. *Am J Hum Genet* 1967 ; 19 : 112-29.
2. Antonarakis SE. Diagnosis of genetic disorders at DNA level. *N Engl J Med* 1989 ; 320 : 153-56.
3. Wallace DC. Choose better human genes. *TIG* 1993 ; 9 : 128-33.
4. Johnson JP. Genetic counselling using linked DNA probes. *J Pediatr* 1988 ; 113 : 957-59.

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