GENETIC COUNSELLING - THE MOTIVATING TOOL

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ABSTRACT: Genetic disorders are almost always serious, are not curable and relatively few are amenable to satisfactory modes of treatment. At the present time the most effective means of preventing genetic diseases remains the provision of genetic counselling for individuals at risk of having a child with a serious genetic disorder, coupled with prenatal diagnosis where possible. This article discusses the indications, practical aspects and effectiveness of genetic counselling with special emphasis on the psychological aspect during counselling. Molecular genetic technology coupled with appropriate genetic screening and counselling not only provides primary prevention but also helps in creating awareness regarding genetic disease and its prevention among the population.

INTRODUCTION

Genetic disorders occur among nearly 30% of admissions to Paediatric hospitals, and mortality rate is between 40 - 50%. Genetic diseases are almost always serious, are not curable and relatively few are amenable to satisfactory modes of treatment. Thus in current situation the prevention of this group of diseases remains of paramount importance. The most effective means of preventing genetic diseases remains the provision of genetic counselling for individuals at risk of having a child with a serious genetic disorder.

The basic principles of genetic counselling are that "An ounce of prevention is worth a pound of cure" and that "A child has a fundamental right to be born with a sound mind and body'.

The study of human genetics began during the early decades of the present century. Charles Davenport of the Eugenics records office in New York began giving genetic advice as early as 1910. It was during the second world war time, that the first genetic counselling clinic was opened in America at Michigan (1940) and Minnesota (1941). In U.K, the hospital for sick children on Great Ormond street London, developed the first such clinic in 1946. Sheldon Reed published his first book on counselling in Medical Genetics, in 1955.

The American Society of Human Genetics in 1975 has defined genetic counselling as follows - "Genetic counselling is a process of communication, the intent of which is to provide individuals and families having a genetic disease or at risk of such a disease with information about their condition and to provide information that would allow couples at risk to make informed reproductive decisions.

INDICATION:

Genetic counselling is valuable for patients and families with known hereditary disease. It also helps in certain conditions, where congenital disorders can be due to mixture of heredity and environmental factors. In every case, accurate diagnostic evaluation is important. Pregnancy in a woman over 35 year of age is associated with high risk of Down's Syndrome, so genetic counselling and prenatal diagnosis offered to such women can prevent the occurrence of defects. In certain conditions, although not strictly genetic, counselling can be provided for women exposed to potential teratogens during their pregnancy. The offspring of consanguineous mating are at increased risk of being homozygous for rare mutant alleles and thus being affected with rare autosomal recessive disease. It should be noted that for all pregnancies there is a risk of approximately 3% that the child would be born with a serious genetic disease at birth. (Table No. 1)