On both sides of the family, examining the affected person and sometimes other family members and arranging appropriate tests (Haematological, biochemical, chromosomal and DNA analysis).

2. Calculating risk of recurrences in the family of similar diseases. This depends on the type of disorder i.e., whether it is chromosomal, single gene or multifactorial.

3. Communication of information to the consultants. This needs adequate time, capacity of the counsellor to communicate information which is understandable to the counsultand.

Genetic Counsellor discusses with the couple information regarding the nature of disorder, its prognosis, natural history of diseases and possible complications.

Counsellor also discusses the risk for future children to have similar problem and regarding availability of prenatal diagnostic tests for the disorder. If the risk of having a child is high reproductive options available are also discussed. Genetic counselling also take into account the complex psychological and emotional factors of the counsultand and tries to allay their anxiety and guilt feelings.

4. Follow-up To know whether consultant have understood the information given to them and for continuation of education of the family members regarding the disorder. Follow-ups studies also help the parents to have answers for their questions which may arise after counselling.

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GENETIC COUNSELLING

Information Brochure

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What is Genetic Counselling?

Genetic counselling is the process by which patients or relatives at risk of a disorder that may be hereditary are advised of consequences of the disorder, the probability of developing and transmitting it and of the ways in which this can be prevented.

What are Genetic Diseases?

Genetic diseases are also called as hereditary diseases, are the most burdensome of all human afflictions and are due to changes in chromosomes, genes or due to interaction of abnormal genes and environment, Genetic diseases are determined at conception but can be expressed at any time in life span.

What are chromosomes and genes?

Human body is composed of cells which are known as units of life. Cells contain a thread like material in the nucleus called chromatin. Chromatin is made up of DNA (Deoxyribonucleic acid). Genes are units of DNA and are responsible for the expression of character in the organisms. During cell division chromatin forms, which take part in cell division resulting in two cells from a single cell. The chromosome number in human being is 46, chromosomes occur in pairs, one derived from each parent. Out of 46 chromosomes 44 or 22 pairs are called autosomes and the remaining one pair (X and Y) are called Sex chromosomes and these chromosomes determine the Sex of the individual. In males the Sex chromosome are "XY" and in females it is "XX".

How any types of Genetic Diseases are there?

Genetic diseases can be divided into three main groups (i) Chromosomal disorders-due to abnormality in chromosome structure or number for e.g: Down's Syndrome (Three 21 chromosomes instead of normal 2). (ii) Single gene disorders-due to changes in the genes present on autosomes or Sex chromosomes (iii) Multifactorial disorders-These are due to the effect of many genes and partly environmental in causation e.g. common birth defects.

Since most of the genetic diseases are not completely curable so prevention of these diseases is important. The methods available counselling include, screening for genetic diseases in population, Genetic counselling and prenatal diagnosis.

Who need genetic counselling?

1. Those who have had a child or family member with
   a) Intellectual Disabilities.
   b) Multiple birth defects.
   c) Anemia nor responding to treatment.
   d) repeated episode of bleeding after injury.
   e) convulsion
   f) growth retardation
   g) Certain types of skin diseases.

2. Couples with history of
   a) blood related marriages.
   b) repeated abortions (More than 3)
   c) still births
   d) malformed babies.

3. If there is history of diabetes during pregnancy.
4. When there is any other familial diseases.

Steps in Genetic Counselling

1. The first step in genetic counselling is to establish a definite diagnosis. This involves enquiring into medical history of affected person, reviewing relevant medical records. obtaining family history