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Research Article

GENETIC COUNSELLING

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ABSTRACT

Genetic counselling is a service that provides information and advice about genetic condition. These are condition caused by changes known as mutation in certain genes and are usually passed down through a family. Genetic counselling is the process through which knowledge about the genetic aspects of illnesses is shared by trained professionals with those who are at an increased risk or either having a heritable disorder or of passing it on to their unborn offspring. A genetic counsellor provides information on the inheritance of illnesses and their recurrence risks; addresses the concerns of patients, their families, and their health care providers; and supports patients and their families dealing with these illnesses. The Heredity Clinic was the first genetic counselling service centre established in 1940 at the University of Michigan, USA. Since then, the many such centers have been opened around the world.

Keywords: Genetics, Counselling, Heriditary, Congenital malformations, Chromosomal abnormalities, Genetic disorder

Introduction:

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 counselling in common disorders is often given by the family doctor, the paediatrician or the obstetrician.

History and evolution of genetic counseling

"Sheldon Clark Reed" coined the term genetic counselling in 1947 and published the book "Counselling in Medical Genetics" in 1955. Most of the early genetic counselling clinics were run by non- medical scientists or by those who were not experienced clinicians. With the growth in knowledge of genetic disorders and the appearance of medical genetics as a distinct specialty in the 1960s, genetic counselling progressively became medicalized, representing one of the key components of clinical genetics. It was not, though, until later that the importance of a firm psychological basis was recognized and became an essential part of genetic counselling, the writings of Seymour Kessler making a particular contribution to this. The first master's degree genetic counselling program in the United States was founded in 1969 at Sarah Lawrence College in Bronxville, New York. In 1979, the National Society of Genetic Counsellors (NSGC) was founded.

General Concept:

Genetic counselling is a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial, and reproductive implications of the genetic contribution to specific health conditions. A communication process that deals with human problems associated with the occurrence or the risk of occurrence of genetic disorders in individuals or families. Genetic counselling is communicative process which deals with human problems associated with occurrence and or recurrence of a genetic disorder in a family

Aims of genetic counseling

The genetic counselling aims to provide the family with complete and accurate information about genetic disorders.

1. Promoting informed decisions by involved family members
2. Clarifying the family's options available treatment and prognosis
3. Explaining alternatives to reduce the risk of genetic disorders
4. Decreasing the incidence of genetic disorders

Reducing the impact of the disorders

Purpose of genetic counseling

- Explaining alternatives to reduce the risk of genetic disorders.
- Reducing the impact of genetic disorders.
- Assisting families in choosing the options most appropriate for them.
- To comprehend the medical facts, including diagnostic and available management. Discussing the options available for dealing with the disorder.
- Provide concrete, accurate information about inherited disorders.
- Reassure people who are concerned that their child may inherit a particular disorder
- Allow people who are affected by inherited disease to make informed choice about future.
- Educate people about inherited disorder and the process of inheritance.
- Offer support by skilled health care professionals to people who are affected by genetic.
- To comprehend the medical facts, including diagnostic, probable course of the disorder
- To appreciate the way hereditary contributes.
- To understand the option for dealing with the risk occurrence.
- Possible adjustment to the disorder in an affected family member.

Indications of genetic counselling

1. Advanced parental age:
- Maternal age ≥35 yrs
- Paternal age ≥50 yrs

2. Previous child with or family H/O:

- Hereditary disease in a patient or family
- Congenital anomaly
- Dysmorphism
- Intellectual disability
- Developmental delay
- Isolated birth defect
- Metabolic disorder
- Chromosomal abnormality
- Myopathy/ Neuropathy
- Ambiguous genitalia

3. Adult-onset genetic disorder (pre symptomatic testing)
- Cancer - Inherited a tendency to develop cancer

4. Consanguinity - Couples are blood relatives.
5. Teratogen exposure
6. Repeated pregnancy loss or infertility
7. Pregnancy screening abnormality
- Maternal serum α-foeto protein
- Maternal triple or quad test
- Fetal ultrasonography
- Fetal karyotype

8. Heterozygote screening based on ethnic risk
- Sickle cell anemia
- Tay- Sachs, Canavan, Gaucher disease
- Thalassemia

9. Follow up to abnormal neonatal genetic testing
10. Other Indication:

- Birth defects
- Mental retardation
- Miscarriages
- Malformations
- Tendency to develop a neurologic condition
- Birth defects and genetic condition.
- Child with defects / genetic condition.
- Child with developmental delay

Mental retardation and other problems with growth and developmen

Types of genetic counselling
They are of 2 types:
Prospective genetic counselling:
This allows for the true prevention of disease. This approach requires

Retrospective genetic counselling:

- Most genetic counselling at present is retrospective, i.e, the hereditary disorder has already occurred within the family.
- The methods which could be suggested under retrospective genetic counselling are: Contraception and Pregnancy termination.

Types of genetic screening
A search in apparently normal population for individual with abnormal genes which increase their risk or their offspring of being affected by a disease.

1. Carrier identification
It is possible to identify the healthy carriers of a number of genetic disorders, especially the inborn errors of metabolism

2. Prenatal diagnosis
Prenatal diagnosis forms an integral step in genetic counselling. In fact, for couples at risk of a disorder, it is desirable to consider, plan

and discuss prenatal diagnosis even before pregnancy. Discussion and planning beforehand will eliminate hurried procedures and emotional trauma as well.

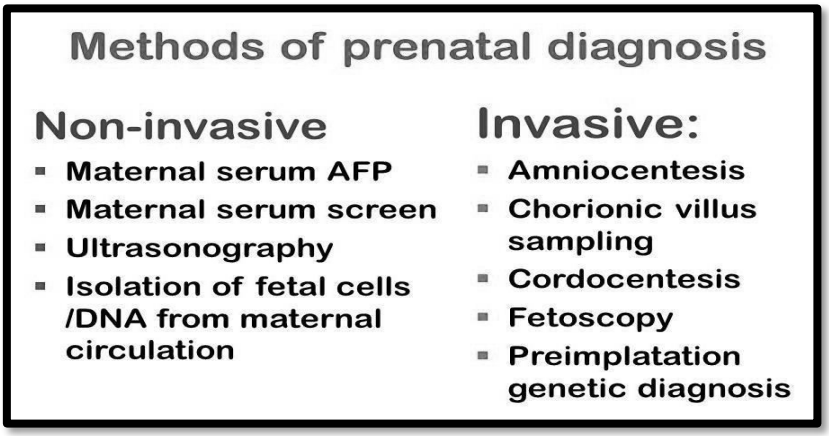

Figure 40: Approaches to prenatal diagnosis

3. Newborn screening

Newborn screening is the practice of testing all babies in their first days of life for certain disorders and conditions that can hinder their normal development. This testing is required in every state and is typically performed before the baby leaves the hospital.

Forensic screening (paternity test)

Paternity tests consist of determining the genetic maps that belong to the two people who undergo the analysis. By comparing the genetic map of the suspected father with that of the child, it is possible to determine their biological kinship.

Diagnosis-based on accurate family history

This is primary beginning phase of counselling in which following tasks are accomplished

- Initial interview with counselee & family for preparation of counselee for counseling.
- Carryout primary assessment of counselee, physical examination etc.
- Considering potential diagnosis based on collected information.

Examination & Investigation

- Most crucial step in any genetic consultation; involves History taking, Examination and Investigation
- In some cases, the goal of genetic evaluation is to make a diagnosis of genetic condition/ syndrome.

- In other cases, the diagnosis already is known, and the genetic counselor probably will confirm the established diagnosis to proceed for next phases of the counseling.

- There are two investigations are said to done as follows:

1. Confirmatory /Supplementing Tests

a. Chromosomal analysis

b. Biochemical tests

c. Molecular DNA testing

d. X Rays, biopsy

e. Immunological test

f. Prenatal diagnosis

g. Linkage analysis

2. Establishment of an Accurate Diagnosis Risk Assessment

This phase includes followings tasks.

Communication

- Communication of results and risk to the counselee and to the family if appropriate.
- Discussion of natural history of disorders.
- Current treatment options and anticipatory guidance.
- Assess the counselee's understanding about facts and relevant hereditary pattern, diagnostic and management options for disorder.

Discussion of Option

- Consultants should be provided with all of the information necessary for them to make their own informed decisions.
- It should include details of all the options available
- If relevant, the availability of prenatal diagnosis should be discussed, together with details of the techniques, limitations and risks associated with the various methods employed
- Various reproductive options –donor sperm, donor ova or PGD Long Term Contact and Support:
- It should present information clearly in sympathetic and appropriate manner
- It be receptive to the fears and aspirations, expressed or unexpressed
- The setting should be agreeable, private and quiet, with ample time for discussion and questions
- Technical terms should be avoided or, if used, fully explained

- Questions should be answered openly and honestly
- Written summary for follow up
- Should contact the consultant at later date
- Should refer to appropriate patient support groups

Pre-requisites of genetic counselling

- Detailed family history.
- Accurate diagnosis.
- Understanding medical aspect of disorder (etiology, history, treatment, prognosis, burden).
- Understanding the inheritance pattern (recurrence risk)
- Understanding the psycho-social impact of the information.
- Training / experience in counselling techniques.
- Understanding the concepts of health / disease / healthcare in the appropriate cultures.

Role of nurses/midwives in genetic counselling

- Receive the client and family and make them comfortable in assessment room for genetic counseling.
- Obtain prenatal, family, and other health histories from individual and family.
- Conduct primary physical information and collect other relevant information.
- Provide psychological support to individual and family throughout the counseling.
- Provide information about hereditary pattern.
- Collect other relevant information from individual and family e.g. – any prior test report and documents others.
- Encourage the individual and other family members to ask questions as much as they can understand about all aspects of disorders.
- Establish a plan of care with the family and coordination care with the family and other healthcare professionals.
- Maintain privacy and confidentiality of all the information.
- Provide referral guidance.
- Follow up care

Recent trends in genetic counselling

Genetic counselors fill a distinctive position in the complicated and varied arena of genomic medicine and health.

- Advances in genetic medicine create an even greater demand for expert health care services.

- Genetic counselors help meet this need, serving in almost every major medical center and across the globe as an increasingly important resource for medical referral and quality patient care.
- For an international list of genetic counselors and further information about genetic counseling, visit the website of the National Society of Genetic Counselors (www.nsgc.org).

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