




Genetic Counseling

Pilar S. Bracero, MS




What is Genetic Counseling?



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

From Practical Genetic Counseling by Peter S Harper




Genetic counseling is an educational process that seeks to assist affected and/or at risk individual to understand the nature of the genetic disorder, its transmission and the options open to them in management and family planning


From *Clinical Genetics and Genetic Counseling* by T.E. Kelly




The Genetic Counselor's Job



Genetic counselors work as members of a health care team, which may include specially trained doctors, social workers, nurses, medical geneticists, or other specialists to help families make informed decisions about their health.



Genetic counselors provide information and support to families that have members who have birth defects or genetic disorders, or who may be at risk for a variety of inherited conditions.




Genetic counselors identify families at risk; interpret information about the disorder; and analyze inheritance patterns and risks of recurrence.

This includes interpretation of family and medical histories.



Genetic counselors review available genetic testing options with the family.

Genetic counselors discuss the risks, benefits and limitations of genetic testing; review available options with families and provide supportive counseling.



Genetic counselors are skilled in translating the complex language of genomic medicine into terms that are easy to understand.



Genetic counselors also serve as patient advocates, educators, administrators, researchers and resource people for health care professionals and the public.

From The Joan H. Marks Graduate Program in Human Genetics




History



First genetic clinics started in 1940 in Michigan.

First genetic counseling program started in 1969 at Sarah Lawrence College.



Genetic counselors traditionally have a Master of Science degree in genetic counseling.



Genetics and Healthcare in 21st Century



**Human Genome Program has brought
inherited diseases to the forefront.**



Genetic risk assessment, screening, and testing is becoming part of primary care.

- **Primary care**

 - Identification of those at risk

- **Tertiary care**

 - Refinement of risk



Clinical genetics and primary care need to work together to provide appropriate services.

- **Primary care**


Screening

- **Tertiary care**

Diagnosis




Counseling session



The initial contact phase is when the counselor and families meet and build rapport.


- Obtain a family history (pedigree)
- Address issues



The encounter phase includes dialogue between the counselor and the client about the nature of screening and diagnostic tests.

Explain genetics


- chromosomes
- genes
- inheritance patterns



The summary phase provides all the options and decisions available for the next step.

If clients wish to go ahead with testing, an appointment is scheduled.

The genetic counselor acts as the person to communicate the results.



The counselor's job is to present all options, then allow the client to make decision.

Many factors involved:

- religion
- previous history
- family support

Patient Centered Care

Institute of Medicine (IOM) defines patient-centered care as:

“Health care that establishes a partnership among practitioners, patients, and their families (when appropriate) to ensure that decisions respect patients' wants, needs, and preferences and that patients have the education and support they need to make decisions and participate in their own care.”



Importance of Genetic Counseling



Education, Education, Education