Chapter 5 : Genetic Counseling

As members of a health care team, genetic counselors provide information and support to families affected by or at risk of a genetic disorder. They serve as a central resource of information about genetic disorders for other health care professionals, patients, and the general public. This chapter provides an overview of the role of genetic counselors, and their approach to educating patients and identifying individuals/families at risk of a genetic disorder. In addition, some useful resources are provided where patients may be referred to for additional information.

Genetic counselors play an important role in providing expert genetic services. They are trained to present often complex and difficult-to-comprehend information to families and patients about genetic risks, testing, and diagnosis; discuss available options; and provide counseling services and referrals to educational and support services.

5.1 Role of Genetic Counseling

Genetic counselors work as part of a health care team, providing information and support to families affected by or at risk of a genetic disorder. They help to identify families at possible risk of a genetic disorder, gather and analyze family history and inheritance patterns, calculate risks of recurrence, and provide information about genetic testing and related procedures. In particular, genetic counselors can help families to understand the significance of genetic disorders in the context of cultural, personal, and familial situations. Genetic counselors also provide supportive counseling services, serve as patient advocates, and refer individuals and families to other health professionals and community or state support services. They serve as a central resource of information about genetic disorders for other health care professionals, patients, and the general public.

The most common indications for genetic counseling include advanced maternal age, family history of a genetic condition, and suspected diagnosis of a genetic condition. **For more information about genetic counseling or to find a genetic counselor in your area, please see the National Society of Genetic Counselors' web-site at *http://www.nsgc.org*.**

5.2 Process of Genetic Counseling

In general, a genetic counseling session aims to:

- Increase the family's understanding about a genetic disease(s), the risks and benefits of genetic testing and disease management, and available options.
- Identify with the individual and family the psychosocial tools required to adjust to potential outcomes.
- Reduce the family's anxiety.

It is not unusual for multiple genetic counseling sessions to occur and, at a minimum, include a pre-testing and post-testing session. During the initial genetic counseling visit, the genetic counselor will determine why the patient/family is seeking genetic counseling, identify what information they wish to obtain from the session, collect and record a family history, and assess and record the psychosocial history of the patient.

Among the topics discussed during a pre-test session are the clinical presentation of the condition(s) the patient may be at risk for, the pattern of genetic inheritance of the condition, risk of recurrence, available testing procedures and test limitations, reproductive options, and follow-up procedures if needed. General questions relating to suggested treatment, therapy, and the function of related proteins are also addressed. Referrals may be made to specialists regarding specific issues which fall outside the scope of genetic counseling practice.

If the patient decides to have genetic testing performed, the genetic counselor is often the point person to communicate the results to the patient/family. However, the post-test session involves more than the provision of medical information and often focuses on helping families cope with the emotional, psychological, medical, social, and economic consequences of the test results. In particular, psychological issues such as denial, anxiety, anger, grief, guilt, or blame are addressed and, when necessary, referrals for in-depth counseling are offered. Information about community resources and support groups are provided to the patient/family.

If the genetic test is positive, testing should be considered in additional relatives of this individual. Genetic counseling referrals for other family members for risk assessment are then discussed and it may be necessary to refer relatives to other genetic counselors due to geographical and other constraints.

At the conclusion of the genetic counseling sessions, the patient should be offered a written summary of the major topics discussed. The summary is often provided in the form of a letter which serves as a permanent record of the relevant information discussed, as well as relaying additional information that may have become available after the final counseling session. The patient may also choose to share the letter with other family members.

5.3 Patient Education

Patients rely most upon their primary health care providers for information related to their condition. In general, though, your patients will require information you may not have. Before providing patients with any educational materials, please be sure to review that the information is produced by a credible source and is current.

Books and pamphlets are most widely distributed and appreciated by patients, even by patients who are web-savvy. Patient advocacy groups generally provide the best and most up-to-date information on specific conditions. The organizations listed below are excellent sources of information about genetic diseases that can be helpful to patients:

Genetic Alliance
4301 Connecticut Ave, NW
Suite 404
Washington, DC 20008
Ph: (202) 966-5557
Fax: (202) 966-8553
URL: ***http://:www.geneticalliance.org***
E-Mail: info@geneticalliance.org

Genetic and Rare Diseases Information Center (GARD)
P.O. Box 8126
Gaithersburg, MD 20898-8126
Ph: (888) 205-2311
TTY: (888) 205-3223
Fax: (240) 632-9164
URL: ***http://www.genome.gov/Health/GARD***
E-mail: GARDinfo@nih.gov

National Organization of Rare Diseases (NORD)
55 Kenosia Avenue
PO Box 1968
Danbury, CT 06813
Ph: (203) 744-0100
TTY: (203) 797-9590
Fax: (203) 798-2291
URL: ***http://www.rarediseases.org/***
E-mail: orphan@rarediseases.org

Selected References

Genetic Alliance–Disease InfoSearch *http://www.geneticalliance.org/ws_display.asp?filter=diseases*
Provides accurate and reliable information developed by the advocacy organizations which form the Genetic Alliance. Users can search for information about advocacy support groups related to specific genetic conditions, the clinical features of a wide number of genetic conditions, and updates on management, treatment, and other related topics.

International Society of Nurses in Genetics *http://www.ISONG.org*

March of Dimes *www.marchofdimes.com* (Spanish *http://www.nacersano.org/*)
Provides information about improving the health of babies by preventing birth defects, premature birth, and infant mortality.

MedlinePlus *http://www.nlm.nih.gov/medlineplus/*
MedlinePlus has extensive information from the National Institutes of Health and other trusted sources on over 700 diseases and conditions. There are also lists of hospitals and physicians, a medical encyclopedia and a medical dictionary, health information in Spanish, extensive information on prescription and nonprescription drugs, health information from the media, and links to ongoing clinical trials.

National Human Genome Research Institute–Health *http://genome.gov/Health/*
The site provides useful information about basic genetics concepts, genetic conditions, current research, and valuable tools to help make genetics an important tool in determining health.