As members of a healthcare team, genetic counselors provide information and support to families affected by or at risk for a genetic disorder. They serve as a central resource of information about genetic disorders for other healthcare 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. Patient resources are also provided.
5.1 Role of Genetic Counseling

Genetic counselors help identify families at possible risk of a genetic condition by gathering and analyzing family history and inheritance patterns and calculating chances of recurrence. They provide information about genetic testing and related procedures. They are trained to present complex and difficult-to-comprehend information about genetic risks, testing, and diagnosis to families and patients. Genetic counselors can help families understand the significance of genetic conditions in relation to cultural, personal, and familial contexts. They also discuss available options and can provide referrals to educational services, advocacy and support groups, other health professionals, and community or state services. Genetic counselors can serve as a central resource of information about genetic conditions for other healthcare professionals, patients, and the general public. (See Appendix O for Making Sense of Your Genes: A Guide to Genetic Counseling.)

5.2 Process of Genetic Counseling

In general, a genetic counseling session aims to:

- Increase the family’s understanding of a genetic condition
- Discuss options regarding disease management and the risks and benefits of further testing and other options
- Help the individual and family identify the psychosocial tools required to cope with potential outcomes
- Reduce the family’s anxiety

It is not unusual for multiple genetic counseling sessions to occur and, at a minimum, to 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 medical history, and assess and record the medical and psychosocial history of the patient.

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

If the patient decides to have genetic testing performed, the genetic counselor often acts as the point person to communicate the results. 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 psychosocial counseling are offered. Information about community resources and support groups can be provided to the patient/family.

If the genetic test is positive, testing may be considered for additional relatives of the individual. Genetic counseling referrals for other family members for risk assessment may be discussed. It may be necessary to refer relatives to other genetic counselors due to geographical and other constraints.
At the conclusion of the final genetic counseling session, the patient may receive 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 information discussed and can include additional information that became available after the final counseling session. The patient may choose to share the letter with other family members or healthcare providers.

5.3 Patient Education

Many patients rely heavily on their primary healthcare providers for information related to their condition. In general, though, patients will require information providers may not have. Before providing patients with any educational materials, providers should be sure to check that the information is current and produced by a credible source.

Books and pamphlets are appreciated by patients, even those who are web-savvy. Patient advocacy groups generally provide the best and most up-to-date information. The organizations listed on the following page are excellent sources of information about genetic diseases that can be helpful to patients.
Selected References

American College of Medical Genetics, Newborn Screening Act Sheets and Confirmatory Algorithms
www.acmg.net/resources/policies/act/condition-analyte-links.htm

Genetic Alliance Disease InfoSearch
www.geneticalliance.org/dis

International Society of Nurses in Genetics
www.isong.org

March of Dimes
www.marchofdimes.com (Spanish at www.nacersano.org)

MedlinePlus
www.nlm.nih.gov/medlineplus

National Human Genome Research Institute–Health
www.genome.gov/health

National Society of Genetic Counselors (NSGC)
www.nsgc.org

Genetic Alliance
4301 Connecticut Avenue, NW, Suite 404
Washington, DC 20008
Ph: 202.966.5557
Fax: 202.966.8553
Email: info@geneticalliance.org
www.geneticalliance.org

Genetic and Rare Diseases Information Center (GARD)
P.O. Box 8126
Gaithersburg, MD 20898
Ph: 888.205.2311
TTY: 888.205.3223
Fax: 240.652.9164
Email: GARDinfo@nih.gov
www.genome.gov/health/gard

Genetics Home Reference
Reference and Web Services
National Library of Medicine
8600 Rockville Pike
Bethesda, MD 20894
Ph: 888.346.3656
Fax: 301.496.2809
Email: custserv@nlm.nih.gov

National Organization of Rare Diseases (NORD)
55 Kenosia Avenue, P.O. Box 1968
Danbury, CT 06813
Ph: 203.744.0100
TTY: 203.797.9590
Fax: 203.798.2291
Email: orphan@rarediseases.org
www.rarediseases.org