**GENETIC COUNSELING**

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

**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.