GENETIC COUNSELING

What Is Genetic Counseling?

The National Society of Genetic Counselors (NSGC) defines genetic counseling as the process of assisting people with understanding and adapting to the medical, psychological, and familial implications of genetic contributions to disease (National Society of Genetic Counselors, 2012). This process includes the interpretation of family and medical histories to assess the chance of disease occurrence or recurrence. Genetic counseling usually involves providing education about inheritance, testing options, disease management, and prevention. Genetic counseling also promotes informed choices and adaptation to the risk or condition (National Society of Genetic Counselors, 2012). Counseling usually has a specific therapeutic focus such as prenatal, pediatric, psychiatric, or cancer genetic counseling. The aim of genetic counseling is to support individuals in their ability to make an informed decision regarding genetic testing and the implications of such testing (Sequeiros & Guimarães, 2007).

Individuals seek genetic counseling for a variety of reasons. People who have questions about origins of diseases or traits in their family or ethnic group are typical clients of genetic counseling. Those who may find genetic counseling helpful include those who have, or are concerned they might have, an inherited disorder or birth defect. Physicians also refer pregnant women whose ultrasound examinations or blood testing indicate that their pregnancy may be at increased risk for complications or disability, as well as women over 35 who are pregnant. Couples who already have a child with a genetic disability or who give birth to infants diagnosed with a genetic disease by routine newborn screening may also seek genetic counseling (March of Dimes, 2012).

Who Provides Genetic Counseling?

Genetic counselors are health professionals with specific education, training and experience in medical genetics and counseling (National Society of Genetic Counselors, 2006). Genetic counselors usually work as part of a healthcare team, providing information and support to families who have members with birth defects or genetic disorders and to families who may be at risk for a variety of inherited conditions (National Society of Genetic Counselors' Definition Task Force, 2006). Genetic counselors interact with clients and other healthcare professionals in an assortment of clinical and nonclinical settings, such as university-based medical centers, private hospitals, private practice, and industry settings (American Board of Genetic Counseling, 2012). More and more, primary care practitioners are providing facets of genetic counseling and genetic services, resulting in a need to train nurses, social workers, and physicians. Genetic counselors provide a critical role in educating providers and developing standards of practice. Genetic counselors also afford health professionals and patients the opportunity to communicate with others, such as policymakers and the media, about new genetic services and technologies (National Human Genome Research Institute, 2012).

What Does Genetic Counseling Consist Of?

Genetic counselors assess the risk of occurrence or recurrence of a genetic condition or birth defect using a variety of techniques, including knowledge of inheritance patterns, epidemiologic data, and evaluation of clinical data. They obtain and review medical and family histories and explain the nature of genetics evaluation to clients. They explain medical information regarding the diagnosis or potential occurrence of a genetic condition or birth anomaly (National Society of Genetic Counselors' Definition Task Force, 2006). Also discussed are potential treatment options and possibilities and limitations of tests and assessments in determining the genetic status of the client. Prenatal diagnosis using cytogenetic or biochemical analyses of fetal cells, amniotic fluid, or mother's blood can provide distinct answers to genetic questions (Zych, 2008). In particular, populations where the probability of genetic disorders, such as Tay-Sachs disease or sickle cell anemia, is remarkably high, screening programs have been organized to counsel clients before they start families.

The discovery of disease and susceptibility genes brought forth by the sequencing of the human genome has brought challenges to the field of genetic counseling. The traditional role of genetic counseling has significantly widened to address a diversity of developing needs, ranging from individuals looking for disease susceptibility testing to those looking to find out if a therapeutic treatment option is the right one for them (National Human Genome Research Institute, 2012).

What Common Types of Genetic Disorders Are Discussed and Considered in Genetic Counseling?

Genetic disorders may be detected at any time during the lifespan; however, most disorders are detected during the gestational period or soon after a child is born. Results during the gestational period may reveal Down syndrome or spina bifida, while postnatal testing may reveal phenylketonuria or hypothyroidism. Most disorders occur when one or both of the parents pass on their genes. Couples who are carriers may choose to have their DNA tested before conception to determine if they are carriers for Tay Sachs, cystic fibrosis, or Huntington's disease.

Encyclopedia of Special Education, edited by Cecil R. Reynolds, Kimberly J. Vannest, and Elaine Fletcher-Janzen.

Ethics and Genetic Counseling

Genetic counselors must communicate not only the risks of prenatal testing but also the significance of such testing and the potential for therapeutic intervention (National Human Genome Research, 2012). Most people have limited knowledge about disabilities and prenatal decisionmaking during the screening process occurs within a limited timeline, while requiring families to learn new medical information (Roberts, Stough, & Parrish, 2002). These choices are laden with uncertainty and raise challenging ethical, legal and social issues (National Human Genome Research Institute, 2012). Genetic counselors are trained to facilitate decision-making to promote informed choices. Genetic information can have profound psychological meaning for clients, particularly for members of families affected by a genetic condition (National Human Genome Research Institute, 2012).

REFERENCES

American Board of Genetic Counseling (2012). *Genetic counselors' scope of practice*. Retrieved from http://www.abgc.net/docs/GC_Scope_of_prractice_final.pdf

March of Dimes (2012). *Genetic counseling*. Retrieved from http://www.marchofdimes.com/pregnancy/trying_geneticcounseling.html

National Society of Genetic Counselors' Definition Task Force. (2006). A new definition of genetic counseling: National Society of Genetic Counselors' Task Force report. *Journal of Genetic Counseling*, *15*(2), 77–82.

National Society of Genetic Counselors (2012). *Genetic counseling as a profession*. Retrieved from http://www.nsgc.org/Home/ConsumerHomePage/PatientFAQs/tabid/338/Default.aspx

National Human Genome Research Institute. (2012). *The Johns Hopkins University/National Human Genome Research Institute Genetic Counseling Training Program: Introduction*. Retrieved from http://www.genome.gov/10001156

Roberts, C. D., Stough, L. M., & Parrish, L. H. (2002). The role of genetic counseling in the elective termination of pregnancies involving fetuses with disabilities. *Journal of Special Education*, *36*(1), 48–55.

Sequeiros, J., & Guimarães, B. (2007). Definitions of genetic testing. *EuroGentest Network of Excellence Project*. Retrieved from http://www.eurogentest.org/web/files/public/unit3/DefinitionsGeneticTesting-3rdDraf18Jan07.pdf

Zych, K. A. (2008). Genetic counseling. In E. Fletcher-Janzen & C. R. Reynolds (Eds.), *Encyclopedia of special education* (3rd ed.). Hoboken, NJ: Wiley.

Laura M. Stough
Marcia L. Montague
Texas A&M University