

GENECARE

*By Philip Buchanan, PhD
and Virginia W. Norris, MGC, CGC*

Who are Genetic Counselors?

Genetic counselors are specially trained medical professionals who provide information and support to individuals and families who have an increased risk for a genetic condition or of having a child with a birth defect or genetic condition. Genetic counselors are a valuable member of a team of medical specialists, which can include pediatricians, obstetricians, oncologists, neurologists, endocrinologists, cardiologists, nurses and many others. Genetic counselors provide information and supportive counseling, coordinate testing, and connect families with community resources.

As more genes are identified by the Human Genome Project, more testing and treatments for specific genetic disorders will become available. The genetic counselor will become an even more valuable resource for individuals. A genetic counselor can review personal and family medical histories to determine available testing and provide specific information to the patient. A counselor will also discuss the many implications of testing. Huntington disorder (HD) is an excellent example to demonstrate the implications of testing.

Beth is a 35-year-old woman who has been referred for genetic counseling because of her increased maternal age. While taking a detailed family history, the counselor learns Beth's paternal grandmother died of HD. Beth's father is 56 and does not have any symptoms of HD. Beth's paternal aunt is 61 and has recently developed some early symptoms of HD. Beth's other paternal aunt is 59 and does not have any symptoms of HD. Beth is concerned about her risk of developing the disorder. She thinks her father has a 50 percent risk of developing it; and, if he does, she also has a 50 percent chance of developing the disorder.

The counselor discusses the inheritance of HD and confirms Beth's knowledge about her father's 50 percent risk. Beth reports her father does not want to know if he will develop HD. The counselor discusses with Beth some of the issues surrounding testing. The first issue is how a positive test result may affect her life knowing she has inherited this disease. If she learns she has inherited HD, she will automatically know her father also has inherited the disorder. How will she be able to keep this information to herself? Having a positive test result may affect her insurance. Does she want her insurance company to have this information? How will positive results affect her self-image? A negative test result means she will not develop the disease, but how will she feel if her older sister inherits the disease?

Beth decides to pursue testing, so her prenatal genetic counselor refers her to a special Huntington disorder clinic to meet with their genetic counselor. After a lengthy meeting with this second counselor, Beth and her husband have a change of mind and heart, and decide *not* to pursue testing. The knowledge that she and her father are destined to develop Huntington Disorder would be too difficult a burden to live with, Beth decides. She is aware she can pursue testing at a later date, should she change her mind.

Genetic counselors work with many emotional issues in all areas of medicine. Genetic counselors are trained to impart knowledge and to empower patients to make their own decisions.

Dr. Philip Buchanan is the director of GeneCare Medical Genetics Center. Ginger Norris is a certified genetic counselor at GeneCare. GeneCare provides genetic counseling, screening and laboratory services. For more information, contact their website: www.genecare.com or call (919) 942-0021 or (800) 277-4363.