Pediatric Genetic Counselor

Interview by Amy Entwisle

Carolyn Applegate

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In college, Carolyn Applegate studied biology and psychology, a perfect combination, as it turns out, for someone who wants to pursue a career in genetic counseling. Here, she talks about the unique combination of science and counseling inherent in the job, the rapidly changing nature of the field, and why the most difficult part of her job is also the most rewarding.

How did you become interested in genetic counseling?

I remember hearing about genetic counseling in my high school biology class. I kept it in the back of my mind, and then when I was in college, I realized that, in addition to medicine and science, what I really enjoyed was interacting with patients and helping them understand and deal with the medical conditions they were faced with.

Why did you decide to pursue pediatric genetic counseling in particular?

Genetic counselors generally work with three kinds of patients: couples at risk for having a child with a serious, genetically based health problem; parents of children with a genetic disorder; and adults at risk for a genetically linked, adult-onset condition like Alzheimer's disease or certain types of cancer.

I happen to love working with kids and helping parents plan for how to care for their child. And because I see kids with a range of rare genetic diseases, I'm constantly learning.

What's your typical day like?

One day a week, I see patients in clinic. Typically, they've been referred by a pediatrician, developmental pediatrician, or other specialist. I find out what their symptoms are, which specialists they've seen, and how they've been treated. Then I work with the physician to determine the best approach to testing.

Genetic testing is very expensive and fairly complex, and there are many different levels. I explain why we're doing particular testing, what it can and cannot detect, and what our plan will be if the test is negative. If it's positive, I explain the implications. For example, since about 15 percent of children with a developmental delay or autism have a chromosomal abnormality, I always recommend a chromosomal microarray analysis. If an abnormality is found, we make additional recommendations based on the genes and their function within the abnormal chromosome region. We may recommend that a patient see an eye doctor, or have an ultrasound of the heart, for instance.

For the most part, I spend the other days answering phone calls from parents and following up on those test results. I meet with families to go over the results and what they mean. I also do inpatient consultations, working with physicians to see children who are in the hospital.

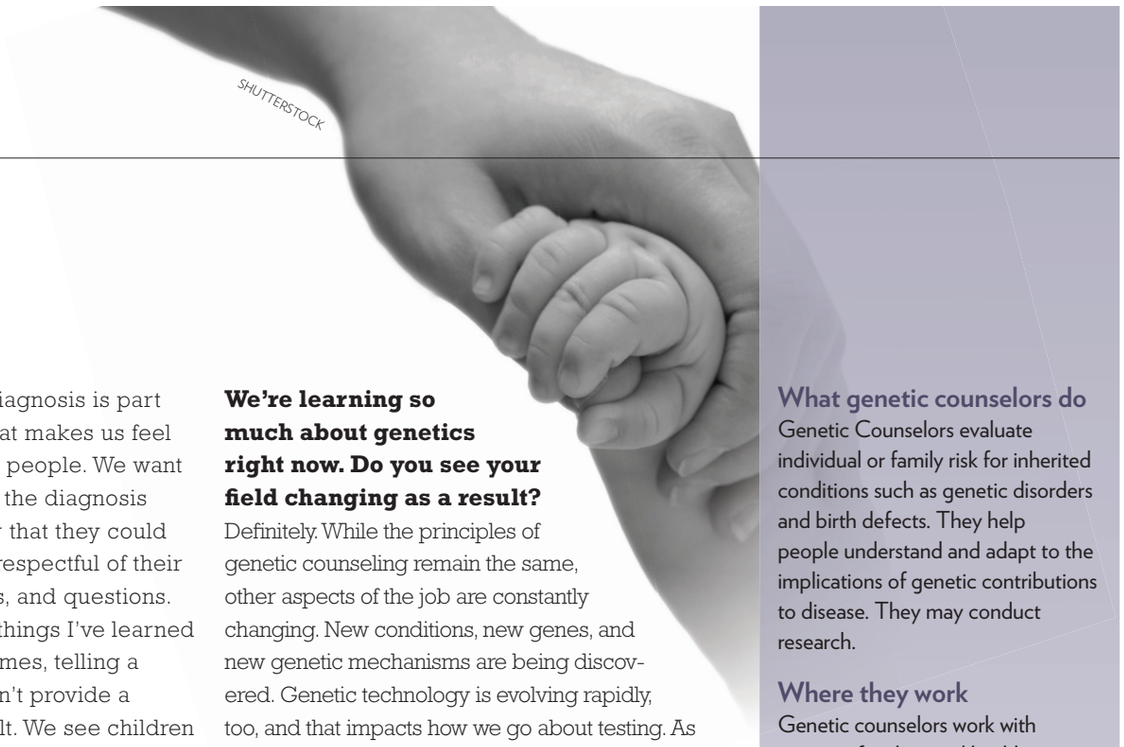
Who informs the families of a diagnosis once it's been determined?

One of the members of the medical team—the genetic counselor, the medical geneticist, or another physician involved in the child's care—tells the family the diagnosis. If it's something that's obvious in clinic, the doctor might say, "This is what I think it is," but then I follow up with them to explain in more detail what that diagnosis means, inform them of any associated medical conditions, recommended surveillance, prognosis, and if available, treatment options.

As an example, a one-year-old was referred to us for a developmental delay. When I walked into the room, the child reached out for me to hold her. She reached out to the doctor, and then the genetic counseling student. This is an unusual reaction for a one year-old to have: usually at this age, children want to be in the arms of their parents, particularly at the doctor's office! Her very outgoing personality, combined with specific facial features, made it obvious to us that the child had Williams syndrome, which is caused by a missing piece of chromosome 7. We ordered testing to confirm the diagnosis, but we were able to tell the family at that visit that their child most likely had Williams syndrome.

It must be challenging to have to share such news with families.

Because it can be emotionally and psychologically draining, it is helpful to have colleagues and friends to



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talk to. But sharing a difficult diagnosis is part of our training, and it's also what makes us feel like we're doing something for people. We want them to feel that they received the diagnosis in a caring, supportive manner that they could understand, and that we were respectful of their emotional state, beliefs, wishes, and questions.

One of the most interesting things I've learned through my work is that sometimes, telling a family that their child's test didn't provide a diagnosis can be just as difficult. We see children who we're certain have a genetic condition, but we haven't been able to identify the specific gene and/or mutation that causes it. This is often difficult for families to accept because they have to deal with the uncertainty, versus having a diagnosis and an understanding of what to expect based on what others with the same condition have experienced.

Are you involved in research?

Yes. Sometimes we enroll the family in a study or connect them to studies related to their child's condition. In one study, Hopkins is doing whole exome sequencing (looking only at protein-coding parts of the genome) on patients. We're enrolling families who have known genetic conditions with the goal of finding genes not previously known to be associated with genetic disorders.

The genetic counselors also have a spinoff project where we're looking at the psychosocial aspects of whole exome sequencing. Right now we're focusing on the consent aspect. We typically spend a half-hour to an hour with families explaining the process and obtaining consent for studies, and we want to know what they actually hear: what was important to them, what made a difference in their decision to participate in the study or not. How do they think the study is going to help them, or their family, or science? So we also develop our own research projects to explore and improve the process of genetic counseling.

We're learning so much about genetics right now. Do you see your field changing as a result?

Definitely. While the principles of genetic counseling remain the same, other aspects of the job are constantly changing. New conditions, new genes, and new genetic mechanisms are being discovered. Genetic technology is evolving rapidly, too, and that impacts how we go about testing. As the understanding of the genetic basis of disease is growing and genetic testing is becoming more widely available, genetic counselors are becoming involved in many other specialty areas. Cardiology clinics, for example, are increasingly integrating genetic counselors into their practices. It's exciting to see how the profession is evolving and to be part of that.

What skills or qualities does one need to be successful in your field?

In addition to having a strong understanding of biology and genetics, you need to be a good listener and be able to empathize with people. You also have to be a good teacher, one who can break down information and explain it to people wherever they are in their emotional process and educational level.

What advice do you have for students who might be interested in becoming a genetic counselor?

It's never too early to meet a genetic counselor or spend time with one in clinic. We're open to having people rotate with us to see if they like that type of environment. There's a "Find a Genetic Counselor" link on the website of the National Society for Genetic Counselors. Don't be afraid to email a counselor to express your interest. You can also get experience in just about any kind of medical or counseling setting. Volunteer at a crisis center, a suicide hotline, or other group that does counseling, and see how you feel in that role. **i**

What genetic counselors do

Genetic Counselors evaluate individual or family risk for inherited conditions such as genetic disorders and birth defects. They help people understand and adapt to the implications of genetic contributions to disease. They may conduct research.

Where they work

Genetic counselors work with patients, families, and healthcare professionals in medical centers, hospitals, private practice, and industry settings, as well as with organizations that help people with disabilities.

Education and training required

Genetic counselors must have a master's degree in genetic counseling from a program accredited by the American Board of Genetic Counseling. In addition, most employers require certification, and some states require licensure.

Salary range

According to the National Society of Genetic Counselors, the 2011 median annual salary for genetic counselors was $63,000.

For more information

American Board of Genetic Counseling
www.abgc.net

National Society for Genetic Counselors
www.nsgc.org