WRITTEN BY-MIKALEE BYERMAN

Is it in the genes?

Answers about cancer can be found through genetic testing

IT'S NOT OFTEN THAT A SPELLING MISTAKE IN AN INSTRUCTION MANUAL CAN CAUSE DANGEROUS RESULTS. But when the "instruction manual" is the genes that comprise a human being, then one spelling mistake can be catastrophic. In fact, it can cause cancer.

This is the analogy made by genetic counselors whose work involves trying to discover a patient's genetic predisposition for hereditary cancer syndromes. While only 10 percent of cancers are hereditary, people at high risk for hereditary cancers — those with a significant family history or who have cancers that have been diagnosed at an early age — can be genetically tested to determine a way to manage their health.

Brandy Smolnik, MS, a Reno-based genetic counselor, describes discovering hereditary cancer as looking for the spelling mistakes in our personal instruction manuals. "When there are mistakes, a gene doesn't work properly, and cancer can result," Smolnik said.

The mistakes, she noted, are called mutations. And while genetic testing is far from perfect, it can give a patient peace of mind — or the answers they've long wondered about.

Smolnik described 60% of cancers as "sporadic," meaning there is an age and environmental influence; 30% are familial, where several genes and the environment can play a role; and only 10% are hereditary, meaning a single gene mutation is at fault. Genetic testing only covers the cancers in this last category.

"Primarily we are looking for hereditary breast and ovarian cancer syndromes, including male breast cancer, and hereditary colon cancer syndrome which can include cancers of the colon, uterus or pancreas," she said. Genetic counselors also assist with developing a plan to maintain health and screenings appropriately.

The testing process typically starts with a patient who discusses cancer concerns with a primary care physician. That doctor may refer the patient to a practice like Smolnik's, who works with Robert Nathan Slotnick, MD, PhD, a medical geneticist at Perinatal Associates of Northern Nevada and Medical Director of Genetics at Renown Institute for Cancer.

"Brandy and I work together every day," Dr. Slotnick said. "We talk about our patients' concerns and develop a course of action. This is the best way — to have a genetic forum with a doctor and counselor working together very closely on behalf of the patient."

Before beginning the genetic testing process, a patient should spend time investigating his or her own background and history.

"The details make all the difference," Dr. Slotnick said. "Those who come to us with a lot of information about their own family — when their mom was diagnosed, at what age grandma died of breast cancer, etc. — that gives us more to work with. Many of our patients also educate themselves through their own research on the Internet or talking with family members."

Genetic testing itself involves a simple blood draw followed by the complex part: a two- to four-week wait.

"Before we test, I'll sit down with patients and discuss the family history, assess the situation and then explain the process," Smolnik said. "And regardless of the results, I'll always meet with the patient in person to offer support and explain what happens next."

Patient Mary Moffitt discusses her genetic testing results with Brandy Smolnik, MS, a Reno-based genetic counselor.

> "Reality is so much better than denial."
>
> *Mary Moffitt, after undergoing genetic testing for breast cancer*

That level of personalized service is what provides comfort to patients like Mary Moffitt. The 57-year-old Minden resident sought answers from Smolnik and Dr. Slotnick concerning her three bouts with breast cancer that started at age 32.

"My mother died at 58 from breast cancer," she said. "My sister was diagnosed at 40. So I talked to my doctor about my concerns, and he asked if I wanted to know if I carry the gene. That led me to Dr. Slotnick and Brandy."

But her initial genetic testing — the most common variety, called a BRCA 1 and 2 test — came back negative. Smolnik, not satisfied with the result, contacted other genetic counselors throughout the country seeking alternative suggestions. The outcome: a positive result in a test for a rare condition called Cowden Syndrome.

"This diagnosis gave me the answers I've been waiting for since 1983," Moffitt said. "Now that I know I have this syndrome, a lot in my life makes sense. The testing itself has been such a wonderful experience.

"Reality is so much better than denial," she said. Now we can move forward with answers instead of more questions."

After a history of multiple surgeries for cancer, Moffitt says the time she spent going through genetic counseling has been refreshing and rewarding.

"Brandy is such a great listener, and she really spent time trying to understand my history and explaining options," she said. "She left me to make my own decisions. I only wish I had done the genetic testing sooner, as I'm just relieved to have the answers." ■

R. Nathan Slotnick, MD, PhD

R. Nathan Slotnick, MD, PhD, is Medical Director of Genetics at Renown Institute for Cancer and a Perinatologist at Perinatal Associates of Northern Nevada. Dr. Slotnick has a master's degree in Molecular Genetics from the University of Washington and a doctorate degree from the State University of New York, Buffalo. He earned his medical degree from the University of Pittsburgh. Dr. Slotnick completed his residency and Fellowship training at University of California San Francisco.