



Cancer and Genetics

Lisa Graves was 50 years old when she was diagnosed with colorectal cancer last year. Even though she leads a busy life, she makes time to have an annual gynecological exam – a practice that probably saved her life. “Because of the results from a test my doctor did during my last visit, he told me I should have a colonoscopy,” Graves said. The colonoscopy found polyps that turned out to be malignant. “The surgeon had to remove a part of my rectum, but because they caught the cancer early, I didn’t need any other treatment. People should know that early detection really is a lifesaver,” she said.

Graves recalls her surgeon talking to her after her surgery about genetic counseling, a service of the Cancer Program of Our Lady of the Lake and Mary Bird Perkins. Genetic counseling for colon and rectal cancer is part of the Cancer Program’s multidisciplinary colorectal cancer approach. When initially discussing the consideration of having genetic counseling, “I was too distracted to pay attention,” Graves admitted. But when approached about it later, Graves said she was more prepared to listen. “The counseling was very educational. When I found out there are colon cancer

genes, I wanted to be tested. I was concerned that if I was a carrier, I could have passed the gene on to my 34-year-old son.”

This is where geneticist Duane W. Superneau, M.D., Our Lady of the Lake Genetic Services, comes in. Dr. Superneau offers genetic counseling for individuals or families at risk for various hereditary cancers. During genetic counseling, Superneau and his staff simplify complex scientific concepts to provide clear and clinically relevant information about genetic risk factors. This information is a key component in the early detection of cancers in high-risk patients and in the decision of those considering whether or not to have genetic testing.

Superneau explained that all cancer has a genetic basis, but not all cancer is hereditary. In fact, he said, only a small portion of all cancers – maybe 5 or 10 percent – can be

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attributed to heredity. Colon and rectal cancer inherited through a parent's genes, which can be documented by genetic testing, falls into the 5 percent group. The other 95 percent of colon and rectal cancers are due to local changes in the colon tissue.

When asked if he's noticed an increased interest in the role genetics plays in cancer, Superneau replied, "We're seeing more people because they're more aware of the subject, but we're not necessarily seeing the right people." He explained that not enough people are getting screened who should be screened due to a family history. "The majority of people we see already have cancer. Sometimes we see individuals concerned about their family history, but they might not be the candidate who should be tested." He said that he is a resource for genetic counseling whose role is to determine which family member is the appropriate one to be tested, if the family decides to take it that far. "The big take-away from all of this," Superneau emphasized, "is to know your family history and obtain appropriate screening studies for cancer when indicated."

Graves said she knew colorectal cancer could be hereditary, and it scared her. Her children would benefit from Graves knowing if she had the gene because they could be more diligent about early detection practices. "Moms are universally concerned about their kids," shared Dr. Superneau. "Through genetic counseling they learn that if they have the gene, they got it from their parents, and in similar fashion, it could be in their children." He explained, however, that medically his first priority is to the patient. "If Lisa has the colon cancer gene mutation, that means all the cells in her body have the mutation. Colon cancer gene mutations also cause uterine and ovarian cancers, putting her at a greater risk for both." Superneau said there would be two options to lower the odds of her contracting either cancer – removal of the uterus and ovaries or more frequent imaging of the same. "Genetic counseling then becomes a medical education about your own health concerns," Superneau said. "Lisa would, of course, have to have more frequent colonoscopies, for instance, which insurance then covers. But, we would also have more information about her chances for a recurrence."

Thankfully, Graves' test proved to be negative. "Now I feel relief," she said. "If it would have been positive, I would have been in a worried state for myself and my son the rest of my life." She added, however, that she would have been better prepared to take the necessary steps for her health care. She believes the genetic counseling resource being offered as an option for cancer patients is a good thing. "I recommend it. I didn't know anything, and I found it very helpful. Being that my test was negative, it was also very reassuring."

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Lisa Graves

Genetic Counseling Session Preparation

The best way to prepare for a genetic counseling session for adult cancer is to find out as much as you can about your family medical history. Talk to your family members and try to find out medical information about your siblings, parents, aunts and uncles, cousins, grandparents, children and grandchildren. At minimum, this information should include:

- Your relation to each family member, including whether family members are adopted or half-relatives
- Major health conditions that affect each family member, such as cancer, diabetes or heart disease
- The age of onset for each condition
- Age and cause of death (where relevant)

Try to confirm each health condition that affects family members. In many cases, your risk may be different depending on exactly what condition your family member had. For example, if you think that a relative had lung cancer when in fact they had breast cancer, it could seriously affect the accuracy of your risk assessment.

that early detection

- Lisa Graves