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Is it hereditary?

Genetic counseling about cancer can inform and guide future decisions

By Kimberly Ballard

In 1999, Leslie Valley was diagnosed with lobular breast cancer. Fully aware of the hereditary nature of her ailment, her immediate fear was for her three daughters and two nieces. Could she have passed on the dreaded breast cancer gene? Under the care of board certified gynecologic oncologist Dr. F. Joseph Kelly at Huntsville's Clearview Cancer Institute, Valley underwent genetic counseling and testing at CCI to find out whether she carried either the BRCA1 or BRCA2 genes.

The presence of the BRCA markers does not make cancer a certainty, but increases the propensity for hereditary cancers. The existence of the BRCA1 gene is related to breast and ovarian cancer, as well as uterine, pancreatic, and colon cancer. The presence of a harmful mutation of the BRCA2 gene heightens the risk of pancreatic, stomach and gallbladder cancers, as well as melanoma.

As the instruction books that allow our bodies to function, genes mutate in matched pairs, but due to the complexity of the process mismatched genes will sometimes pair off. "The beauty of the human genome is that the body has a built-in mechanism to correct or repair mismatched genes when they are working properly," Mason says. "But when that mechanism ceases to make the correction, the mutation of mismatched genes becomes dominant." This harmful mutation causes cancer. "We all have good genes, but some good genes just go bad. Why that happens is still the long term goal of cancer research."

A HUGE ROLE

Genetic counseling for certain types of prevalent genes, followed by genetic testing or screening, has played a huge role in helping patients or potential patients take preventative measures against different forms of cancer. Patients are also advised on the best options for their families.

"We can start with a simple telephone interview to gather as much information about your family medical history as possible," says Carla Mason, a licensed and certified genetic counselor specializing in hereditary cancers. "This preliminary information leads us to the need for a deeper investigation." For the average family, going back farther than the grandparents can be problematic. Most people have no idea what illness their great-grandparents suffered. Prior to the mid-20th century, the fact someone died of a stomach ailment or pains in the chest does not tell us what unknown and undiagnosed causes they suffered. "If you believe that your grandmother and perhaps a great



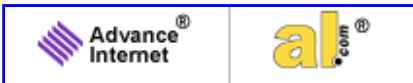
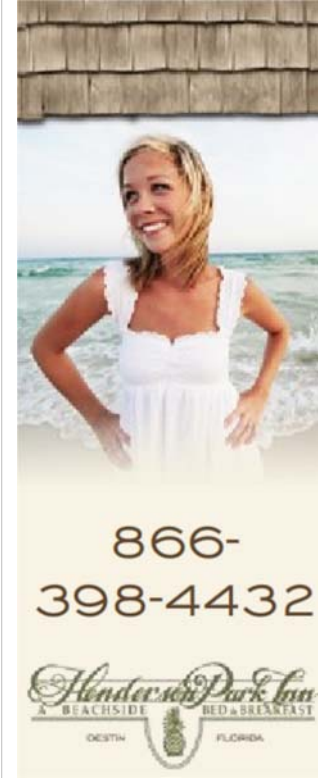
Eric Schultz

Genetic counselor Carla Mason, left, and Leslie Valley look over some notes at the Clearview Cancer Institute.

aunt died of cancer, we will want you to try and find out as much as you can about that," Mason says. "It may require doing some homework."

The next step is scheduling a consultation. "It takes about an hour, and we request you bring your homework - any more detailed information you uncovered, to that meeting," Mason says. "With your family medical history, we start putting together a puzzle."

"Both of my parents were only children, so the lack of family medical history was a problematic piece of the puzzle," Valley says. "There was no history of breast cancer in my family as far back as I could trace, but there was also not enough family history to paint a very clear picture."



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Valley opted for genetic testing. "In this consultation, we educate the patient," says Mason. "We do not push them to make any decisions, but if there is a level of risk due to genetic markers, we make sure they understand their options."
 The first option involves non-invasive testing. With breast cancer, a blood test will show whether the patient has the genetic markers. Ultrasounds can detect some forms of cancers, and there is formidable testing for colon and prostate. Cervical cancer, which is not hereditary but caused by sexually transmitted diseases, is easily preventable with annual pap smears. There are no reliable pre-screening techniques for many forms of cancer like brain and lung cancer, endometrial, ovarian, and uterine cancer. In some cases, by the time the symptoms appear, the cancer has already spread, as in the case of liver cancer.
 "Being at risk does not mean you will get cancer, but we do offer a secondary preventative option for patients with a familial cancer history. Oral chemotherapy has been shown to lower the risks of some cancers by

up to 50 percent," Mason explains. "We have also had very high success with the birth control pill, which has been known to reduce ovarian cancer by up to 60 percent." Going onto medications, particularly oral chemotherapy, for purely preventative reasons is a big decision for a patient to make, according to Mason. "Again, we educate the patient on their options, and the final decisions are completely up to them."
 Preventative measures are just one of the genetic counselor's tools. "Patients who are in the high-risk category have some more extreme options. For women with multiple generations of breast cancer, a radical mastectomy to remove the breasts is an alternative." This surgical procedure includes the removal of the chest muscle, lymph nodes and underarm pit.
 "In families where two or more generations of women have suffered from breast cancer, this far-reaching solution is not unheard of, but even a full mastectomy is not a guarantee," Mason says. "The breast wraps around to the back, under the arm,

and deep into the chest. If potential cancer cells are left behind, women are still at risk."
 The same is true of an oophorectomy in which both ovaries are removed. "The success rate is high, about 90 to 96 percent," Mason says, "but the ovarian cells also line the abdomen and leaving those behind can be a long-term concern."
 Leslie Valley's story has a successful conclusion. Her mastectomy and 6 months of chemotherapy and radiation treatments have given her a clean bill of health. Furthermore, through genetic counseling and testing she, her daughters and her nieces have some peace of mind.
 "A blood test showed I did not have the BRCA gene. It was a relief to know I had not passed it along to my daughters, but that does not mean we are not diligent to keep a watch on it," Valley says. "Screening is so important! I cannot stress enough the importance of annual check-ups like mammograms and pap smears – and for men, tests for prostate cancer and colonoscopies. Do not delay!" ■

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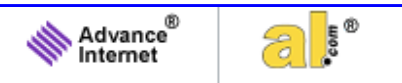
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