

Detecting cancer early

How genetic testing can help

Four years ago, at age 39, Bill was told he had colon cancer. It's not the kind of information anyone wants to receive, but after rounds of surgery and chemotherapy there was some good news: Bill's cancer had been eradicated.

However, Bill has a family

history of cancer, including his deceased mother, who had colon, uterine and kidney cancers. So he wasn't prepared to simply wait and see whether he might get another cancer.

Toward the end of his chemotherapy treatment, Bill learned that

he could have a genetic risk assessment to find out more about his future health—and the risk of disease for people related to him.

Empowering the family

After talking with a genetic counselor at the Cancer Genetics Risk Assessment Clinic at St. Vincent Indianapolis Hospital, Bill opted to undergo genetic testing, which involved a simple blood draw.

A month later, Bill learned that he's a carrier for a gene mutation called MSH2, one of the genes associated with Lynch syndrome, which predisposes people to colon cancer, as well as other cancers. His two children, now ages 5 and 18, each have a 50 percent chance of carrying the mutation. Bill's sister chose to have genetic testing and discovered she, too, carries the familial mutation.

"My children and sisters were my driving motivation," said Bill. "It wasn't so much about me anymore, because I've already had cancer. Although I didn't know for sure if my cancer was hereditary, I wanted to know if there was a strong potential for me to have it again or if other family members were at risk."

The Cancer Genetics Risk



Genetic questions?

If you feel you may be at risk for cancer, contact the Cancer Genetics Risk Assessment Clinic at **(317) 338-RISK (7475)**. More information is available at cancergenetics.stvincent.org.

➔ Screening guidelines

Even if there's no family history of colon cancer, the American Cancer

Society recommends that starting at age 50, men and women should use one of the screening tests below. The tests designed to find both early cancer and polyps are preferred. Talk to your doctor about which test is best for you.

TESTS THAT FIND POLYPS AND CANCER:

- a flexible sigmoidoscopy (a thin tube with a camera attached that checks the lower part of the colon) every five years *
- a colonoscopy (a thin tube with a camera that checks the entire colon) every 10 years
- a double-contrast barium enema every five years *
- CT colonography (virtual colonoscopy) every five years *

TESTS THAT MAINLY FIND CANCER:

- yearly fecal occult blood test (FOBT) *
- yearly fecal immunochemical test (FIT) *
- stool DNA test (sDNA) *

*Colonoscopy should be done if test results are positive.



Assessment Clinic aims to identify people at risk for hereditary cancer, provide risk assessment, discuss the benefits and limitations of available genetic testing and advise them about current screening recommendations based on their family history and genetic test results. Ultimately, the goal is to reduce deaths from cancer. “Our goal is to give people a sense of empowerment, which can change the way they approach their medical care,” said Stephanie Cohen, a board-certified genetic counselor. “It also gives family members the impetus to get screened early.”

Genetic counselors help

Knowing there's a hereditary link to cancer can be a relief for patients, Cohen said, because it

means their lifestyle wasn't to blame for their cancer diagnosis. Others, who may be distressed by the testing results, can look to Cohen and her colleague, board-certified genetic counselor Dawn McIlvried, for support. They talk to patients about how to share the results and what it might mean to their relationships with family members.

Bill said that knowing he has a genetic mutation has put him in charge of his health. “There's no such thing as too much information. I wanted to know so I could deal with it on my own terms. Then I could feel like I was controlling the cancer and it wasn't controlling me.”

Not every person with a mutation will get cancer, said Cohen. “But their risk is much higher. Those

who carry a gene predisposing them to Lynch syndrome, for example, have up to an 80 percent lifetime risk of developing colon cancer.”

Stepping up the screening

Bill's concern now is his children. Because they have a 50 percent chance of carrying the gene, they both need to be screened for cancer beginning at age 25, according to guidelines for people who have Lynch syndrome or are at high risk for it.

Because they both know exactly which gene mutation they may be carrying, testing will be much faster and less expensive because doctors will be looking specifically for this mutation, rather than putting them through a battery of tests.

To further help, Bill is keeping track of medical records. The testing he underwent was aided by the fact that his mother had carefully mapped the family's medical history before she died. “It's very helpful for family members to keep track of records pertaining to cancer diagnoses, so that information is available for more accurate risk assessment for hereditary cancer,” said McIlvried.

For Bill's sister, who doesn't have cancer, knowing about the gene mutation will help her if she should get abnormal test results in the future, she said, because doctors don't always consider colon cancer in patients under age 50. She will now be armed with her genetic testing results to push for more extensive screening. ■