



THE INS AND OUTS OF GENETIC TESTING

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From the time of Hippocrates (400 BC) all the way through to the nineteenth century, prevailing scientific thought held that the workings of the human body were dictated by the four “humors,” or bodily fluids: black bile, yellow bile, phlegm and blood. Ridiculous, sure, but also comforting; all medical problems were thought to constitute either a deficiency or a surplus of fluids, meaning any illness could theoretically be cured by correcting the imbalance.

Today we know that genetics are actually responsible for much of the way our bodies behave. Genetics dictate everything from our hair color to our shoe size; they can also dictate our predispositions for various types of disease, including cancer. And, unlike Hippocrates’ four humors, our genes are outside of our control (so far).

Not all forms of cancer have a hereditary component, but some of the most infamous do: breast cancer, ovarian cancer, colon cancer, prostate cancer. If you have multiple relatives with the same type of cancer, relatives who were diagnosed with cancer at young ages, or an unusual number of rare cancers among your family members, there’s a chance that a cancer-causing genetic mutation runs in your family. The only way to know for sure is a genetic test.

According to Kim Ranieri, certified genetic counselor and program coordinator of Genetics at the Cancer Institute of New Jersey, there are now around a dozen clinical tests available. “The most common types are genetic testing for breast, ovarian and colon cancer,” she said. “There are lots of other very rare cancer symptoms we can test for, such as hereditary melanoma and hereditary pancreatic cancer, but those are the ones we see most often.”

Genetic testing consists of examining a sample of a patient’s blood for an anomaly in their DNA that indicates a mutation is present. Predictive gene tests already exist for a number of cancer-causing mutations. Mutations on the BRCA1 and BRCA2 genes indicate an increased susceptibility for breast and ovarian cancer in women and prostate cancer in men; familial adenomatous polyposis gene mutations practically guarantee the development of colon cancer in both men and women.

“Generally speaking, there are two reasons people come in for genetic testing,” Ranieri said. “One, they decide there’s something they might do differently if it turns out they’re at a higher risk. Or two, they’re worried about their family members. A woman will come in because she’s had a breast cancer diagnosis and she’s worried about her daughter or her sister.”

Both are compelling motivations. Before undergoing genetic testing, however, it’s important to weigh the advantages and disadvantages.

First of all, Ranieri stresses that genetic testing is not for everyone. “Less than 10% of cancers are caused by an inherited predisposition,” she said. “A lot of people have this misconception that all cancer is hereditary, which is almost the opposite of the truth. And one of the biggest limitations of testing is that we don’t know every gene under the sun. A negative test result doesn’t get anyone off the hook.”

The next thing to think about is how you’ll handle the information should your results turn out to be positive. “We encourage people to think about whether this is information that will cause them a lot of psychological distress,” said Ranieri. “Or do they view it as empowerment? This isn’t going to be beneficial for everybody. There are a lot of things to think about. Two people with the exact same medical history can make very different decisions about getting tested or very different decisions about what to do with the results.”

For many people, Ranieri says, genetic testing provides serious relief from anxiety about cancer risk -- even if the results are positive. “There’s one response, which is, ‘Here I am, feeling like this has been hanging over my head, and now I feel relieved,’” she said. “Or the reverse, which is, ‘My test is positive, but at least I know.’”

And positive test results give patients the ability to take preventative steps that aren’t recommended for people without an inherited susceptibility. “If we learn that someone’s at an increased risk for cancer, we can change their care,” said Ranieri. “There’s increased surveillance, like mammograms in women who aren’t really old enough to get a mammogram; then there’s chemo prevention, which is taking a medication like Tamoxifen to lower the risk; and the third thing people can do is preventative surgery.”

Additionally, there can be unexpected fringe benefits to learning you have a genetic predisposition to cancer. “We work very closely with a clinical psychologist and she’s been doing a study about the behavior of kids whose parents test positive,” Ranieri said. “It causes them to change their lifestyle behaviors. The two most common changes are they quit smoking and they start exercising.”

But there can also be a downside to sharing your results with your family -- after all, if you have a genetic mutation, your blood relatives may have it too. “Just because one family member wants genetic testing doesn’t mean other members of the family want that information,” Ranieri points out.

Discrimination is also increasingly a concern -- so much so that in May, President Bush signed the Genetic Information Nondiscrimination Act (GINA), which prohibits health insurers and employers from discriminating against people based on the results of genetic tests. GINA says nothing, however, about life insurance, and "if someone has a genetic predisposition, a life insurance company may be able to ask for and use that information," said Ranieri.

The good news is that Ranieri and her colleagues are trained to help patients make these difficult decisions. "I would encourage anyone who's concerned to get genetic counseling," she said. "Get all of the information. Don't make any assumptions. There are trained professionals who are happy to sit down and go over unbiased information, then let people make their own decision about what's best for them."

For one woman's real-life account of genetic testing and its aftermath, check out Michelle McBride's [BRCA Journal: The Journey of a New Mom](#)

For more on genetic testing and civil liberties, visit [the ACLU Blog](#)

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