

Genetic testing can reveal if you have one of the 3,000 genes linked to diseases. But the bigger question is would you want to know?

OR NOT TO TEST

Based on her family history, Beverley Docherty believed it was only a matter of time before she developed breast cancer.

“My grandmother on my mother’s side died of breast cancer in her early 50s, and my aunt on my mother’s side got breast cancer at 33. My cousin, my aunt’s daughter, found a lump last year and had to have a double mastectomy, her ovaries removed, and was put into menopause at 31.”

Docherty, also 31, decided to confront her fate after receiving genetic counselling at Alberta Children’s Hospital in Calgary. In April 2010, she decided to have her DNA tested. “My mother tested positive for the gene but never had breast cancer,” explains Docherty, “but with the experiences of the other women in my family, I suspected I could be facing breast cancer in my lifetime.”

Until recently, only time could tell if a person would develop breast cancer.

Since two telltale genes were identified in the mid-1990s, individuals with a family history of breast cancer, which one in 5,000 Canadian women will develop, can find out if they were born with one of the defective genes that cause the disease.

The decision to be tested for a genetic disorder, such as hereditary cancer or Huntington’s disease, is not to be taken lightly, says Deepti Babu, a genetic counsellor at the University of Alberta in Edmonton. “Predictive genetic testing is much

more than a blood draw. There are implications that can be far-reaching, and these can have a ripple effect on family members.”

While the decision to test is yours alone, the results can affect your entire family. “Genetic testing does not just reveal information about an individual. It reveals information about a whole family,” says Dr. Victoria Seavilleklein, who works at Red Deer Regional Hospital Centre as a clinical ethicist for Central Zone of Alberta Health Services. “If an individual were found to be a carrier, that would tell you that at least one of the parents was also either a carrier or actually had the condition, and would have implications for the parents and siblings.”

In guiding families and health-care teams on issues surrounding medical interventions, including genetic testing, individuals, families and health-care providers are faced with considering the benefits of telling someone their risk for a certain disease they may never get or be able to do anything to prevent.

In fact, a positive genetic test for a disease doesn’t definitely mean you’ll get it. It means you are at a higher risk than the average person. Likewise, this kind of test can’t predict when you could get a disease or how severely it may affect you.

But just knowing you are more susceptible to a disease can significantly alter your health. Seavilleklein says a new disease category, called

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What is genetic testing?

Genetic testing is used to determine if your genes have variances that might cause a disease.

The genes that determine everything from the colour of your eyes to the curl in your hair are found in the 23 chromosome pairs in your cells. You inherit half your chromosomes from your mother and half from your father. Genes are made up of DNA and sometimes contain small variations, called mutations, which may or may not lead to a health problem. Everyone's genes carry some normal variances.

In genetic testing, a blood sample is taken and the DNA sequence studied for a specific gene mutation to diagnose a health concern. In predictive genetic testing, an individual who has no signs of a genetic condition is tested for a particular gene change. Test results usually take several weeks or months, but may be available sooner if the situation is urgent or a mutation has been found previously in a family member. Most predictive test results show probabilities of increased risk—even a positive result does not guarantee a person will become sick. Some tests can be difficult to interpret and none are 100 per cent determinative. Genetic counselling is an important part of helping people understand the complexities of what testing can mean and how to deal with the results.

pre-symptomatically ill, describes people who have tested positive and are not sick, but start to perceive themselves as ill or becoming ill down the road.

And testing negative can still be emotional. A person who screens negative, while another family member is diagnosed with a disease, often experiences survivor's guilt. Instead of feeling relief, the one spared feels undue remorse, or even responsibility.

Tara Johnson-Ouellette was sure she wouldn't feel guilty if she tested negative for Huntington disease, an inherited brain disorder that affects one in every 10,000 Canadians. In 1994, when she was 20, her mother was diagnosed—a year after researchers had isolated the mutant gene. Johnson-Ouellette had a 50/50 chance of inheriting the gene.

"All of a sudden this bomb is put on your lap," recalls Johnson-Ouellette, whose family became the first in Calgary to receive the new protocol regarding genetic counselling for Huntington's disease. "Your mom is dying, slowly, there is no cure, and by the way, you're at risk. I didn't think it was just a test, I thought I had it. For life."

Johnson-Ouellette was prepared for the worst when the results came back nine months later. "They opened the envelope and handed it to me, saying: 'You don't carry the gene.' I went into shock. It was not what I expected." But the immediate relief she felt vanished on the short ride home from the hospital, and guilt assumed its place when she told her dying mother the good news.

She says her brother, who was too young to be tested in 1994, has never wanted to know if he carries the gene. While carrier screening may be offered to other family members after a diagnosis, Seavilleklein stresses it's very important for siblings to decide for themselves if they want to be tested, and to get separate counselling.

Although the decision to test for a genetic disease is yours alone, Babu emphasizes, you

don't have to make it alone. Before a hereditary cancer genetic test is arranged, an adult without symptoms is referred to a genetics clinic for genetic counselling to help understand the risks, benefits and limitations of testing. In Alberta, only a genetic counsellor or clinical geneticist can order a hereditary cancer genetic test.

Dr. Stacey Hume is a molecular geneticist and director of the Genetic Laboratory in Edmonton, which tests for about 40 different genetic disorders. For adults, the most commonly ordered tests are cystic fibrosis, hemochromatosis (iron overload), Huntington's disease, amyotrophic lateral sclerosis (ALS, or Lou Gehrig's Disease), deafness and colon cancer.

Hume says a person is susceptible to developing hereditary breast or ovarian cancer when they inherit a mutation in either their BRCA1 or BRCA2 gene. The BRCA genes are the ones responsible for controlling cell growth in the body. "If you are born with one mutated gene, you have a 50 to 70 per cent chance of acquiring the second mutation and developing cancer by age 70." Some, like Docherty's cousin, may acquire the second mutation early, while others, like Docherty's mother, may never acquire it.

Through genetic counselling, Babu helps individuals do what is important to them. That includes ensuring the motivation to test is from the individual and not someone else. Pressure from concerned family members is common with predictive testing, she says. "I'm not comfortable until I've helped someone make a decision to test—or not test—that is consistent with their own personal life values and priorities. These are not easy decisions for anyone."

Making the decision involves understanding options and implications. Docherty knew if she tested positive for breast cancer, she could choose increased cancer screening or preventive surgery.

After learning she had the BRCA2 gene mutation, she had a double mastectomy and reconstructive surgery in October 2010.

"I wanted to get my breasts before they got me," says the Calgary mother of two-year-old Greyson. "I wanted to do (the surgery) on my own terms, when I'm healthy, rather than live under that cloud of being screened every six months and waiting for the test results."

Having control over her reproductive health was another strong motivator; Docherty didn't want to face the same fate as her cousin: "I wanted to be free to choose whether to have more children or take hormones when I go into menopause naturally."

Now 37 and the new mom of son Carter, Johnson-Ouellette, has been channelling her survivor's guilt into helping others who are considering testing. For 17 years, she has been sharing her experiences through the Southern Alberta Chapter of the Huntington Society of Canada, encouraging families to seek professional support before and after a test. "People who test negative think 'I'm good, problem's over,' but that's a false reality. They need to understand how it will change their lives and how to deal with it in a positive and constructive way."

One of the disadvantages families face with an inherited condition is possible discrimination based on genetic information. Johnson-Ouellette was only able to get life insurance after testing negative for Parkinson's. She learned individuals who acknowledge a family history of Huntington's disease may be turned down by insurance companies as a health risk—even if they have no symptoms of the disease and have never been tested. Today, she is helping voice the need for Canadian laws to prevent insurance companies and employers from using predictive genetic testing to decide who is insured or employed. (Americans have been protected from the misuse of genetic information in health insurance and employment since 2009.)

Not everyone wants to undergo genetic testing. For some, the fear of knowing is greater than the fear of not knowing. For others, being tested is the only way to take control of their destiny. Knowing what her future could have been gave Johnson-Ouellette's life new purpose. Docherty, who was back in the gym with her doctor's okay 10 days after surgery, felt privileged to stare down fate with genetic information her grandmother didn't have. *a*

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