

Genetic Testing: Promise and Peril

By J. Riediger-Duebel

On Sept 23, 2004 the Provincial Museum of Alberta, in partnership with Genome Prairie, hosted a public forum on genetic testing in Edmonton. I was invited to be on the panel at the forum. Each panelist addressed the audience briefly on the subject of genetic testing. My presentation is reprinted below.



In high school I learned about the DNA double helix and Mendel's yellow and green peas. I never imagined the impact genetics would have on my life. In the last ten years I've come to know personally about some of the Promise and Peril of genetic testing.

My talk today is specifically about genetic testing for hereditary breast and ovarian cancer. Some of the points I raise may not apply to genetic testing for other diseases or conditions. I will focus on the impact of genetic testing on women. Also, I have limited my remarks to the impact of positive or negative genetic test results. Unfortunately, some test results are inconclusive or uninformative at this time.

THE FACTS

Five to 10% of breast and ovarian cancers are hereditary. Two genes were discovered in the mid 90's. They were named BRCA1 and BRCA2. Women with a mutation in one of these genes have a lifetime risk of 50 to 85% of breast cancer, and many will develop breast cancer before the age of 50. These women also have a 20 to 50% lifetime risk of ovarian cancer. If a parent carries a mutation, the children will have 50/50 odds of inheriting it as well.

MY STORY

In 1995 my sister lost her battle with ovarian cancer. My mother had died of breast cancer at the age of 49. In total there were almost a dozen cases of cancer on my mother's side of the family.

A year after my sister died, two of my cousins who were living with cancer agreed to have genetic testing. Our family was found to carry a BRCA2 mutation. For the first time, all the cancer in our family made sense. It wasn't because we hadn't eaten our veggies. It wasn't the "sins of our fathers". It was one tiny mistake on one gene. What havoc this mistake had wreaked in our family. I tested next and found that I also carried the mutation.

Through the Hereditary Breast & Ovarian Cancer Society, I've been fortunate to meet many families in Alberta who have gone through, or are considering genetic testing. There are ethical, legal, economic and political issues to consider, but today I'd like to look at the personal consequences of genetic testing.

THE PROMISE OF GENETIC TESTING

Why do people decide to be tested?

1) A GENETIC TEST RESULT PROVIDES INFORMATION THAT IS THE BASIS FOR INFORMED DECISION-MAKING

This is possibly the most important promise of genetic testing for those who test positive for a gene mutation.

Why is the information provided by genetic testing so critical?

Let me give you an example.

Some years ago I got to know a woman who was five when her mother died of breast cancer at age 44. In her family there were 40 known deaths from breast or ovarian cancer. It was 1981. The BRCA genes had not been discovered. Genetic testing was not an option. She was a nurse and had seen cancer up close. She was determined not to be the next in line. At 34 she made the decision to have surgery to remove her breasts. Some years later she had her ovaries removed. By then, two of her four sisters had been diagnosed with breast cancer. When genetic testing became available years later, her family was found to carry a BRCA1 mutation. When she tested, her result was not what she expected. She did NOT carry the mutated gene.

I said earlier that genetic testing can be a basis for informed decision-making:

A woman who tests **positive** for the BRCA1 or 2 gene mutations has several choices.

i) First of all, she can increase surveillance.

Recommendations for screening of high risk women differ significantly from those of the general population. Screening begins earlier and includes more tests than are generally recommended.

ii) Secondly, she can choose prophylactic surgery to remove her ovaries and/or her breasts. Prophylactic surgery may seem like a radical choice to some, but it remains the option that reduces risk most significantly.

iii) Thirdly, she can enroll in clinical trials. There are on-going studies that are looking at agents that may reduce cancer risk, imaging techniques that might find cancers earlier and treatments that may work better for cancers that are hereditary.

The PROMISE of genetic testing for those who test positive is that they can be proactive. They can make informed choices based on their individual genetic test result.

2) GENETIC TESTING GIVES THOSE WHO TEST NEGATIVE A NEW LEASE ON LIFE

If I would pick the best thing that genetic testing has done for families with a BRCA gene mutation, it would be this. In a BRCA family, the odds of not inheriting a mutation are 50/50, the same as the odds of getting it. The people who get the negative result win the lottery!

They can live their lives without the fear of cancer. Imagine thinking your whole life that you wouldn't live past 40 or 50, and suddenly finding out you might live twice that long. People have told me that they had to drastically alter their thinking when they found out they tested negative. For the first time they began to seriously plan for retirement. For the first time, they had hope that they might live to see their grandchildren.

For those who test negative, there is no longer the need for high risk cancer surveillance. They can go back to the screening protocols recommended to the general population. Invariably this results in reduced stress and fewer medical appointments.

For women who test **negative**, there is no need to agonize over the decisions of surgery versus surveillance.

Both women and men who test negative have the immense relief of knowing that their children will not inherit the gene mutation.

SO, WHETHER POSITIVE OR NEGATIVE, A GENETIC TEST RESULT CAN GIVE HOPE TO FAMILIES WITH HEREDITARY CANCER.

Those who do carry the mutation can be proactive in managing their risks. Knowing that they are at high risk empowers them to take action. They no longer feel that dying from cancer is their inevitable fate.

Those who don't have the mutation can carry on their lives without the fear of cancer. They can reasonably hope for a long and healthy life.

THE PERIL OF GENETIC TESTING

There may be Perils for individuals, families and society. Let me name a few.

For individuals there may be:

Fear of insurance or employment discrimination. Even though there is no compelling evidence of widespread discrimination, the fears remain. Will life insurance be denied if I test positive? Will an employer prefer to hire someone who is less likely to need time off work for illness? If I move to another country will I be able to obtain health care insurance?

Fear of how others will respond. Who should I tell? Will they understand why I chose to have genetic testing? Will they support my decisions if I test positive?

Feelings of guilt. How will I feel if I test negative and my sister is positive? Will I feel guilty if I pass on the mutation to my daughter?

For families there may be perils.

There is potential for divisiveness in families. Who is responsible to tell family members about an identified cancer gene? What if someone doesn't want that information? Can I keep my genetic test result a secret if it identifies a gene mutation in the family? Or am I obligated to tell my relatives? Where do I find support if I decide to have genetic testing but others in my family want nothing to do with it?

There may be perils for society.

Genetic testing may create more demand on health care resources. Some would argue that this peril will be offset by the reduced demand by people who test negative and no longer require increased services.

Genetic testing will necessitate a reconfiguration of the provision of health care. This might better be described as a challenge rather than a peril. Families with hereditary cancer are a small demographic. They face issues that fall both within and outside the mandate of current cancer services. Most cancer programs are geared to deal with people with disease. Genetic testing has given us the ability to predict which individuals in a family will most likely get cancer. What services should be available for those individuals? Who should provide them? Where should those individuals go for advice, services, support and information?

IN CONCLUSION

When I think about the Promise and Peril of genetic testing, I have to come down on the Promise side.

There are many challenges. But with vision and leadership, open discussion and the continuing advances in research, those challenges can be overcome. Information and education will help to alleviate the many fears surrounding genetic testing.

More importantly, there is Promise. Promise for individuals and families to break the curse of cancer in their lives. There is Promise for future generations. Promise that they will have more and better options to live a long and fulfilling life. ☀