Expert endorses genetic testing for BRCA1 and BRCA2

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Professor Kelly Metcalfe, of U of T's Lawrence S. Bloomberg Faculty of Nursing, is leading the charge against hereditary breast and ovarian cancers by helping establish the standard protocol for addressing cancers associated with BRCA gene mutations.

Women with these mutations are at far higher risk for breast cancer, and if they do develop cancer, it appears at an earlier age and is more aggressive than sporadic forms of the disease. Metcalfe has shown that surgical interventions for women with BRCA mutations can reduce the risk of developing breast cancer by 90 per cent and increase cancer survival by 50 per cent.

"Genetic testing for BRCA1 and BRCA2 can save lives," says Metcalfe. "I want women to have this genetic information so they are in a position to prevent a cancer before it happens or receive personalized cancer treatment to reduce their risk of dying."

Metcalfe holds the Bloomberg Professorship in Cancer Genetics at the Lawrence S. Bloomberg Faculty of Nursing, which is ranked No. 6 in the world. She spoke with U of T News about her research.

What is your area of research?

It's mainly focused on the two main breast cancer genes – BRCA1 and BRCA2.

How do we identify people who are at higher risk of developing breast cancer, [and] once we identity them, how do we prevent breast cancer from ever happening. How do we target their treatment so they have the highest chance of surviving the disease?

I'm very committed to ensuring women have the most up-to-date information so they can make a decision that's going to be the best for them.

What's the importance of knowing if you have the breast cancer genes – BRCA1 and BRCA2?

When it comes to breast cancer diagnoses, most women don't know if they have a BRCA1 or BRCA2 mutation. This is valuable information to have to help women best evaluate their surgical options and improve their survival rates.

With the BRCA1 or BRCA2 mutation, the most aggressive type of breast cancer surgery – a bilateral mastectomy (removal of both breasts) – is often the best approach as it reduces the risk of dying by about half. This is different than in women without a BRCA1 or BRCA2 mutation, where there is no survival advantage associated with a bilateral mastectomy compared to breast conserving surgery.

Genetic test results typically take a long time to come back – up to months – so even if a woman was tested at the time of her breast cancer diagnosis, she wouldn't know the results before she had to decide on the surgery. I wanted to change this because with current genetic technology we can offer these tests much faster.

As part of a study, we're [doing genetic] testing on 1,000 women at the time of breast cancer diagnosis. We give them their genetic test results

back within 10 business days, prior to them having their surgery, and then we're evaluating whether they change their surgical decisions based on their results.

To date, we've tested just over 700 women. The preliminary results show that women use their genetic information to make surgical decisions. The uptake of bilateral mastectomy is over 90 per cent for those women who have the BRCA mutation.

Does having genetic tests done at the time of diagnosis cause even more stress for the patient?

I was concerned that may be the case so we are evaluating that as part of the study.

To date, the results have suggested that those identified with a BRCA mutation are no different in any type of psychological measure – anxiety, depression, cancer-related distress. It isn't adding an increased burden to what they are already experiencing as a result of their breast cancer diagnosis.

Women want this kind of information. When the study comes to an end, we will try to ensure that we can integrate this type of service into clinical care so that women can continue to benefit from fast genetic test results.

How is genetic testing for breast cancer genes currently being done?

In Ontario – and it's very similar across all provinces in Canada – there are very strict criteria as to who can have genetic testing for the two main breast cancer genes – BRCA1 and BRCA2. This is mainly based

on personal and family history of cancer.

Do I think they're perfect? No. Are we missing people who have these genetic mutations based on this criteria? Absolutely.

For example, we did a large study a few years back with the Jewish population. We know BRCA mutations are more frequent in this population. We offered free genetic testing to 6,000 Jewish women [in Ontario], and we were able to identify women with the BRCA mutation before cancer happened.

We also found the majority of the women we identified with the BRCA mutation would not have met [standard] criteria for testing in the province. Based on that, we know we're missing people who have these genetic mutations.

We want to change this and are currently offering a study where we provide genetic testing for BRCA1 and BRCA2 to any Canadian over the age of 25 for a very low cost. It is important to evaluate providing population based genetic testing, even to people without a family history of cancer. We think that every Canadian should have access to this test if they want the information. Anyone can sign up for the study at thescreenproject.ca.

What's next for you in your research?

In the last 20 years, my research has contributed to what we know about the clinical implications of having a mutation in BRCA1 or BRCA2.

However, there are newer cancer causing genes that we know less about. One example is PALB2. Women are having genetic testing for this gene in addition to others, and we really don't know what to tell them if they get a positive result. I want to change that. I want to be able to tell a

woman with a PALB2 mutation what she should do to maximize her chance of not getting or dying of breast cancer.

What kind of feedback have you received about your research?

A big part of what I do is translate our research findings for the research, medical and patient communities.

[These patients] are very engaged in research because this is a genetic mutation that gets passed down from generation to generation. Many of the women that we see also have a very significant family history. They've seen many of the women in their family develop and die of cancer.

I try to ensure the research is translated to the patient so they know the latest research that's happening, which many of them have contributed to.

We organize a biannual event where we invite all the people who have contributed to our research, and we share the most recent findings with them. I'm very focused on trying to get that research back to the patient.

Here in Canada and also throughout the world, there are many different patient conferences where I try to go and share some of the research findings.

How has the Faculty of Nursing supported your research?

I've been very fortunate. I have a professorship now from the Faculty of Nursing from the Bloomberg donation that supports my research

financially so I'm able to do research without having to worry all the time about applying for external research funding.

Research at the University of Toronto is a big part of what we do as professors. My goal is to not only conduct world leading research in the area of cancer genetics, but also to translate the research findings so that patients all over the world can benefit from this research. U of T provides valuable resources to enable me to do this.

What would you like others to take away from your research?

Genetic testing for cancer-causing genes can benefit those with and without cancer. I think it's important for people to know that these genes do exist. We can test for them, and if we do find that there is a problem or a genetic mutation, we can do something about it. I believe genetic testing should be available to every person who wants the information, and I think that we are slowly making our way there.

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