Coping with genetic predisposition to cancer

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Kelly Metcalfe, a professor at the Bloomberg Faculty of Nursing, focuses her research on a unique population of women faced with some really tough decisions and helps them reach satisfactory conclusions.

What makes these women unique?

They have a strong history of breast cancer in their families and are at a high degree of risk in developing the disease due to a mutation in the BRCA 1 and BRCA 2 genes.
For these women — five to 10 per cent of those who get breast cancer — preventing breast cancer takes on a different meaning.

BRCA1 and BRCA2 are tumour suppressor genes. Most people have two working copies but these women usually have only one, which gives them less protection against tumour growth. According to the Ontario Ministry of Health guidelines, women with a strong family history of breast cancer can qualify for genetic testing to detect for these mutations.

“We all have the BRCA1 and BRCA2 gene; when they work, they suppress tumours and prevent cancer from starting. Those who have the gene mutation only have one good gene and when that stops working that’s when the cancer starts to develop,” said Metcalfe.

Women with the mutation have an 80 per cent chance of getting breast cancer and a 40 per cent chance of getting ovarian cancer, whereas average women have only an 11 per cent chance of getting breast cancer

and a one per cent chance of getting ovarian cancer.

For the average woman, minimizing the risks is about fundamental lifestyle choices, such as regular exercise and a good diet. However, it will not help reduce the risk of breast cancer for women who have tested positive for the BRCA1 and BRCA2 gene mutation. For these high risk women, the options include preventative removal of the breasts and ovaries and risk-reducing drugs such as tamoxifen.

Although these options may seem aggressive, consider their efficacy: a preventative mastectomy reduces their risk of getting breast cancer by almost 100 per cent; a preventative ovariectomy (before age 50) reduces the risk of breast cancer by about 50 per cent, with a risk reduction of about 90 per cent for ovarian cancer; and tamoxifen reduces the risk by about 50 per cent.

For the women who undergo genetic testing, cancer has played a significant role in their lives and they want to take precautions to make sure that it doesn't happen to them.

"We know this group of women will get breast cancer and we can actually do something about it. I know there is something these women can do to prevent it," said Metcalfe. Deciding on what preventative steps to take is where Metcalfe's research comes in.

Metcalfe developed a decision aid tool — currently undergoing randomized trials — that helps these high-risk women review all of their prevention options, the "pros" and "cons" of each option and the side effects. It also helps a woman determine what is most important to her. For example, if it's important for a woman to keep her breasts, then the tool rules out preventative mastectomy and helps her consider some of the other options, so she can keep her breasts.

“It’s not just an educational tool to teach them about the options, it helps them work through what would be the best decision for them,” said Metcalfe.

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