Helene Hudson Memorial Lectureship CANO 2008

by Patricia Benjaminson

Abstract

One in nine women will develop breast cancer in her lifetime (Canadian Cancer Society, 2007). Hereditary breast cancer accounts for only five to 10 per cent of all breast cancers. However, women carrying a single high-penetrance gene mutation have a 40% to 80% chance of developing breast cancer (Fackenthal & Olopade, 2007). Most of these breast cancers occur in women under the age of 50. The BRCA 1 gene mutation was first reported in 1994, and the BRCA 2 gene mutation in 1995.

The BRCA 2 gene mutation is often carried in males, and accounts for approximately six per cent of male breast cancer. Women with this gene mutation have a lifetime risk of developing breast cancer of between 50% and 85%, a second breast cancer of between 30% and 50%, and ovarian cancer between 10% and 20%.

Conférence à la Mémoire de Helene Hudson

Congrès de l'ACIO 2008

Abrégé

Une femme sur neuf développera un cancer du sein au cours de sa vie (Société canadienne du cancer, 2007). Le cancer du sein héréditaire représente seulement de cinq à 10 pour cent de tous les cancers du sein; cependant, les femmes porteuses d'une seule mutation génétique de pénétrance élevée ont de 40 % à 80 % de chances de développer un cancer du sein (Nat. Rev. Cancer, 2007). La majorité de ces cancers du sein surviennent chez les femmes âgées de moins de cinquante ans. C'est en 1994 que la mutation génétique BRCA1 a été signalée pour la première fois tandis que la mutation génétique BRCA2 l'a été en 1995.

Ce sont souvent les hommes qui sont porteurs de la mutation BRCA2 laquelle est responsable d'approximativement six pour cent des cancers du sein masculins. Les femmes porteuses de cette mutation génétique ont un risque à vie de développer le cancer du sein situé entre 50 % et 85 %, un risque de développer un second cancer du sein situé entre 30 % et 50 % et un risque de cancer de l'ovaire variant entre 10 % et 20 %. Chaque parent porteur de la mutation génétique BRCA2 a 50 % de chances de transmettre cette mutation génétique à ses enfants (NCI, 2006).

Il est difficile de prévoir l'impact émotionnel qu'aura la prise de connaissance d'une telle information sur le risque de cancer. Lorsqu'on fournit à la personne concernée de l'information sur la chirurgie visant à réduire les risques, sur la chimioprévention, sur l'évitement des risques de cancer et sur l'augmentation de la fréquence de dépistage, comment cette personne prend-elle des décisions?

Je vous invite à marcher à mes côtés tandis que je relate comment ma famille a découvert que nous étions porteurs de la mutation génétique fondatrice d'Islande, les pas que nous avons faits ensemble durant le processus de dépistage et enfin, le processus décisionnel suivi par les membres de la famille ayant obtenu un résultat positif au test génétique. Nous allons diriger notre attention sur ma sœur Rita—des journées ordinaires, un être extraordinaire.

Each parent with the BRCA 2 gene mutation has a 50% chance of passing this gene mutation to their children (National Cancer Institute, 2006).

The emotional impact of receiving cancer risk information such as this is difficult to predict. When presented with information about risk-reduction surgery, chemoprevention, risk avoidance and increased screening, how does one make decisions?

Walk with me, as I share how my family discovered we carry the Icelandic founder gene mutation, the steps we took together during the testing process, and the decision-making by the family members who tested positive. We'll focus on my sister Rita—ordinary days, an extraordinary woman.

I was honoured to have been awarded the Helene Hudson Memorial Lectureship at the 2008 CANO Conference in beautiful St. John's, Newfoundland, and pleased to have had this opportunity to share with my oncology nursing colleagues some of what I have learned about hereditary breast cancer, genetic screening, and risk reduction options.

The Helene Hudson Memorial Lectureship honours the memory of Helene Hudson, a remarkable oncology nurse who is remembered for her vision, compassion, and determination. Helene had a wonderful sense of humour and a real appreciation for the "storytelling" aspect of nursing. She took her job very seriously, but never herself. Helene and Winnipeg oncologist Ian Maxwell established the oncology clinic at the Victoria General Hospital. Helene was one of the organizers of the first National Oncology Nursing Symposium held in Winnipeg in 1983, where discussions took place that would lead to the establishment of CANO a few years later. Helene was a founding member of CanSurmount, a support group for cancer patients and their families.

In 1989, Helene reflected on what it meant to her to be an oncology nurse: "I do not", she stated, "consider lack of cure or progression of disease a failure. Death is not a failure either. There is a place for skillful and creative nursing care for these patients, from diagnosis to cure or to death. ...The opportunity to reach out and help another human being in a meaningful way should not be taken for granted. It is the very essence of nursing. Working with cancer patients brings joy, satisfaction, and meaning. Patients I have worked with have touched my life in a very special way. Health care professionals are valiant soldiers in the war on cancer, but the real heroes are our patients, whose courage and vitality serve as an inspiration to us all."

Before her sudden death in 1993, Helene touched the lives of so many people who will always remember her dedication to cancer patients and their families. Helene's laughter is still heard in the memories of the many people she worked with and cared for.

During my talk, I briefly reviewed BRCA gene mutations, provided a short history of breast cancer in my family, and discussed genetic screening and risk-reduction options for men and women who test BRCA gene mutation positive. I focused on my sister Rita's decision-making and her surgeries, the dilemma of when to tell the children and living with the knowledge of being at high risk for breast and other cancers.

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BRCA gene mutations

It is estimated that between five and 10 per cent of all cancers can be attributed to inherited mutations in specific genes involved in cancer regulation. A defective or mutated tumour suppressor gene cannot stop cells from uncontrolled proliferation rendering the cell unable to suppress the changes that lead to cancer.

BRCA 1 and BRCA 2 are tumour suppressor genes believed to have a role in DNA repair, gene transcription, and cell-cycle control. The BRCA 1 gene, discovered in 1994, is located on chromosome 17. The BRCA 2 gene is located on chromosome 13 and was identified in 1995. Commercial testing for mutations in these genes was first available in 1996. Close to 2,000 distinct mutations and sequence variations have been described in the BRCA 1 and BRCA 2 genes.

Although numerous factors affect the likelihood of developing breast cancer, an inherited mutation in the BRCA 1 or BRCA 2 gene is the strongest breast cancer predictor known.

In North America, one in every 300 to 800 people is estimated to carry BRCA gene mutations. Many people incorrectly assume that paternal transmission is not possible. However, mutations in these genes are inherited in an autosomal dominant pattern. This means that a mutated gene can be transmitted by either parent to their children. A parent who is a carrier of the gene mutation has a 50% chance of passing the altered gene to each of his or her children. Another way of saying this is that each child of a carrier parent has a 50-50 chance of inheriting the gene mutation. A person only needs to inherit one altered copy of the gene to be at increased risk.

A small number of prevalent mutations, called "founder" mutations, have been identified in different ethnic groups. For example, two specific BRCA 1 mutations and a BRCA 2 mutation are common in Ashkenazi Jews who trace their roots to Central and Eastern Europe.

The Icelandic founder mutation, BRCA 2 999 del 5 is the sole high-frequency founder mutation in Iceland.

Women who carry a BRCA 2 gene mutation have a lifetime risk of developing breast cancer of up to 85%, a second breast cancer of up to 50%, and ovarian cancer up to 20%. Men have a risk of breast cancer of greater than 6% and are at increased risk of prostate cancer, which some studies have shown to be more aggressive than sporadic

cases. Both men and women are thought to be at an increased risk of developing cancers of the pancreas, buccal cavity, pharynx, stomach, and gallbladder, as well as melanoma and hematopoietic malignancies.

Short family history

My grandfather, Skuli Benjaminson, was born in a small village in Iceland in 1879. He immigrated to Canada with his parents and older sister in 1883. They settled on Lake Winnipeg 10 miles north of Gimli, Manitoba. In 1895, when Skuli was 16, his father died after suffering ill health for more than two years.

Several years later, Skuli married Laufey, also of Icelandic descent and, in just six years, they had four children, two girls and two boys. Laufey died in 1921 following a lengthy illness when their youngest child, my father Fred, was only two. We were never aware of the cause of her death.

When my grandfather's second wife, Setta, suffered a debilitating stroke, my father's unmarried sister, my Aunt Inga, moved back home to help with her care. Our extended family was very close, frequently gathering for Sunday dinners at the home my grandfather built on the outskirts of Winnipeg.

In 1965, to cope with Setta's increasing care needs, my grandparents, Aunt Inga, my parents, my two brothers, two sisters and I all moved into an eight-bedroom house so my mother, a nurse, could provide Setta's care.

Aunt Inga was diagnosed with breast cancer when she was 46 years old. She was admitted to hospital on February 16, 1963, for a radical mastectomy and was discharged three weeks later on March 7. Inga started radiation treatments April 4.

Because we all lived in the same house, we shared certain experiences with Aunt Inga to which, as children, we would not normally have been exposed. Inga had considerable lymphedema and decreased strength in her right arm. She also experienced recurring bouts of depression and difficulty coping with her change in body image and life roles. A notation in our family diary dated April 12 stated—"Inga deep in gloom today". She frequently expressed her embarrassment when her prosthesis would slip from the confines of her bra becoming visible above the neckline of her blouse or dress, or when she had difficulty rising from a chair.

Figure One.

In February, 1990, at the age of 73, Auntie Inga died of metastatic carcinoma of unknown origin.

While vacationing with her husband in British Columbia in January, 1992, my sister Rita, age 44, discovered a lump in her left breast. She knew intuitively that she had breast cancer and on her return to Manitoba sought a referral to a surgeon. I accompanied her to the surgeon's office to receive her diagnosis. She had a biopsy, which was positive for a grade 3, T1 N0 M0 ER/PR negative infiltrating and intraductal comedocarcinoma. Rita faced her diagnosis and treatment with incredible strength of character, humour, and a positive outlook. Rita had a segmental resection and axillary dissection followed by radiation therapy. Rita continued on to receive chemotherapy as per the NSABP B.23 clinical trial. This was a double-blinded clinical trial randomizing women to receive CMF or AC plus or minus tamoxifen. Rita received her chemotherapy at the oncology department in the Victoria General Hospital. One of the wonderful oncology nurses administering treatment and caring for Rita was Helene Hudson. Rita is a laboratory technologist and x-ray technician who always takes the time to provide information, care and comfort to patients and their families, interacting with gentle humour and Helene provided care to Rita with the same sense of compassion and humour.

In February of 2006, our 40-year-old niece discovered a breast lump. She had a positive core biopsy and, while waiting for her surgery, she researched diagnosis and treatment of breast cancer. The province in which she lives was experiencing an acute shortage of medical oncologists. Not content to wait passively, she organized her own out-of-province PET scan, which revealed a metabolically active axillary node. She proceeded to have a mastectomy and node dissection with immediate reconstruction, then started what she was told would be a very lengthy wait to see a medical oncologist.

In an e-mail correspondence she stated, "Auntie Pat—I don't know how to cope with this delay—I feel like I have a time bomb waiting to go off inside my body". With her permission, I began to actively advocate for her earlier treatment and assisted her to navigate her way through the health care system. In a discussion with one of the referral nurses at her local cancer centre, we made arrangements for her to be seen at CancerCare Manitoba in Winnipeg. Her work-up was completed and she was booked for appointments for assessment with radiation oncology and medical oncology and received her first cycle of chemotherapy in Winnipeg. With this done, her care could be assumed in her home province and she continued her treatments without delay. At her request, either her Auntie Rita or I accompanied her to her appointments. The radiation oncologist reviewed our family history and referred her to the hereditary breast cancer clinic.

Genetic screening (how we found out)

It is important that all women considered at risk for carrying a BRCA gene mutation should be referred to a specialized hereditary breast cancer clinic for genetic counselling and management.

Genetic information is complex and can easily be misunderstood by people with little medical knowledge. In Canada, our health insurance is not affected by results of genetic testing. However, there is a potential for the results to be used by life insurance companies to deny or limit coverage to people who are known to be gene mutation carriers. As you must disclose such information if asked, it is suggested you review all your insurance policies including life and disability, before you have any genetic testing done. A recent federal legislation in the United States, the Genetic Information and Nondiscrimination Act, was signed into law in May 2008 and, when enacted, will provide protection with respect to insurance and employment issues.

Genetic testing always starts with a family member who has been diagnosed with breast or ovarian cancer. However, genetic test results impact not only the person who is tested, but also that person's entire biologic family. There are many questions that should be considered before genetic testing. Is there an ideal time to involve family? Is it before the testing? Or once the results are received? What if I don't want to share my results? What if some members of my family don't want to know? Can this impact my insurance? Will this knowledge impact my children's future? Is it possible someone may not wish to marry a person who carries a gene mutation? Or employ someone with a gene mutation?

The genetics counsellor reviews family medical information including first-degree relatives (parents, siblings, children), second-degree relatives (aunts, uncles, grandparents), and third-degree relatives (cousins). This can be complicated, as our relatives' medical information is not always known. The genetics counsellor must have knowledge and sensitivity and the ability to provide support to the patient and family throughout this process. In order to ensure informed consent, she must provide education and counselling, including the purpose of genetic testing, the implications of possible test results (including psychosocial), the implications for the family, how and when and by whom results will be communicated, and the options for risk reduction or increased surveillance.

The Hereditary Breast Cancer Clinic at CancerCare Manitoba encourages patients to bring family members to their appointments, if desired. Our niece invited her sister, their father (my brother), Rita and me. My self-appointed role at these appointments was to take notes, ask appropriate questions, and act as an advocate and support for my family.

Knowing your genetic status has the potential to be empowering or frightening or overwhelming. It is important to consider how you will feel **before** you are tested. As one of my nursing colleagues stated when speaking of a friend, "It's funny how some people feel safer not knowing".

Risks associated with genetic testing are the psychological and psychosocial impact. People found to carry a cancer susceptibility mutation might experience anxiety, depression, anger, feelings of vulnerability, and guilt about possibly having passed the mutation to their children.

The genetics counsellor must be sensitive in assessing the individual's response to the news. At our first visit, despite being reminded that Rita had also been pre-menopausal at diagnosis, the medical resident was initially adamant that our niece was the only one who qualified for initial testing. When the resident left the room, our niece stated she did not wish to share her results with the family, as she did not want her children to know the results and, if she kept the information to herself, there would be no way they would inadvertently discover the information. Our initial response to this declaration was, understandably, somewhat emotional and had the potential to increase the stress among the family. Her sister, their Aunt Rita and I all had the same reaction—what about our children! Happily we were all able to remain quite calm and supportive, but I could see how easily situations like this could cause considerable family distress. The genetics counsellor gently assured our group that, although individual test results would be kept confidential, the appropriate family members would be offered testing based on the results. When the geneticist returned with the medical resident, he agreed that initial testing should include Rita, as well as our niece. Because of our Icelandic heritage, the testing would focus on the ethnic-specific alleles.

Genetic testing of families can be a very lengthy process. Patients who have had breast or ovarian cancer are tested first, then, if positive, that patient's siblings and their children (if adult). It can take up to three months to get each result. Contacting extended family members, especially in an Icelandic family with many surnames and uncertainty regarding who are biologic cousins, can be complicated.

There are basically three potential results:

- a positive result means that a gene mutation has been identified
- a negative result with a known mutation in the family (also referred to as a true negative)
- a negative result with no known mutation in the family (considered to be indeterminate or inconclusive—essentially little information has been gained).

While there remains some controversy, it is generally believed that a true negative result means that the person has the same risk as the general population. The general population risk for a woman developing breast cancer in her lifetime is reported as 11% to 14%.

Figure Two illustrates the cancer cases in my family tree. The dark gray represents breast cancer and the black other cancers not believed to be related to BRCA gene mutations. The top two are my paternal grandparents—I've placed question marks on them as we do not know which one of them carried the gene mutation. Of their four children, we know my father (on the right with the plus sign) was a carrier. Aunt Inga probably was too, and it is possible that my uncle carried the gene mutation as two of his three daughters had breast cancer. Their test results are not yet known. I only continued with subsequent generations for those who are carriers after my generation. Of my siblings and I, three of five tested positive and of their children, three are positive, two are negative and two are not yet tested. Their children are all minors and are, therefore, untested.

Figure Two.

The cancers that occur in people with a BRCA 1 or BRCA 2 gene mutation do not occur in childhood. As there is no medical benefit in this case in offering genetic testing to minors, it is recommended to wait until the individual is adult and able to make her or his own informed decision about genetic testing. It is probable that by the time these children reach adulthood, there will be more effective surveillance and risk-reduction strategies available.

“Survivor” feelings

The emotional impact of receiving genetic test results is very individual. It may be very different in someone who has already had a cancer diagnosis and someone who tests positive, but has never had cancer. Those found not to carry a mutation might experience a mixture of profound relief and guilt. In our family, it was easy to share negative test results with Rita, as she was very clear that she was praying for everyone else to be negative and had no “why me and not you” feelings that are frequently experienced in other families.

It has been reported, however, that some people who had prepared themselves for a positive test result, when told their result was negative, found it difficult to emotionally “go back” to a general

population risk. They continued to feel high-risk, but without the options to decrease their perceived risk. This feeling is only enhanced by the controversy in the literature, as some studies seem to indicate that those who test negative are, indeed, still at increased risk.

Because each generation of Icelandic people change their surnames and men and women have slightly different surnames, as a child I used to joke with my father about how every Icelandic person I met turned out to be our cousin. I grew up proud of my Icelandic heritage and was proud of my “good strong Icelandic genes”. On discovering my family carries an Icelandic founder gene mutation, I experienced an overwhelming feeling of angry betrayal by our ancestors.

Risk reduction/monitoring: Men

Men who have inherited a BRCA gene mutation have an increased risk of cancer, but not as dramatic as for women. It has been reported that despite this increased cancer risk and perhaps because the risk is not as dramatic as for women, there seems to be a focus on informing and testing female family members. In our case, the men in my family received equal attention during the information session and testing process. However, those who tested positive for the gene mutation received little or no risk reduction/monitoring information and were not referred further. With their permission, their family physicians were notified and the onus was left with them for follow-up. I, therefore, provided each of my brothers and nephews with a copy of the National Comprehensive Cancer Network Hereditary Breast and Ovarian Cancer (NCCN HBOC) Management Recommendations for men, which advises:

- breast self-exam training and monthly practice
- semi-annual clinical breast exam
- consider baseline mammogram, annual mammogram if gynecomastia or parenchymal/glandular breast density on baseline study
- adhere to screening guidelines for prostate cancer as described in the NCCN prostate cancer early detection guidelines.

Risk reduction/monitoring: Women

Compared to sporadic cases of breast cancer, BRCA cases tend to have medullary pathology; estrogen and progesterone receptor negative; and higher grade. When compared with women with sporadic breast cancer at five, 10 and 15 years, women who are BRCA carriers and were treated with lumpectomy and radiation have a considerably higher risk of local recurrence, as well as contralateral breast cancers.

Data seem to indicate that some BRCA mutation carriers will **never** develop breast or ovarian cancer in their lifetime, but identification of these individuals is currently impossible.

As with any other statistic we use with our patients when discussing risk and outcomes, the risk is not patient-specific but, rather, a calculated average risk. We are unable to state “Rita you have a 60% chance of another breast cancer,” as we do not possess the knowledge to be that specific—rather we tell her she has “up to” when, in fact, she may never get another cancer. It is important to emphasize—not everyone who tests positive will develop cancer. Not everyone who tests positive and has had a breast cancer will get another breast cancer. This is what makes risk-reduction decisions so complicated. The role of the genetics counsellors and oncology nurses is vitally important in assisting patients to understand that the only right answer is the one they choose.

It is believed by some researchers that breast cancer risk declines with age for carriers of BRCA1 gene mutations, but not for BRCA 2 gene mutation carriers.

**NCCN HBOC practice guidelines in oncology,
2008 recommendations for women:**

- breast self examination training and monthly BSE starting at age 18
- clinical breast exam starting at age 25
- annual mammogram and breast MRI screening starting at age 25 (individualized based on earliest age onset in family)

- discuss option of risk-reducing mastectomy
- discuss option of risk-reducing salpingo-oophorectomy
- for those not electing to have risk-reduction surgery, concurrent transvaginal ultrasound + CA-125 every six mo starting at age 35 or five to 10 years earlier than earliest age of first diagnosis of ovarian cancer in the family
- consider chemoprevention
- consider investigational imaging and screening studies when available.

It bears repeating that women who test positive for a BRCA gene mutation face difficult choices to manage their cancer risk. Little data are available regarding this decision-making process.

To make it slightly less complicated, the nurse can assist patients to understand that there are essentially three options: increased surveillance, chemoprevention, prophylactic surgery. These options should be carefully explained.

(Some but very few still consider a fourth option:

- risk avoidance

Certain lifestyle behaviours that decrease cancer risk such as regular exercise and limiting alcohol intake, have been shown to decrease risk in the general population, but the effects on people with BRCA 1 or BRCA 2 gene mutations are unknown.)

None of these options eliminate the risk of cancer.

Surveillance

Careful surveillance will identify many, but not most, early breast cancers. BRCA mutation carriers should be taught breast self-exam and encouraged to practise monthly. Beginning at age 25, they should have yearly MRI and mammography, as well as clinical breast exams every six months.

Screening for ovarian cancer, as specified in the NCCN guidelines, is thought by many to be ineffective and inefficient and is, therefore, not an option in many centres. A new blood test hoped to detect ovarian cancer with 99% accuracy is being assessed in a phase three evaluation.

It should be stressed to patients that the best result of a surveillance approach is detecting breast or ovarian cancer at an early stage. This approach, obviously, does nothing to prevent cancer.

Chemoprevention

Chemoprevention is another option. Tamoxifen has been shown to decrease the risk of breast cancer by about 50% for BRCA 2 mutation carriers, but not for BRCA 1 mutation carriers.

Prophylactic surgery

Although still considered by some to be a radical and controversial intervention, prophylactic surgery is the most effective preventive strategy available. Prophylactic bilateral salpingo-oophorectomy (PBSO) is reported to reduce the risk of ovarian cancer by 85% to 95% and breast cancer by 46% to 68%. Bilateral prophylactic mastectomy reduces the risk of breast cancer by 85% to 95%. Both surgeries have potential risks and benefits that complicate the decision-making process.

When is the best time to have prophylactic surgery?

Decision-making (Rita)

At the time of receiving positive results of the genetic test, our female family members were advised that they could be referred to the gynecology-oncology clinic and the surgical oncology and plastic surgery clinics for discussion of risk management options. Despite the fact that these clinics provided excellent information and support, perhaps because they were separate appointments, there was a feeling that something was missing. Studies have shown that many women feel they received insufficient information about prophylactic surgery.

It has been stated that when decision-making closely follows disclosure, women may experience increased levels of anxiety and distress, affecting their ability to retain information, thus affecting their ability to make decisions.

Rita's advice is to take at least one family member with you (in addition to your husband) to write down information and ask questions you and your spouse may be feeling too overwhelmed to think of asking. I would like to encourage nurses to offer to assume that role when patients attend appointments alone. Nurses and physicians should not assume that because the person has an appointment to discuss the surgical risk-reduction options that this means she has already made a decision. The genetics counsellor and the surgical-oncology nurses all provided their phone numbers and encouraged patients to call with questions, but not everyone is comfortable with this self-directed approach. A planned follow-up appointment with the genetics counsellor or the surgical nurses to review the options and answer additional questions would be very helpful in the decision-making process.

Rita was advised that she would need to be off work for at least eight weeks following bilateral mastectomy with immediate reconstruction and three to four weeks following the laparoscopic bilateral salpingo-oophorectomy with vaginal hysterectomy. She was offered the opportunity to have the surgeries at the same time, which would decrease her time off work, but would require more than eight hours of anesthesia and increase her risk for DVTs.

Following the breast surgery, she would be in hospital for four or five days, and would have hourly ultrasound of the reconstructed breasts for the first 24 hours. To avoid stress on the abdomen where the tissue and blood vessels were harvested, she would not be able to stand straight or lie flat and would need to wear a three-panel abdominal binder and a sports bra for four weeks. She should not lift anything heavier than five pounds, avoid sexual activity and strenuous exercise for at least four weeks and should not drive for three weeks.

Recently, the public is being made more aware of hereditary cancers through news stories about famous people such as 36-year-old actress Christina Applegate who, in August of 2008, announced her diagnosis of breast cancer, her positive test for the BRCA 1 gene mutation, and her bilateral mastectomy, which she called a "logical decision". And, on October 1, 2008, PBS aired a documentary called "In the Family" by filmmaker Joanna Rudnick who, at age 27, tested BRCA mutation positive.

Despite the recent increase in public awareness, some women have reported that their friends and family reacted with "shock" and "horror" when told of their decision to have prophylactic mastectomy, adding to their stress and making them hesitant to share the information with anyone else. I discovered that this is not, unfortunately, an uncommon reaction. More than one oncology nurse, unaware that I and my family were having genetic testing done, stated casually in reference to other patients, "I don't know why anyone would have genetic testing done—there is nothing they can do about it," and "How could they think of having horribly disfiguring surgery, which is probably of no benefit anyway—if it was me, I'd rather take my chances." Or the oncologist who was aware that I was awaiting my genetic test results who said, "Eww, if your test is positive you're not going to have that surgery are you?"

Prior to prophylactic surgery, nurses should discuss with patients the possibility of experiencing altered body image and the potential impact on the patient's sexual relationship. Women who are considering these surgeries should be counselled to take time with their partners to explore the meaning of their sexuality.

Rita initially decided to have the surgeries together and was booked for March 2007, but when the breast surgery was postponed she decided to proceed with the PBSO alone. Rita had been advised that her breast surgery might need to be cancelled if the surgical slot was needed for a patient diagnosed with cancer. Rita's breast surgery was postponed twice, adding to her stress and family anxiety and creating difficulties at work.

As oncology nurses, we have all met patients whose employer or coworkers think they are using their medical history as an excuse to take extra time off work for follow-up tests and appointments. They may imply that, because the patient is not really sick anymore and because they required so much time off when they were receiving active treatment, they should be scheduling their appointments on their days off or vacation time so as not to further inconvenience their coworkers and employer. Coworkers and employers who follow this line of thinking are not likely to be supportive when you require up to 12 weeks off for prophylactic surgeries and then have the date of one of the surgeries changed twice. And how on earth do we educate the public when some oncologists' and oncology nurses' initial response to prophylactic surgeries is "Eww!" or "If it was me, I'd rather take my chances!"?

Decision-making for women with BRCA gene mutations is highly individual and requires weighing the inborn risk against other life priorities and quality of life issues. For many women, after risk reduction surgery, there remains a need to confirm that "the right thing" was done by continuing to read and research risk-reduction options, and attend relevant conferences and symposiums.

Rita's surgeries

At her September 2006 appointment, Rita was advised by the gynecology-oncology team that her risk for developing ovarian cancer was up to 20%, but probably somewhat lower as she had used oral contraceptives and tamoxifen in the past. She was advised that she should also consider a hysterectomy, as there was evidence to indicate she was also at an increased risk of endometrial cancer. Rita, who has three children and, at age 58 was past her childbearing years, found the decision to proceed with these surgeries relatively easy. Rita was scheduled for a laparoscopic prophylactic bilateral salpingo-oophorectomy and vaginal hysterectomy, both minimal access surgeries with low morbidity. She was advised she would need three to four weeks off work. Rita and her husband had a vacation to Thailand planned in late December and surgery was, at her request, scheduled for March 2007.

The decision regarding managing her breast cancer risk required more consideration and deliberation. Rita initially seemed to be leaning towards increased surveillance. At her October 2006 appointment, the surgical oncologist reviewed the possible options. She advised Rita that she did not consider surveillance to be a good option for her, as surveillance is simply hoping to detect an early breast cancer. Chemoprevention was also not considered to be a good option, as Rita had already taken tamoxifen in the past. She stated it was difficult to estimate Rita's risk of another breast cancer, but thought it would be at least 25%, probably greater. Her recommendation was prophylactic bilateral mastectomy with or without reconstruction. She assured Rita she certainly did not need to make an immediate decision and recommended she first meet with the plastic surgeons to discuss reconstruction options. While awaiting Rita's decision, she requested breast MRI and booked a six-month follow-up appointment.

Rita met with the plastic surgeon in November. He reviewed the risks, benefits and alternatives of reconstruction using autologous tissue, describing the technique called DIEP flap (deep inferior epigastric artery perforator procedure) and microvascular anastomosis. This method of reconstruction utilizes blood vessels from the muscle along with overlying fat and skin. Because the muscle is left intact, abdominal strength is not compromised and there is less pain. And, as a bonus, the woman gets a tummy tuck to go along with the reconstruction. Immediate reconstruction occurs while the patient is still on the operating table following mastectomy, thus decreasing total days spent in hospital, as well as time off work. It has been estimated that DIEP surgery is readily available to only approximately 20% of women in North America and we are very lucky that this surgery is routinely offered in Manitoba. To further enhance the cosmetic result, the oncology surgeon performing the

mastectomy uses a breast reduction-type pattern skin incision, which was explained to Rita that she would use on the right breast and potentially on the left, but there was some concern regarding the viability of skin due to her previous surgery and radiation. Because of this, she may require an additional surgery a few months later to "tweak" the cosmetic result. Following healing, nipples are created and areolas are tattooed.

One of Rita's recommendations is that the surgical nurses need to show the patient what the surgical site looks like and what to expect prior to discharge from hospital. It would also be helpful to review the "what ifs"—what to do if one of the drains falls out, what to do if one of the sutures opens, etc. This teaching should include the patient's husband and be offered to her adult children if she desires. Nurses need to be aware of the potential problems with body image and sexuality and associated distress for the first year following prophylactic surgery. Supportive counselling should be offered to patients and families prior to surgery and up to a year following.

Telling the children

As referred to earlier, in the case of BRCA testing it is generally recommended to wait until the individuals are old enough to make their own informed decision about genetic testing. Most believe the "appropriate age" to screen is age 25 or five to 10 years before the earliest diagnosed breast cancer in the family.

A nursing colleague who has a friend with a BRCA 1 gene mutation stated, "It is so hard to know when it's the right time to tell the kids."

While there is little information available about how and when to disclose information to children, members of my family have strong and divergent opinions on this subject.

Rita and her family believe it is important to hide nothing from her grandchildren. "Let them see what I am doing to prevent further cancer, see how well I am coping, how happy I am."

One of our nieces is adamant that her children remain unaware until they are old enough to make their own decisions. She believes her children have been through enough learning their mother had breast cancer and watching her cope with surgeries, chemotherapy, and radiotherapy.

Her sister, who also has the BRCA 2 gene mutation but has never had cancer, shared her information with her children with age-appropriate information, and matter-of-factly explained why she was undergoing prophylactic surgery. She stated that she didn't feel right hiding this information from the children as "it is a fact of our lives". She confided that she worries, however, that the knowledge could cause stigmatization. Would it create difficulties in future relationships—maybe some people won't want to risk marrying someone or hiring someone who may test positive. She also worries that one of her children may talk about it with their cousins and cause her sister more distress.

There is a critical need for more research and information about the impact of disclosure on children. This is an excellent opportunity for nursing research to fill the gap.

Living with—Living beyond the knowledge

It has been said that cancer changes lives in many ways and it changes lives forever.

Nearly every cancer survivor will agree that having had cancer has given them a deeper appreciation for life. However, I don't think I have met anyone who has had cancer who hasn't worried to some degree about a recurrence, especially acute around the time of another illness or just prior to routine follow-up appointments.

In the spring of 2008, Rita developed a cold that she couldn't seem to shake followed a few weeks later by the development of a harsh, dry cough, especially troublesome at night accompanied by shortness

of breath on exertion, considerable fatigue and lassitude. She pushed herself to continue to work full-time, as well as taking call as one her coworkers needed time off to assist her ill father. After a few weeks, Rita experienced a piercing right-sided lower ribcage pain with each cough and thought perhaps the harsh coughing had resulted in a fractured rib. She spoke with one of the physicians at the hospital where she works who ordered an x-ray, which revealed a right pleural effusion. This physician provided Rita with a prescription for antibiotics and, aware of Rita's history of breast cancer, arranged an urgent CT chest, which revealed no other abnormalities in addition to the pleural effusion. Rita was then referred to chest medicine for assessment and diagnostic thoracentesis. Cytology was negative for malignant cells. The effusion was determined to be exudative, inflammation caused by pneumonia. Although we all knew the pleural effusion was unlikely to have been malignant—after all, following her mastectomies all tissue had been free of malignancy—the fear, no matter how distant, is always present.

Rita's approach to life is to "take it as it comes" and "worry is a waste of time and imagination". She is a good example to everyone, as she makes the most of every day and truly enjoys life. One of Rita's three children also tested BRCA 2 gene mutation positive. Prior to receiving his test results, he stated there was no point in worrying about something that had already been determined. He believed he was prepared for receiving a positive result, yet, on receiving the news, was surprised at how upset he felt initially. This news didn't just affect him—it has the potential to impact the health and well-being of his children, but he must live with this uncertainty until they are adults. Living with the knowledge, we are aware that, as the next generation enters adulthood and make their decisions about genetic testing, we will be again praying for the 50-50 chance to be on "our side."

I am aware of people in other families who have declined genetic testing, as they "feel safer not knowing," and others who have received positive results and have spent months and months agonizing over their options. There are many, many others, however, who believe that the knowledge has empowered them to know their cancer risk and have options for decreasing that risk. While I was awaiting my genetic test result, a dear friend who has been living with metastatic breast cancer for just over five years said, "If you test positive, do not hesitate. Do everything you can to decrease your risk. I would give anything to have been given that opportunity. Life is just too precious."

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We must not allow the clock and the calendar to blind us to the fact that each moment of life is a miracle and a mystery.

— H.G. Wells

Conclusion

The impact of receiving predisposition genetic test results can last a lifetime and longer. Nurses need to assess how we interact with women who have received a positive or uninformative BRCA mutation test, discuss risk information with patients and families, provide options for follow-up after genetic testing, and offer long-term planning for risk reduction. Women contemplating prophylactic surgery need understanding, well-informed people to talk to, and a safe place in which to express their feelings.

It has been 16 years since Helene Hudson's untimely death and 17 years since my sister Rita's diagnosis of breast cancer. During those years, we have acquired the knowledge and ability to more accurately identify individuals at high risk of breast cancer and offer risk-reduction options. It is a challenge for nurses to keep sufficiently informed to be able to incorporate up-to-date genetics information in patient teaching and support but, as core members of health care teams, it is a challenge we must meet. I know Helene Hudson would if she was here.

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