Genes, discovery and life decisions

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I was 34 years old when I found out that I had inherited the *BRCA1* mutation. I cannot remember what the day was like when I met with the genetic counsellor to receive my results. Was pathetic fallacy at play? If the day was foggy, stormy and rainy, I may have anticipated that I had inherited the *BRCA1* mutation on chromosome 17. If it had been sunny with a blue sky, then chromosome 17 would have been spared. Fifty-fifty chance, the flip of a coin. I probably just wandered from one unit of the hospital to my appointment and entered into a role reversal. Me the patient instead of the medical resident. I had just started my training to become a psychiatrist.

I had tried to prepare myself psychologically for this result. I thought about other family members who did not have the opportunity to make these decisions. I never met my maternal grandmother, Jeanette Mednick, who graduated from the Ontario College of Pharmacy in 1946 and was one of the earliest recipients of the Parke Medal for having the highest marks in third year. She had three children and liked ballet, opera, and theater. Jeanette died in 1968 at the age of 43 years from ovarian cancer. I did get to know my aunt, Adrienne Yamaguchi, my mother's sister who lived in Hawaii. She was a nurse who also had three children. Adrienne's breast cancer was diagnosed in her early forties, but even after hormone therapy and radiation, she passed away in 2006 at the age of 50 years. I was 25 years old at the time and this was heartbreaking. I have felt her passing and loss even more as I approach her lifespan.

Deciding to get tested was not the difficult decision; it was when to get tested that was more complicated. I made that decision after my husband and I had two children. I wanted to wait until I could make better use of the information if I was going to proceed with any risk reducing measures. However, even after I found out, I still couldn't decide what to do. I could take control and get ahead of the possibility of developing breast or ovarian cancer. I knew that I could potentially develop other cancers and also succumb to any other form of death, but this way I could be in control of this specific situation. Or I could do nothing beyond the usual screening protocol.

I was now known as a "cancer previvor," a term used to refer to people who have not received a diagnosis of cancer but have a known predisposition because of a genetic mutation. I felt that carrying around this knowledge was not unlike having a diagnosis of an unruptured intracranial brain aneurysm less than 7 mm in size, where the suggested management is to observe with repeat imaging unless the patient is young or has other risk factors. In both cases, developing cancer or the possibility of aneurysm rupture are there in the background — present but not necessarily causing any harm. Or is it? Studies have shown that there is a psychological burden to receiving *BRCA1/2* test results, as well as news of a cerebral aneurysms. I wanted to remain a previvor, but I also felt like a cancer diagnosis was looming.

The timing of these surgeries was also a challenge. It is not easy to plan for time off during a rigid training program. At first, I chose increased surveillance with breast MRI and mammography. My husband supported this decision as we discussed the options. However, I did eventually want to proceed with the prophylactic surgeries because the thought of always waiting for MRI and mammography results made me nauseous and fearful. My fertility journey was not over quite yet. When I was 36 years old, we had a third child. Then I felt ready. I pictured my fallopian tubes, ovaries and breasts hanging on the clothesline like undergarments, cleansed of their unwanted genetic legacy. Leaving my body with a map of scars.

The weight of these decisions was at times suffocating. I was straddling two worlds, diagnosed and undiagnosed, diseased and healthy. I felt like my feet were on either side of a rushing stream looking down into the whirlpools, not knowing on which side I would eventually land. On one side was the risk of cancer, and the other side was surgically induced menopause. Which one was I going to choose? When my youngest son was 9 months old, I chose to have surgery to remove my fallopian tubes and ovaries to reduce my risk of developing ovarian and breast cancer.

The hormone changes I experienced cannot be overlooked. I was worried about my ability to continue to breastfeed after surgery while estrogen, progesterone and testosterone were acutely depleted from my body. The symptoms — vaginal dryness, hot flashes, insomnia, irritability, night sweats and mood changes — were uncomfortable and unappealing. I was 37 years old and found it hard to tease out menopausal symptoms from the sleep disturbance, mood changes and overall grouchiness of parenting. Fortunately, I was able to meet with a gynecological oncologist who gave me expert advice about postsurgical menopause management.

As if that was not enough, while I was experiencing hormone adjustment and raising children, I was also studying for my Royal College psychiatry exam, and the entire world was dealing with a global pandemic. Despite feeling overwhelmed and nervous, I still decided to proceed

with the double mastectomy. First, I got an updated breast MRI and mammogram. After the mammogram, my family doctor called me to say that I would need further testing. I knew this was a possibility but, immediately, I thought that cancer had caught up with me. My elevated risk was there; I had not been able to outrun it. As I sat waiting for my repeat mammogram, the waiting area plexiglass barriers acted as containers of my angst and uncertainty. I imagined the worst, but fortunately received the "all clear." At that moment, any ambivalence I had harboured about my decision to have the breast surgery was quashed.

Timing, career, family, the juggle of life is a topsy-turvy unbalancing act. The breast surgery was psychologically more complicated than I expected. Before the operation, I occupied my time with finding pillows. Wedge pillows to support back sleeping and heart-shaped pillows to position under the armpit to prevent rolling onto the surgical site. I leaned heavily on support from friends who had mastectomies after a cancer diagnosis and family members who had had prophylactic surgeries. Thankfully, they were quick to reply to my cascade of questions. What type of bra could I use with drain bulb management? How long is the recovery process? What would the scars look like? I also sought lived experience by looking at blogs and reading group chats online. The courageous women who publicly told their stories guided me through my expectations and allowed me to visualize what was to come.

I felt an intense relief the moment I realized my breasts were gone. But my body did not feel like mine anymore. The breasts that had nursed my children were replaced with numb inflatable balloons. I was uncomfortable and it was awkward dealing with the surgical drains, along with collecting and measuring the fluid. Was it getting darker or lighter? Did I have an infection? I was grateful for the home visits from the community health nurses who provided dressing changes and unflappable reassurance.

This story is about me and my family, our genes and our losses that have been felt throughout the generations. Jeanette exists in black and white photos, in my mother's piano music, and in my uncle's love for opera. As a woman who thrived in a scientific field, she had a quest for knowledge and a desire to learn. It has been almost two years since I had the double mastectomy and reconstruction. I do not regret my decision. I see *BRCA1* knowledge as a gift that we did not have before. Jeanette did not live to see the future of gene discovery. I am hoping that there will be more to come and women will have more evidence-based prophylactic measures from which to choose. I am now a practising psychiatrist, carrying with me this experience as a patient, always present, navigating the turns and tornado-twists of life.

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