

Genetic Counseling



A Patient's Perspective: Breast Cancer Risk

After my breast biopsy that afternoon, I had already begun my search for the top medical oncologists in my area. How could I not have cancer? My maternal grandmother and aunt were diagnosed with breast cancer in their 40s, my mother was diagnosed with breast cancer in her 50s and passed away not even two years later, and my first cousin was currently being worked up for a lump she felt just last week. Since I initiated mammograms at 35 years old, I knew it was only a matter of time.

Nine days later, I had my first visit with the medical oncologist to discuss my cancer diagnosis, prognosis, and treatment plan. I surprised myself at how calm I was during the majority of the appointment. I told him I was tentatively scheduled for a lumpectomy with my breast surgeon. Toward the end of our appointment, he asked about my occupation, social support and family history. I relayed to him the cancer diagnoses in the family including my mother, aunt, and maternal grandmother. He replied, "Do you still have your ovaries?" I looked up and made eye contact, "I am 39 years old, and of course, I still have my ovaries." The medical oncologist said it might be useful for me to speak to a genetic counselor. I was a bit confused — "I have breast cancer, only breast cancer... why the heck was he asking about my ovaries?"

As he handed me the genetic counselors contact information, he asked me if I would like to speak with her while she is here today. As I gathered my things, I answered, "why not?" A knot grew in my stomach as I entered her office. We discussed the doctor's concern and my confusion about his question. Why had no one ever questioned me about the excessive breast cancer history in the family, and more shockingly, how come I never asked? I was surprised to learn that breast cancer and ovarian cancer can actually be linked in families that carry a mutation in two particular genes, BRCA1 and BRCA2. We reviewed my family history and individualized risk assessment. We discussed reasons for and against genetic testing, why I was a good candidate, and what it would mean to carry a mutation in either of these two genes. I decided to pursue genetic testing that same day.

At my follow-up appointment, I learned that I did carry a mutation in my BRCA1 gene, and was at an increased risk to develop breast and ovarian cancer. Although I was scared, I realized not everyone has the opportunity to identify and prevent such a risk in their family. Later that month, after much talk with my family and husband, I decided to undergo a bilateral mastectomy. Having three young children, I chose to take the most risk reducing strategy.

Shortly after my recovery, I made the appointment to have my ovaries removed by a gynecologic oncologist. As I disclosed this newly found information to my family, they too jumped on the band wagon and began seeing genetic counselors to assess their risk. We were out to put an end to all this, and it felt empowering!

For more information about Genetic Counseling, please contact Beth Souders, MS, CGC, at 215.829.6528 or visit PennMedicine.org/Karnell.