

Breast cancer: a father's legacy

In this first of an episodic series of vignettes, we hear the perspective of patients, genetic counselors, and patient advocates on screening for and coping with hereditary cancer. The patients' stories are instructive and open the door to discussion on genetic testing and counseling within community practices.

A patient's story

It was early fall of 1994. I had come home from work early, to find my parents sitting in the dark, holding hands. When I asked what was going on, my mother looked at me and said, "Daddy has breast cancer." Two days later, he underwent a modified radical mastectomy with 26 nodes removed, none of which showed any activity. He went on tamoxifen and no further treatment. Genetic testing was never mentioned. Dad himself was a physician; he was being treated by a well-known oncologist, affiliated with a comprehensive cancer center.

After Dad had two strokes, his doctors forced him to go off tamoxifen. Within a matter of months, he discovered his metastatic recurrence. It was just 4 weeks shy of his 5-year survival mark. He underwent various treat-

ments, but for the most part, they were palliative. The disease was too widespread to be contained. My father died March 15, 2001.

Along the way, my sister and I both asked our gynecologists if we should be concerned about our risk of breast cancer. My doctor told me there was no known "father-daughter breast cancer link." My sister was told that when men get breast cancer, "it's a fluke." We trusted their judgment.

While nursing her daughter in July 2004, my sister found a lump in her breast. Two weeks later, she was told she had stage III invasive ductal carcinoma. She underwent chemotherapy, a modified radical mastectomy, an oophorectomy, more chemotherapy, radiotherapy, and a contralateral prophylactic mastectomy. Along the

way, my sister tested positive for the *BRCA2* mutation. In June 2005, she finished her treatment and is showing no evidence of disease today.

After my sister tested positive for the *BRCA2* mutation, it was recommended that I be tested, too. Before I got my results, I knew that if I was positive, I would have prophylactic surgery and breast reconstruction. So I began researching my options and meeting with teams of physicians. I was not surprised when the results were indeed positive. I had my surgery a month after my sister finished treatment. Two weeks after my surgery, I received a phone call from my surgical oncologist's office. The PA said, "Wow...did you ever dodge a bullet. We found stage 0 breast cancer, ductal carcinoma in situ." If I had ever questioned my decision, that would have erased any doubt.

I was given a gift—the gift of knowledge. Unfortunately, neither my sister nor my father was able to benefit from such a gift. Had they known of their risk in advance, I am certain things would have turned out differently. My father would have been more than a namesake: he would have been able to know his three incredible grandchildren who are named after him.

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Commentary by Sue Friedman, Board Chair, FORCE, Tampa, FL

Many of the people who contact Facing Our Risk of Cancer Empowered (FORCE) were informed by healthcare providers that paternal family history of breast cancer is not significant. This misinformation sometimes comes from providers who use breast cancer risk assessment models that do not take into account medical information from the paternal lineage, such as the Gail Model. Although these models are widely used, many healthcare providers do not explain to their patients the limitations of the models and disregard other risk factors.

FORCE frequently hears stories from people who were unaware of the red flags for a hereditary cancer syndrome in their family. Sadly, for some, the suspicion is not raised until yet another family member has been diagnosed. Because of her knowledge of risk, this patient was diagnosed during surgery when her cancer was preinvasive. However, had the identification of a hereditary component been made after her father's diagnosis, her sister may have been able to take preventive steps too and avoid chemotherapy and the possibility of metastatic relapse.

Ms. Friedman can be reached at suefriedman@facingourrisk.org.

Commentary by Kami Wolfe Schneider, MS, CGC, Baptist Memorial Health Care, Memphis, TN

Approximately 10%–15% of all male patients with breast cancer may have a mutation in the *BRCA1* or *BRCA2* genes. Although men in the general population have approximately a 1 in 1,000, or 0.1%, lifetime risk of developing breast cancer, men with *BRCA2* mutations have approximately a 6% lifetime risk of developing the disease. Similarly, increased risks of male breast cancer have also been observed in men with *BRCA1* mutations.

Men with mutations in these genes are also at an increased risk to develop prostate cancer, especially at younger ages than men in the general population. Current recommendations for men with *BRCA1* and *BRCA2* mutations include:

- Breast self-exam training and regular monthly practice;
- Semi-annual clinical breast exam;
- Consideration of mammography;
- Adherence to population screen-

ing guidelines for prostate cancer.

In this vignette, the patient and her sister were given incomplete information by their doctors about their hereditary risk for cancer. It is important to remember that hereditary risks for cancer can be inherited from either the maternal or paternal side of a family. Breast and ovarian cancer risk assessments can be especially complicated for paternal family lineages, because the penetrance of cancer in males with a mutation is lower than in females with a mutation (because males generally have very little breast tissue and no ovaries). Therefore, men with these risk factors are less likely to show evidence of the inherited mutation, making women who may be at risk for breast and ovarian cancers more difficult to identify.

Genetic testing for mutations in *BRCA1* and *BRCA2* allows individuals to make appropriate personalized medical management decisions based on the information. With compre-

hensive pretest genetic counseling, patients are given information regarding the risks and benefits of multiple risk reduction strategies so that cancer can be either prevented or detected early.

In this vignette, the patient was diagnosed with ductal carcinoma in situ as a result of a prophylactic mastectomy. Studies have also indicated that some women with *BRCA1* and *BRCA2* mutations who undergo prophylactic oophorectomy are found to have early stages of ovarian cancer. These cancers might not have been detected at these stages otherwise.

This vignette clearly illustrates the importance of genetic counseling for males diagnosed with breast cancer and the consequences that can occur when individuals at risk for a genetic mutation are not appropriately identified and referred for genetic counseling.

Ms. Schneider can be reached at kami.schneider@bmbcc.org.

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ABOUT THE AUTHORS

Affiliations: Ms. Rich is a Certified Genetic Counselor, and Dr. Perrier is Associate Professor, Department of Surgical Oncology, The University of Texas M. D. Anderson Cancer Center, Houston, TX.

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