
Genetic Services Policy Project

Hereditary Breast and Ovarian Cancer: A Vignette

Dr. Jamie Brown is a family physician in a mid-sized community in Oregon. She is part of a six-person family medicine group and has been in practice for 10 years.

Recently, Jennifer McCarthy presented to Dr. Brown's office for a new patient visit and well-woman check-up. Jennifer was a 25-year-old, healthy young woman of apparent Eurasian descent. Her medical history was unremarkable except for a family history of diabetes in several relatives on her mother's side and breast cancer in her mother and an aunt. Her major concern was weight gain of 15 pounds in the past two years. She was on birth control pills, which she felt contributed to the weight gain. In addition, since graduating from college three years ago, she had worked in a sedentary job that required frequent lunch meetings or after-hours drinks with clients. Her work hours made it difficult to get any regular exercise. After completing her physical exam, Dr. Brown counseled Jennifer on nutrition, physical activity, and reducing alcohol intake. She also provided patient education materials on weight management, including helpful tips for busy people and offered a referral to a dietician. As the visit was ending, Dr. Brown asked if there were any other concerns. Jennifer replied, "Well, I wonder what you think about the genetic test for breast cancer. While I was in the waiting room, I read an article about a woman who had a family history similar to mine: a mother and an aunt with breast cancer. This woman got tested through a web-based genetic testing service, and she turned out to have the gene. She had ovarian surgery and was considering breast surgery to reduce her risk of getting cancer. Should I get tested too?"

Though Dr. Brown had initially noted the family history of breast cancer, she hadn't focused on this issue given Jennifer's other immediate concerns and her young age. Dr. Brown questioned Jennifer further about her family history. Her mother, whose family emigrated from Japan to the United States, was diagnosed at age 50 with early stage breast cancer. She was doing well after lumpectomy. No one else on that side of the family had breast or ovarian cancer as far as Jennifer knew. Jennifer's paternal aunt had breast cancer and a mastectomy sometime in her early 40s. Her aunt and her father were twins and had been adopted as infants, so additional family history (e.g., ethnicity or history of cancer) was not available. Jennifer didn't think either her mom or aunt had had testing for the breast cancer gene mutations (BRCA1 and BRCA2). From Dr. Brown's recollection about cancer genetics, Jennifer's history suggested some increased risk but it wasn't clear-cut. The individuals with breast cancer were on different sides of the family, and other important data about previous generations were not available. Dr. Brown offered to do more research about the testing and suggested a follow-up appointment to further discuss the breast cancer concerns as well as check on progress with her weight control program.

At home later that evening, as Dr. Brown considered Jennifer's situation, a number of questions and thoughts came to mind: Would Jennifer or any of her family members be good candidates for the breast cancer mutation test? What additional information was needed from the affected family members? Was Jennifer's mother's Asian background a risk factor? How did her family

history fit in with other potential risk factors for breast cancer—weight, oral contraceptives, alcohol, nulliparity (no pregnancies)? Could the new electronic medical record system being installed in the family medicine office help summarize this information and provide a risk estimate based on all the factors? What would be the ramifications for Jennifer of a positive test showing that she had a mutation? Dr. Brown remembered hearing somewhere that people might have problems with insurance and genetic tests, either that insurance companies would not cover the tests or that companies might discriminate against people if they had a positive test.

After spending an hour of her evening “free time” reviewing online material from a variety of reputable sources, Dr. Brown came to the conclusion that BRCA mutation testing was unlikely to be beneficial to Jennifer at this time, particularly if the genetic status of the paternal aunt was not known. Given the aunt’s history of breast cancer at a relatively young age (<45) and lack of additional family information, the aunt might be a candidate for testing. Without data from multiple generations, an online BRCA risk calculator estimated that the aunt’s chance of having a mutation was 6.8 percent. Given the mother’s older age at diagnosis and no other affected relatives on that side of the family, the mother’s history was less concerning for a BRCA-related breast cancer. There was no evidence that the mother’s Asian background increased the risk of a BRCA mutation. Though Jennifer’s family did not appear to meet the United States Preventive Services Task Force’s evidence-based criteria for an “increased risk family,” Dr. Brown wondered if Jennifer could benefit from genetic counseling to sort through the family issues. Dr. Brown definitely did not feel competent to provide the counseling. Since there were no genetic counselors in town, Jennifer would have to make the trip to the nearest center, which was an hour away. She could also consider the web-based testing service. Dr. Brown explored the website that Jennifer mentioned and was fairly impressed with the comprehensive services that were offered, including genetic counseling and physician reports. However, the cost of testing seemed quite high (\$3,400) and might not be covered by insurance. Dr. Brown was also concerned that Jennifer might be pressured to be tested even if it wasn’t really indicated.

Given the late hour of the day, Dr. Brown decided to send Jennifer a quick email the next morning outlining her thoughts and repeating her suggestion for a follow-up visit in a few weeks. Even if they didn’t proceed with counseling or testing, Dr. Brown thought it was probably a good idea to monitor Jennifer more closely for any breast changes, encourage breast self-exam, and maybe even recommend a mammogram sometime in the next few years. Even without a BRCA mutation, Jennifer’s family history did increase her risk of developing breast cancer. A reasonable next step would be for Jennifer to contact her aunt for more information and to raise the possibility of genetic testing. Dr. Brown decided that the next time she saw her oncology colleague, Dr. Callahan, she would ask about his experience with hereditary breast cancer and if he had any additional thoughts on how to proceed.

Case Issues for Discussion

1. As reflected in this scenario, genetic issues make up only one component of overall care and considerations in primary care practice. Jennifer's priority issue and subsequently Dr. Brown's primary concern is her weight gain; the breast cancer genetic testing question comes up almost as an afterthought.
 - a. Is the current primary care delivery system set up to adequately address genetic concerns? If so, in what ways? If not, why not?
 - b. What are alternate models that might improve identification and exploration of genetic health issues in primary care?

2. While most physicians are likely aware that genetic tests such as BRCA1 and BRCA2 exist and may be helpful in certain situations, experience and proficiency with use in practice may be limited.
 - a. What type of training and/or resources would assist physicians and other health care providers in appropriately utilizing this technology?
 - b. Do these resources already exist? If so, where?
 - c. What mechanisms exist to connect physicians to these resources and how might these connections be increased?

3. Physicians may be more aware of non-genetic risk factors for a disease such as breast cancer.
 - a. How can familial/genetic information be integrated with other risk information for accurate assessment?
 - b. What tools could assist in this process?

4. The physician in this scenario took a general family history during the exam, but spent limited time on this activity until the patient asked a specific question. Afterwards, the physician spent considerable time on the breast cancer issue, both during and after the visit.
 - a. What additional issues were brought up in the family history that the physician did not address? How should she address those issues in her future care of Jennifer?
 - b. Should this time be billable/reimbursable? Why or why not?

5. The 3-generation pedigree is a gold standard in genetic services practice. Dr. Brown did inquire about additional family history, but did not construct a pedigree.
 - a. Should this be an expectation in primary care? Why or why not?
 - b. If so, how can it be implemented in a cost-effective, time-efficient way?

6. After researching the breast cancer genetic testing issue, Dr. Brown felt that a genetic counseling visit could be beneficial for Jennifer even though evidence-based guidelines suggest that her family does not meet the "increased risk" criteria for BRCA1/2 mutations.
 - a. Under what conditions should Dr. Brown recommend a genetic counseling visit to Jennifer?

7. "Curbside" consultations are common in medicine. Dr. Brown thought that her oncology colleague might have additional information about hereditary breast cancer.

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<http://depts.washington.edu/genpol>

- a. What are the potential benefits and pitfalls of such a consultation?
- b. What other types of professionals might she consult with?
- c. Are there effective models for such consultation? If so, what are they?

8. Jennifer's question about breast cancer genetic testing arose after she read an article in a popular magazine about the testing. The web-based testing service was a new concept for Dr. Brown. It was difficult to assess the quality of this retail service, and she was concerned that individuals might be pressured into testing even if they were not at high risk.

- a. Should web-based services be certified or monitored in some way? By whom?
- b. Should direct-to-consumer or direct-to-provider advertisements for genetic tests be regulated or restricted? Why or why not?

9. Dr. Brown wondered about the ramifications for Jennifer of a positive genetic test. A new term, "pre-vivor," has been coined for individuals who have a predisposition for cancer, based on a genetic risk or family history, but who have not been diagnosed with the disease.

- a. What unique concerns might pre-vivors have that cancer survivors or unaffected individuals do not have? Example resource: www.facingourrisk.org

10. Dr. Brown looked for evidence-based approaches to Jennifer's care, but as is common in many clinical situations, found gaps in the available data and recommendations. She considered ordering a mammogram on Jennifer in a few years, though the literature provides limited guidance on this issue. The American Cancer Society suggests that for someone with a family history of early breast cancer, a mammogram at age 30 could be appropriate. MRI screening may also be warranted.

- a. What options could Dr. Brown recommend for Jennifer?
- b. Are there potential harms in suggesting that Jennifer undergo increased screening procedures, when her actual risk is unclear?

11. Insurance coverage of genetic counseling and testing is a potential issue for Jennifer and her family members. BRCA testing and prophylactic treatment (surgery) based on positive results are costly.

- a. What is the impact of insurance companies denying coverage for these services?
- b. Should insurance companies cover testing in non-enrolled family members?
- c. Should Dr. Brown recommend that Jennifer or her aunt avoid using insurance for counseling or testing because of concerns for potential discrimination (increased rates, etc.)?

12. The high cost of testing and follow-up services raises concerns about inequitable access to services. Studies have demonstrated that a significant number of individuals who are good candidates for BRCA mutation testing forego the test due to cost issues. This sets up a situation where individuals with resources have the opportunity to benefit from genetic testing, while those without resources do not.

- a. Is this an appropriate distribution of resources? If not, how can public policy rectify this situation?

13. The use and cost of BRCA1/2 tests are controlled by the company that developed and currently holds the patents for the tests.

- a. How does patenting affect patient access to services?
- b. Should there be limits on the patenting of genes and genetic tests? Why or why not?

What genetic services delivery issues does this scenario raise?

- Lack of trained genetics professionals in the community
- Lack of organized referral or consultation networks (or lack of awareness of such resources)

What provider issues are identified?

- Criteria for use of genetic tests
- Educational resources: where to go for credible information, what information to collect
- Effective/efficient methods to identify high-risk individuals
- Risk interpretation tools
- Risk communication
- Evidence-based guidelines

What consumer issues are identified?

- Impact of media on patient requests
- Potential pre-vivor phenomenon
- Potential for discrimination (a real or perceived concern?)
- Getting information and sharing it with family members
- Cost sharing and out-of-pocket expenses

What payer or coverage issues are identified?

- Lack of reimbursement for time spent researching issues
- Variable coverage of preventive services, including genetic testing and prophylactic treatments
- Potential for discrimination

What industry issues are identified?

- Gene patenting

What other policy issues are raised by this scenario?

- Regulation of retail genetic services and oversight of quality
- Impact of patenting on genetic service delivery and access