

Helping your patients to deal with a predisposition to genetic disease

CASE STUDY

After seeing a cancer genetic counselor and undergoing genetic testing, Sara has been told that she carries the *BRCA1/2* mutation—the same mutation that her mother has and that is responsible for her mother’s recently diagnosed breast cancer. Sara is 32 years old and has a 1-year-old daughter. Until a few weeks ago, she thought that finding good childcare as she prepared to go back to work was her biggest problem. Now, Sara is feeling overwhelmed and very anxious.

Sara’s mother is scheduling bilateral mastectomies based on her own result, and she is asking Sara to take her to appointments. Sara is making appointments for herself to talk to oncologists, surgeons, and plastic surgeons as she wonders if she should have more children sooner than planned so she can have her ovaries removed by age 35 or 40 years to reduce her risk of ovarian cancer. She wonders too if she should herself consider prophylactic mastectomies.

Sara is preoccupied with worry that she could get sick and die and not be able to take care of Mimi, her daughter, or future children. She isn’t sleeping well. She also worries about Mimi and wishes she could get her daughter tested to see if she passed on this “hor-

rible gene” (as she thinks about it) to Mimi. Her husband thinks Sara is getting ahead of herself but doesn’t know what to do to make her feel better. Sara sees her PA as part of a regularly scheduled visit and tells him what is happening in her family. After talking with Sara, the PA suggests that she could benefit from talking with a psychologist to try to sort out the many layers of feelings she is having so she can feel better and make the most appropriate decisions for her.

DISCUSSION

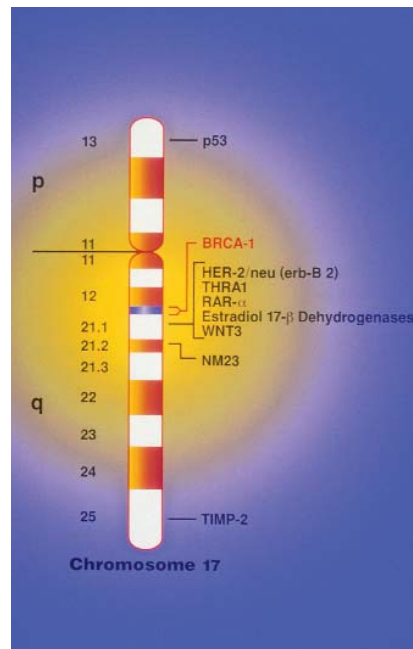
When patients receive information about hereditary disease risks, PAs should consider the psychological as well as the medical implications of the information:

- Focus on the aspect of hereditary disease predisposition that most troubles your patient. Is the patient most worried about his own risks, or are the

risks faced by parents, grandparents, or children (born or hoped for) more a concern?

- Don’t assume that patients have heard and understood what a geneticist has told them about their disease risks, risks of family members, or the risk-reduction options available. Many patients say that after a genetic counselor told them that their genetic test result was positive, they heard nothing more.
- Help patients to express their concerns, and be aware that early, upsetting memories can be powerful images that can significantly color important decisions. Help the patient to understand that the disfiguring radical mastectomy her grandmother had 40 years ago is not what she will face, should she get breast cancer. Help her to remember that medical treatments improve with time. Help parents to remember that if their child inherits the genetic predisposition, the child may have very different decisions to make than those currently faced by mutation carriers.
- Help patients to see the difference between learning that they have a genetic predisposition to disease and receiving a diagnosis of illness. Except for rare cases, such as Huntington disease, being a mutation carrier does not mean that the disease will develop for certain; some mutation carriers never become sick. The odds increase for a carrier, but not to 100%. While uncertainty is not easy to embrace, patients should be aware that knowing they have a predisposition does not tell them if they will get the disease, when they will get it, or in some cases even which of several diseases they might get.
- Remind patients that testing offers opportunities to better understand

Andrea Farkas Patenaude is a clinical and research psychologist at the Dana-Farber Cancer Institute in Boston, Massachusetts, and Associate Professor of Psychology, Department of Psychiatry, Harvard Medical School. She sees patients and family members experiencing distress in the context of undergoing cancer genetic testing in the Cancer Risk and Prevention Clinic at Dana-Farber and has run support groups for mutation carriers. She also conducts research on the psychological impact of cancer genetic testing and is the author of *Genetic Testing for Cancer: Psychological Approaches for Helping Patients and Families*. The author has indicated no relationships to disclose relating to the content of this article.



BRCA1 protein structure

their predisposition and can potentially help them to prevent or detect illness at the earliest possible stage, giving them options that earlier generations did not have. Although “knowledge is power,” remain aware too that we do not currently gain as much power as we might hope to prevent most forms of hereditary cancer or other hereditary disease.

- Patients differ in how they feel about receiving medical information and in how much detail they want about their risks. Some people cannot get enough information and are called *monitors*: people who take in a great deal of medical information and remain vigilant, sometimes hyper-vigilant, for signs of illness.¹ Others, called *blunters*, prefer to know only what is crucial for them to act on and tend to play down, sometimes to the point of not taking preventive or risk-reducing actions, the implications of the news they have received. You can help to balance these tendencies in patients. You can assist monitors to feel less anxious and to sort out what they can do now regarding risk reduction and what can be tabled for discussion at a future point. You can counsel blunters to make a plan about when risk-reduction health behaviors should be implemented and how often they need to be repeated, and you can remind them of when actions should be taken.
- PAs can also play an important role by watching to see if the news of hereditary predisposition is having a major adverse emotional effect on the patient. If it is, referral to a psychologist, psychiatrist, or social worker (preferably one familiar with the relevant illness) might help the patient to better cope with the news. For many people, genetic testing, coming as it does at the intersection of illness and family concerns, brings

out unresolved grief (common in families with many generations of hereditary disease because of the frequency of losses), conflicts, or feelings related to how family members have handled previous illness or loss. Issues may also arise involving communication among family members about health-related matters, feelings about the “fairness” or “unfairness” of life, religious doubts, and feelings about interaction with medical professionals.^{2,3} These responses typical-

“Patients who test negative for a mutation known to affect their family may have difficult emotions to deal with as well.”

ly are better dealt with in a psychotherapeutic setting because they may take time to work through, especially for patients who themselves face a high risk of illness.

- Finally, remember that family members who test negative for a mutation known to affect the family may have difficult emotions to deal with as well. Some feel “left out” of the feeling that family members are “in it together” as they face hereditary cancer or other hereditary illnesses. Many feel guilty about being happy that they have been spared and, particularly, that they will not have to worry about passing on an illness predisposition to future generations. They may feel that they have to hide their relief from those who are at risk or that they should take on additional family responsibilities to make up for their good fortune. Sometimes referral of sister pairs or mother-daughter or father-daughter pairs for psychological sessions can re-open lines of communication that may have been blocked by genetic test results in families where some mem-

bers have positive and some have negative results. These families actually have more difficult times emotionally than families in which all tested relatives have the same results.

CONCLUSION

Genetic tests are increasingly being used for multiple medical purposes,⁴ which makes it increasingly important to have practitioners who can interpret the tests for patients, help them make use of the information they receive,

and, perhaps most importantly, help them to live with the feelings aroused by their knowledge and what it means for both the past and future of their family. PAs who understand both the medical and psychological implications of genetic information⁵ will have critical roles to play in helping to integrate this important new area of medical practice into the lives of those who are the first to reap its benefits and experience its impact. **JAAPA**

Michael A. Rackover, PA-C, MS; Constance Goldgar, MS, PA-C, department editors

REFERENCES

1. Miller SM, Fang CY, Manne SL, et al. Decision making about prophylactic oophorectomy among at-risk women: psychological influence and implications. *Gynecol Oncol*. 1999; 75(3):406-412.
2. Patenaude AF, Dorval M, DiGianni L, et al. Sharing BRCA1/2 results with first-degree relatives: factors predicting whom women tell. *J Clin Oncol*. 2006;24(4):700-706.
3. Tercyak KP, Peshkin BN, Demarco TA, et al. Information needs of mothers regarding communicating BRCA1/2 cancer genetic test results to their children. *Genet Test*. 2007;11(3): 249-255.
4. Patenaude AF, Guttmacher AE, Collins FS. Genetic testing and psychology: new roles, new responsibilities. *Amer Psychol*. 2002;57(4):271-282.
5. Patenaude AF. *Genetic Testing for Cancer: Psychological Approaches for Helping Patients and Families*. Washington, DC: American Psychological Association; 2005.

Tell us what you think! Go to www.jaapa.com to comment on this article.