Healthcare Communication Review
Volume 3 Number 1 Online Edition January 2003

Thinking About Genetic Testing for Breast Cancer

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Introduction

Genes and genetics, especially as they relate to personal and family health, are very much in the news these days. Despite all the hype and sensationalism of human cloning and designer genes, the real potential benefits in terms of understanding and ultimately treating human disease, while still years away, can't be underestimated. In many ways, we are truly at the threshold of a new era of medicine. In a few instances, however, the future is already here. One example is the discovery, in the last decade, of several specific, altered genes in individuals from families where there was a very high incidence of both breast and ovarian cancer. Women inheriting such an altered gene are at increased risk of developing these cancers during their lifetime.

The identification of these cancer-risk genes opened the door to the possibility of offering genetic testing to identify their presence in other women who had a significant family history of cancer. Whether a particular woman does or does not carry one of these genes can greatly affect the medical management of her cancer risk. The process leading to genetic testing can be complicated, however, and the final decision can be based as much on personal reasons as on medical ones. What I will discuss here is how one might approach deciding if this is something to pursue and what is involved in deciding if it is appropriate for particular women at increased risk for developing breast cancer.

The Relationship Between Genes and Breast Cancer

As many as one-in-three Americans will develop some sort of cancer during their lifetimes. Breast cancer is the #1 cancer among women. Approximately 12% of women will develop breast cancer during their lives. Its cause, like most cancer, is far from clear. Although there is firm evidence of complex interactions among genetic and environmental factors, all cancer at some level is genetic in origin. The basic definition of cancer is that of cells growing out of control. Cell growth is the most basic aspect of any living organism. This growth is intricately regulated and monitored by many cellular factors that interact with each other and with environmental factors. In the same way that genes contain the information encoding traits like eye color or blood type, genes also contain information encoding the many molecular factors that regulate and control the cycle of cell growth. Changes in this genetic material can then tip cell-growth regulation out of control.

In thinking about the genetic causes of cancer, it is important to understand that 'genetic' is not always the same as 'inherited'. In fact, only around 5 – 10% of all cancer is inherited, meaning that it can be directly attributed to a genetic factor passed on from a parent. In contrast, most cancer, **by far**, arises from changes that occur in individual body

cells over the course of a person's lifetime. If that change occurs in a cell that is part of breast tissue, for example, and the changes affect genes that regulate breast cell growth, then that cell may start a process of cancerous growth.

Since most breast cancer is not inherited, genetic testing is really not appropriate for the vast majority of women. For which women, then, might it be considered? A key to determining whether a woman is likely to have an inherited breast cancer gene is the close examination of her family history of cancer. Because cancer is so common, most people do, or will, have some cancer in their family. Whether or not that history is suggestive of an inherited cancer-risk gene depends on factors including the number of family members affected with the cancer and how closely related they are, as well as the types of cancer and whether they occurred early or late in life.

Making a Decision About Genetic Testing

If you are concerned about your own breast cancer risk, discuss it with your physician if you haven't already done so. When you do, you can also ask whether he or she thinks genetic testing is appropriate – and it may well be. Because genetics is a specialized and constantly changing medical field, some physicians may be more knowledgeable than others. You can also speak with a genetic counselor. These are individuals who are specifically trained to assess inherited cancer risk and to help you determine if testing is the right thing for you.

Genetic testing is not for everyone, even where there is a significant history of cancer. The process of genetic counseling involves reviewing and discussing the many aspects of the decision process and making sure the decision to proceed, or not, is the best one. For example, as a genetic counselor, I want to make sure that the patient fully understands the benefits, limitations, and any risks involved in DNA testing. I want to know that she is as clear as possible about her reasons for wanting (or ultimately not wanting) to pursue testing at this time.

In making her decision, it is also important to clarify her cancer risk. What exactly is her risk of developing cancer? Is this more or less than she thought? What decisions will she and her doctor make about medical management based on the test result, if at all? How will she and her family be affected by the testing process and by either a positive or negative test result? Informed testing should include all these aspects. It should also address any concerns there might be about privacy and insurance issues. Sometimes these types of personal aspects end up being most important, rather than any medical answers about the value of the test. Every woman is different and being referred for genetic counseling does not necessarily mean that she will end up being tested.

While the decision to be tested is not always straight forward, the actual test process is relatively simple. A blood sample is drawn and then sent to a commercial lab that does the actual gene analysis. Once sent, the process takes about two weeks and the results are reported directly to the physician ordering the test. The doctor or genetic counselor can then go over the results with you and review what it means for you and how to proceed next.

In the end, the result of any genetic testing is just another piece of information. The importance of this information will lie in it's meaning to you and in the decision it will lead you to make. Because each person is unique, that information will hold a different meaning for one woman and her family than it will for others. These are the issues that also should be addressed when asking yourself or your doctor if genetic testing is right for you. □

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