

PERSONAL ACCESS PROGRAM OF HIGH LAKES



BREAST CANCER, THE HUMAN GENOME AND YOU

As you all know, one of my main professional objectives is to find and stop common, deadly diseases whenever possible. Yet the screening tests we employ to find hidden medical problems are far from perfect, sometimes yielding mistaken results (called “false positive” or “false negative” results.) So we must always approach a screening test with a rational, well-considered plan that will prevent us from actually creating more problems through things like unnecessary surgical biopsies or additional invasive screening tests, often the result of indiscriminant testing. This underscores why we don’t blindly screen every person for every disease with our imperfect battery of available tests.

The effective use of screening tests often relies on our ability to select those individuals in a population who have a greater-than-average risk of developing a particular illness and will therefore benefit from a more aggressive screening plan. This process is known as “risk stratification,” and is an essential part of many screening protocols. For example, in an earlier newsletter I shared information about young women in Norway who were at a monstrously high risk for Breast Cancer due to a couple of specific gene mutations known as either BRCA1 or BRCA2. In these

cases, it made sense to use both Mammogram and MRI of the breast every year to screen these patients. But mammogram alone made better sense for the population in general.

Now, with the completion of the human genome project, there have been dozens of commercial genetic tests marketed to the public from as many different companies. The value of most of these tests is hotly debated and their widespread use is rightly criticized in many cases. While “KNOWLEDGE IS POWER!” is the catch phrase for marketing many of these products, the results often lead to more confusion than enlightenment. For unless a test result can clearly dictate a useful, potentially lifesaving course of action for the patient who is found to carry a particular mutation, he or she will experience little value, but possibly plenty of undue stress and anxiety.

So it’s a tough task to convince me to share a genetic test with my patients and readers. Yet I’m going to do so now.

We all hate breast cancer. It is the most common deadly cancer in women world wide and American women suffer from three times the world’s average breast cancer rate. You’ve probably heard me say that the one thing I want my patients to be paranoid about is any breast abnormality they may suspect. That’s because early detection clearly saves lives. So anything we can do to effectively identify real breast

Breast Cancer Genes (continued)

cancers as early as possible makes a lot of sense.

A new genetic screening test known as the **deCode Breast Cancer™** test, offers some real value to all women whose ancestors originated in Europe. (The test has yet to be definitively proven in African American, Asian or Native American populations.) This is how it works:

We can save more lives by identifying women who have an increased likelihood of developing breast cancer and then screen them more closely with MRI of the breast in addition to conventional mammogram. We can also use this risk information to select the women likely to develop “estrogen positive” breast cancer and then treat them with preventive medication such as Tamoxifen. The American Cancer Society currently recommends MRI screening for all women known to have a lifetime-risk of breast cancer over 20%. The American Society of Clinical Oncology recommends preventive medical therapy for women with a 5-year risk of breast cancer greater than 1.66%.

So where do you or your loved ones fall in the scheme of things?

Well, an American woman has a 12.3% chance of developing breast cancer in her lifetime. This number represents the average, combined risk for all women. Yet we know that some individuals have a much higher risk than this because they have an alarming number of close relatives who already developed the disease. In these cases we are often dealing with the well-know breast cancer/ovarian cancer genes BRCA1 or BRCA2. Fortunately, these are relatively rare mutations and account for less than 5% of all breast cancers. These patients should

certainly consider MRI screening and perhaps cancer-prevention treatment. Happily, many of these patients are now being effectively identified and treated appropriately. But they represent a very small minority.

That leaves everybody else. How can we better refine our knowledge of an individual’s personal breast cancer risk? It turns out that there are seven genetic areas that can now be tested in each person, which have been proven to accurately “fine tune” the calculated breast cancer risk for that person. By using a simple swab of the inside of her cheek, a woman can now know her genetic standing in these seven areas which will tell her with much greater accuracy her lifetime risk for breast cancer. The results can range from a risk of under 5% to as high as 50%. The test also predicts the likelihood of developing the kind of breast cancer that can be prevented with medical treatment.

The information obtained from decode Breast Cancer™ test does not replace the risk information a person may have obtained from other sources such as the Gail Breast Cancer Risk Assessment Tool

(<http://www.cancer.gov/bcrisktool/>)

or the results of genetic testing for the more aggressive BRCA1 and BRCA2 mutations. Rather, since it is looking at unrelated sources of risk, it adds to currently available information.

The cost? As of this writing, the current price for this test is \$625 and there is a 10% cash discount (\$563). Many insurances pay for some of the cost.

This letter has been just a brief overview of this new technology and its place in the larger world of breast cancer screening and risk evaluation. I invite you visit the Web site of DeCode Genetics at

<http://www.decodediagnostics.com/BC-general.php>.
