Ask the Doctor December 2015

Should every woman undergo genetic testing?

It was just over 2 years ago when Angelina Jolie was diagnosed as a carrier of a high-risk genetic mutation (BRCA1). This mutation put her at an 85% lifetime risk of developing breast cancer. At the time of her diagnosis genetic testing was limited to women like herself who had strong family histories of breast or ovarian cancer. The cost of genetic testing was in the range of four thousand dollars. Insurance companies only covered the cost of testing in high-risk women based primarily on their family history.[1] This practice of only covering the cost of testing for high-risk women continues today.

Competition in the market place has been driving down the cost of testing ever since the Supreme Court's decision that companies could not hold a patent on naturally isolated DNA.[2]

Recently 17 new mutations have been identified that are associated with a high-risk of developing breast cancer. One new company is likely to turn the world of genetic testing upside down. The company, Color Genomics, offers BRCA1 and BRCA 2 testing plus testing of the 17 other high-risk mutations at a cost of 249 dollars.

In addition no referral is required. A woman can go directly to their web site and review videos discussing the pros and cons of genetic testing. If she chooses to participate, she can be referred to a physician on the web site or she can have her own doctor make the referral.

Women who chose to participate send in the payment and in a few days a kit arrives in the mail. A web site video explains how to provide a saliva sample. Once completed, the kit is returned to Color for analysis. Results are available in a few weeks. Genetic counselors are available on-line to discuss results.

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I am convinced this new approach to gene testing will soon revolutionize the world of genetic testing. Cost is no longer a barrier. The question who should be tested?

If you listen to Mary-Clair King, (who discovered the BRCA1 gene mutation) you are likely to conclude every woman should at least be given the option of testing by age 30. Dr. King points out that women who have no family history can be carriers of high-risk mutations, although she readily admits this is a rare phenomenon.

By identify silent carriers when they are young two important interventions are possible. The first option is to start breast MRI screening at age 25, thus improving a high-risk woman's potential for early detection. The second option is to do prophylactic mastectomies as did Angelina Jolie. This more aggressive option will prevent a future breast cancer in the vast majority of women who select this procedure.

There are many critics of a web-based approach to genetic screening. Experts point out that we are in a steep learning curve when it comes to understanding the complexity of the genetic basis of hereditary breast cancer. They are concerned that wide spread testing will cause unnecessary anxiety in the vast majority of women in order to detect the small fraction of women who are silent carriers.

We have had several months of experience with Color's web-based approach to genetic testing and are very impressed with the results. We find that most of our cancer patients who did not meet standard criteria for genetic testing are anxious to participate. Their major reason for testing is not so much for their own care. These women are concerned about the potential for passing a high-risk mutation to their children.

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Of course, most of these women will prove to be negative which is a big relief to the rest of the family. In the unusual case in which the woman turns out to be a carrier, her children are strongly advised to undergo genetic testing.

We have not yet recommended genetic screening all of our patients, but as cost of testing continues to fall, it seems inevitable that more and more women will decide they want to know if they carry a high-risk gene mutation.

Reference:

1. NCCN guidelines: https://demystifyingmedicine.od.nih.gov/DM10/0413-BreastCancer/NCCN%20br%20genetics_screening.pdf.

2. Supreme Court's decision on DNA patents: http://www.medscape.com/viewarticle/805756

3. Link Color Genomics' website: https://getcolor.com/?gclid=CPDf67CNtskCFcVffgodIDoAVg

The End.