Genetic/Genomic Tests Inform Breast Cancer Treatment Decisions

By Ruth Kava — October 25, 2016

Breast cancer is one of the most intensively investigated forms of cancer. While the research has not yet resulted in a cure, it has informed decisions about how to treat its various forms. Not only are there various types of chemotherapy for treatment, but also effective drugs (e.g. aromatase inhibitors and SERMS) to delay or prevent recurrence. There are also, of course, genetic tests that can help women determine whether they are at increased risk of developing breast cancer, and also genomic tests to indicate which treatments should be undergone.

Women who carry the BRCA-1 or BRCA-2 mutations are now known to be at higher risk of developing both breast and ovarian cancer. Knowledge of these mutations, coupled with a family history of breast cancer may induce a woman to undergo prophylactic bilateral mastectomies as well as removal of her ovaries, especially if she has already has an instance of invasive breast cancer.
Who should undergo testing for these mutations? Women who have an Ashkenazi Jewish background are at increased risk of BRCA mutations. African-American women are at increased risk of having a so-called 'triple negative' breast cancer (ones that have no receptors for estrogen, progesterone, or HER2), which are associated with the BRCA mutations. There are as yet no targeted treatments for triple-negative [2] tumors as there are for hormone receptor-positive tumors such as receptor blockers, although research is ongoing. These tumors are usually responsive to chemotherapy, but are more aggressive than hormone-sensitive tumors and have a greater risk of recurrence. Such genetic tests examine whether or not the actual chromosome is altered by mutation — a gene or genes might be missing or have an altered structure that would affect its activity.

There are now also genomic tests for breast cancer, in particular the oncotype Dx [3] test for estrogen receptor-positive tumors. This test analyzes the activity of genes in the tumor itself. It can predict the odds of tumor recurrence after treatment, whether hormonal treatment should be sufficient or if chemotherapy in addition would be advisable.

Thus, while we still have no actual cure for breast cancer, we do have tests that can help predict its occurrence or recurrence. Indeed, such testing must be at least partly responsible for the decrease in breast cancer deaths — according to the CDC [4] between 2003 and 2012, breast cancer mortality decreased significantly by nearly 2 percent per year. While we still have a long way to go before we can consider breast cancer conquered, the strides we have made thus far should give women reason to be optimistic.