Counsyl Finds Many Women Who Meet Cancer Screening Guidelines Not Getting Tested

Mar 22, 2016 | Monica Heger

Premium

NEW YORK (GenomeWeb) – Following a three-month program where Counsyl offered free inherited cancer screening to women in the San Francisco Bay Area, the firm found that a surprisingly large number of women who met traditional guidelines for testing based on family history had not previously been tested.

Over 2,500 women in the San Francisco Bay Area took advantage of Counsyl's Get Ahead of Cancer Initiative last fall, where it offered its 22-gene hereditary cancer panel for free, regardless of a woman's risk status.

As GenomeWeb reported in December, a preliminary analysis of the approximately 1,500 women who had participated in the program at that point revealed that almost half of those that tested positive for inherited cancer risk did not meet the National Comprehensive Cancer Network's guidelines for screening.

This finding held true in the larger cohort, but the firm also found very different mutational profiles between women who met NCCN testing guidelines and those who did not.

In addition, the firm was surprised by the number of women who had a strong family history of cancer and met the NCCN's testing guidelines, but had not previously been tested, Kaylene Ready, director of inherited cancer at Counsyl, told GenomeWeb.

Of the 2,596 women who received the free testing, 108, or 4.2 percent, had a positive test result. Those 108 women, as well as 195 randomly selected women who tested negative, were evaluated for family history. The researchers estimated that approximately 46 percent of these 303 women met NCCN's family history criteria for cancer screening.

Ready said that the firm would be more closely investigating why that group of women had not

previously been tested. "Either they didn't realize they were at risk, or they realized it but maybe had been told that the cost of testing was really high," she said.

A study reported in *JAMA Oncology* last month suggests that young women are getting such tests, but possibly only after they have been diagnosed with cancer. That study found that around 87 percent of 897 women who had been diagnosed with cancer at age 40 or younger had received testing for BRCA1 and BRCA2 mutations within one year of their diagnosis.

As expected, Ready said many of the women who opted for the free test had some family history of cancer, but that history was not strong enough to meet NCCN's guidelines for testing. That group of women represented around 29 percent of the total. Of the 108 positive test results, 37 were in women whose family history was not strong enough to meet NCCN criteria.

"When we see individuals who have BRCA mutations or Lynch syndrome mutations but fall short of meeting testing guidelines, that does speak to a need to broaden guidelines," Ready said.

The firm even found cancer risk mutations in the group of women with no family history, but those findings were much more rare. About 25 percent of the women who chose to get tested were at low risk, and 14 out of 108, or 13 percent, of the positive results were in low-risk women.

Interestingly, the frequency of positive tests was similar between the two groups of women with a family history of cancer, with 4.8 percent of the highest risk group and 4.9 percent of the intermediate risk group having a positive result.

However, even though overall frequencies of positive tests were similar between the two groups, when the Counsyl team dug deeper into the findings, they noted that the types of positive results differed significantly.

For instance, the highest-risk group of women had a statistically significant higher frequency of well-understood penetrant alleles, including in the BRCA1, BRCA2, and Lynch syndrome genes. BRCA1 and BRCA2 were the most commonly mutated genes in this group, with 14 and 16 positive results, respectively.

Meanwhile, the group that fell short of meeting the NCCN testing guidelines had a statistically significant higher frequency of a mutation in the APC gene, but not of other penetrant alleles. The APC mutation has been linked to a two-fold increased risk for colon cancer in the Ashkenazi Jewish populations, but is not as well understood in the general population. Additionally, in this group, just two women had BRCA2 mutations and none had a BRCA1 mutation.

Ready said that further studies on the APC variant and its link to cancer would need to be done in the general population. However, she said the firm is "continuing to dig into that result to make sure there's not a population bias."

The project also uncovered a few unexpected findings. Ready highlighted one case in particular of a woman who had no family history risk for cancer and was seeing a doctor for *in vitro* fertilization treatment. She decided to take advantage of the free cancer testing and found out she was positive for a PMS2 mutation, a Lynch syndrome gene that confers elevated risk for colorectal cancer. Based on

that information, she postponed her embryo transfer and first had a colonoscopy. In addition, her partner also got tested. Examples such as that help make the case for broader population-based screening.

One aspect of the project that Ready said the firm is continuing to evaluate is the impact of releasing results and offering post-test counseling online. After testing, results were made available online for both the ordering physician and patient. Patients were able to access those results online as well as to schedule online post-test counseling within two hours of receiving their results, which "reduces anxiety that patients face when they have to wait" days or weeks.

"The ability to allow patients to access results online and access to genetic counseling online was key to being able to do this program," Ready said. She noted that about 20 percent of patients who were positive for a BRCA mutation pursued follow-up genetic counseling.

The company is also evaluating the data to see what other conclusions can be drawn. Ready noted that the firm is also considering expanding the program to other cities, which would give them an even larger and more diverse set of data. Repeating the program would also give them the opportunity to improve on it, she said. For the San Francisco program, she said the company did not survey the patients about their motivations for getting tested, but in the future, she said, that is something the firm would be interested in doing.

Counsyl has also not systematically tracked and followed patients after they've been tested aside from anecdotally through talking with their physicians, she said. The firm has the ability to do that through its online interface, but Ready said that the company is trying to figure out the best balance between surveying patients with research questions and respecting their privacy.

Filed Under Sequencing Molecular Diagnostics Cancer Clinical Sequencing breast cancer NGS Counsyl

Related Articles

Dec 01, 2015

Counsyl Free Cancer Screening Finds Many At-Risk Women Who Would Not Have Been Tested

May 25, 2016

Counsyl Launches Oncology Business Unit

Jul 26, 2016

NorthShore University HealthSystem to Launch Genomics-Based Cancer Risk Testing Trial

Sep 30, 2015

Companies Begin Subsidizing Color Genomics' Breast, Ovarian Cancer Testing for Employees

Oct 01, 2015

Study of Aetna-Insured Finds Lack of Pre-BRCA Test Counseling Despite Guidelines

Oct 26, 2015

UC Launches Framingham-Like Study to Inform Personalized Breast Cancer Screening Guidelines