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The Negative Outcomes of a Positive Result

New genetic tests may provide more stress than information for patients



by Maggie Masterson

The rise of genetic testing has led to thousands of women undergoing BRCA testing in the past 15 years, but novel gene mutations are leading to a more expansive and more controversial test. BRCA is a gene that protects us from developing tumors, so a mutation on BRCA implicates a high risk of cancer, but it is not the only gene linked to hereditary cancers. Women are now opting to undergo panel testing, which involves not only testing for a mutation on

the well-known and extensively understood BRCA gene, but also on newly discovered genes, about which geneticists are still at a knowledge deficit, as reported in *Reuters*.



A mutation in either the BRCA 1 or BRCA 2 gene will greatly increase a person's lifetime risk of breast and ovarian cancer. The lifetime risk of developing breast cancer for someone with a BRCA mutation is up to 87 percent, and up to 44 percent for ovarian cancer. Women are the highest risk group for these cancers, since they have more breast tissue than men, and because only females possess ovaries. For this reason, it is mainly women who opt for BRCA testing, although men can also be screened to assess their status as a carrier of the gene. However,

there are women developing hereditary breast cancers who test negative for a BRCA mutation, indicating that there are other genes linked to these hereditary cancers. This is where genetic panel testing comes into play.

“The catch with these novel genes is that their expression and relative risk to certain cancers are mostly unknown.”

A genetic panel test differs from a normal genetic test in that it will screen many genes rather than just targeting one. For example, an Ambry Genetics BreastNext panel test will screen for a mutation in BRCA 1 or 2, as well as on fifteen other genes linked to breast cancer, such as PALB2 and TP53. A test that provides more genetic information should lead to better medical management and risk assessment, right? Well, the catch with these novel genes is that their expression and relative risk to certain cancers are mostly unknown. A woman who would have previously breathed a sigh of relief over a negative BRCA result may now be burdened with the information that she has a mutation on a gene such as TP53. Genetic counselors cannot articulate the exact risks that accompany such mutations and, more alarmingly, cannot confidently say that a mutation in one of these genes is malignant. The genes in question are so new to us that testing for them may cause more harm than good.

We are continually gathering more information about these new genes, which is one benefit of these controversial panel tests: they are slowly helping geneticists learn more about these novel genes. But for now many geneticists are calling for a moratorium on these tests, until more clinically relevant information is available. As time passes, it will become more clear if genetic paneling is helping or hurting patients. 🐾