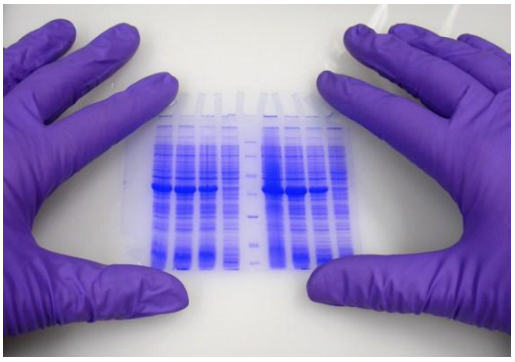

Genetics Corner

Decisions, Decisions;

How to decide what to do next when a BRCA gene mutation is discovered

Cancer risk management guidelines for women with BRCA1 or BRCA2 gene mutation as outlined by the NCCN guide-lines have been previously discussed. However, these guide-lines present women with several options that are not always easy to choose from. This article highlights a variety of re-sources available to women with BRCA gene mutations who are going through the decision-making process about how to manage their cancer risk.

Several Options

Women with BRCA gene mutations must eventually choose how to manage their risk from many options available to them. For breast cancer risk management, one category of options involves managing risk through breast conservation and taking steps to better detect breast cancer if/when it does occur (e.g. Tamoxifen plus annual MRI). Alternatively, women may decide to remove their breasts to reduce their risk as much as possible. Timing can complicate these options further. For example, some women may know that they will want a mastectomy eventually, but they are not sure at what age. Re-garding ovarian cancer risk management, for example, a woman that is not yet finished with childbearing can have a difficult time deciding when (and if) she should have her ova-ries removed.

The Stanford Tool: Detailed Risk by Management Choices

Many of our patients with BRCA gene mutations find that knowing their exact numerical risk helps them to decide which steps to take. Researchers with Stanford School of Medicine have created a tool that patients and providers can use to determine a patient's cancer and mortality risks based on her age and the combination of management options she chooses from. 2 One of the major benefits of the tool is that patients can compare one set of risks based on their possible choices to risks from another set of possible choices. For example, this tool shows that a 25-29 year old woman with a BRCA1 mutation who chooses to have mammograms and MRIs through the age of 70 has an 11% chance of dying from breast can-cer (if she does not have an oophorectomy). Using this tool, she can see that if she instead decided to have a prophylactic mastectomy at 40, she would have a 7% lifetime risk of dying from breast cancer. If she wanted to also know by how much her risk would be reduced if she were to have a bilateral mastectomy at age 30, she could see that her risk would be further reduced to 2%.

This tool also incorporates the impact of oophorectomy on ovarian cancer risk. For example, if the same hypothetical woman were to never have an oophorectomy, her lifetime risk of passing away from ovarian cancer would be 18%. If she instead has an oophorectomy at 35, her risk would be reduced to 4%. The oth-er risks this tool can generate include lifetime risk of being alive with ovarian cancer and breast cancer, life-time risk of living with breast cancer, lifetime risk of never having breast or ovarian cancer, and the impact of oophorectomy on breast cancer risk. The results are displayed both numerically with percentages and visually with colored bar graphs. The user can also compare one set of hypothetical risks side-by-side with several other selections of hypothetical risks. Women can compare these risks to the risks of women who have not had BRCA gene mutations.

The ability to compare an individual's risk both numerically and visually can help to improve patients' under-standing of their risks. However, there are a few of downsides to this tool. The tool is not entirely intuitive at first glance, so some patients may use it incorrectly. It also generates a lot of numbers at once,

which each have very different meanings. Some patients may con-fuse these numbers. Anyone using it must also have access to the internet. For these reasons, we do not recommend this tool for everyone, and also give pa-tients risks during their results sessions based on the literature. For example, based on Chen and Parmigiani's research, we can let a patient in her 40s with a BRCA2 mutation know that she has about a 42% lifetime breast cancer risk and 16% lifetime ovarian cancer risk.[1] However, precisely determining by how much these risks change based on management choices is not always possible.

Health Care Specialists Help Clarify Personalized Management Options

Another important step in making decisions about cancer risk management is speaking with specialists about procedures. All women with BRCA gene mutations should speak with a breast specialist, preferably a breast surgeon, in addition to a gynecological oncologist. Clarifying to patients the details of management options can help to dispel myths, and thus help to prevent patients from making decisions they may otherwise later regret. For example, many women avoid mastectomies because they know of women who have had poor aesthetic out-comes from the mastectomies they had had decades prior. Breast surgeons can explain verbally and through the use of visual aids that mastectomy and reconstruction has improved markedly over the past couple of decades. They may also discuss with women that their options are more limited. For exam-ple, if a woman has already had breast cancer that has spread to the other breast, a breast surgeon may strongly encourage a double mastectomy.

Patients Learn from Others' Experiences

Speaking with others who have been through the procedures already can further help patients in the decision-making process. Patient perspectives can come from in-person and online support groups and literature. Support groups include nation-wide support groups, such as FORCE and Bright Pink. FORCE has Dallas and Fort Worth chapters, and the Fort Worth chapter meets at Moncrief Cancer Institute. There are other local groups, such as 'Young Survivors of Tarrant County'. Several sup-port groups meet here at 'Women's Journey of Hope' group. There are several books for patients that detail the journeys women take upon discovering that they have a BRCA gene mutation. These include Previvors, by Dina Roth Port, and Pretty is What Changes, by Jessica Queller. The former gives several women's viewpoints and experiences in the decision-making process presented with published management options. The latter is a more detailed account of one woman's experiences. Learning more about others' experiences who have already gone through and experienced these procedures can be very valuable.

Other Assistance in Decision-Making

There are other resources available to women who could use some assistance with the decision-making process. For example, some women feel completely overwhelmed upon recently dis-covering they have BRCA gene mutations. Being presented with their management options may further compound the problem. Sometimes these women often benefit from individual psychological counseling. Also, some patients find help from religious institutions. Furthermore, friends and family members who have had experience with the management options can be an important source of information for patients. It is also important for patients to discuss how these decisions may have an impact on family members and friends.

Most women benefit from information that comes from a combination of sources. When a patient tested through our institution tests positive for a BRCA gene mutation, he/she is given a list of resources to help guide them through the decision-making process, including those outlined above. For some women, knowing the exact risks numerically and graphically (i.e. using the Stanford tool) is important for their decision-making process. For others, the exact risk is less important. Instead, gathering information about the details of risk reduction procedures may be more important. We do our best to point out to our patients which resources they would get the most benefit from. Fortunately, there are many sources of

information available to patients with BRCA gene mutations who are going through the decision-making process.

As always, if you have any questions or would like to refer a patient to UT Southwestern's cancer genetics team in Dallas or with Moncrief Cancer Institute in Fort Worth, please call (214) 645-2563.

References

Chen S, Parmigiani G. Meta-analysis of BRCA1 and BRCA2 penetrance. J Clin Oncol. 2007 Apr 10;25(11):1329-33.

Stanford Medicine. Decision Tool for Women with BRCA Mutations. http://brcatool.stanford.edu. Last updated 12/01/2011. Accessed 1/17/2013.