GENETIC TESTING AND CANCER RISK (BRCA1 and BRCA2)

To test or not to test? That is the question.

The advancement and wizardry in medical technology and the availability of genetic testing is manna from heaven. However, with testing of any kind, whether genetic or otherwise, comes uncertainty with the results. This uncertainty comes with a partner in crime…fear. This fear is what makes testing a double edged sword. For women, this is the sword of Damocles…they have the power to test or not to test knowing that the results may be life changing. The idea that this wicked disease called cancer could be their reality is terrifying. The option of genetic testing is a weight that has been dropped squarely on the delicate shoulders of women.

According to research by the National Institute of Health, BRCA1, which was discovered about 20 years ago and BRCA2 later, can now be tested for mutations, which can lead to cancer. We should be singing Hallelujah! And indeed we are singing! However, whenever there is uncertainty, fear rears its ugly head. With this manna from heaven come great responsibility and the daunting question…to test or not to test?

What are BRCA1 and BRCA2 anyway? Simply put, BRCA1 and BRCA2 are human genes that produce tumor suppressor proteins. These proteins assist in fixing damages in the DNA thereby managing the stability of the cell's genetic material. In other words, if these genes (BRCA1 and BRCA2) are okay, they produce the proteins needed for repairs and upgrades to the DNA. However, if either of these genes, BRCA1 or BRCA2, is mutated or defective, the tumor suppressor proteins that are produced are affected. The genes can either produce no protein at all or produce protein that is non functional. As a result, DNA damage is not repaired and this is what leads to cells developing additional genetic mutations that can cause cancer. According to the National Institute of Health (NIH) and the National Cancer Institute (NCI), specific inherited mutations in BRCA1 and BRCA2 increase the risk of female breast and ovarian cancers. These mutations account for about 20 to 25 percent of hereditary breast cancers and about 5 to 10 percent of all breast cancers. The mutations according to NIH, also account for 15 percent of ovarian cancers. As if this is not bad enough, breast and ovarian cancers associated with these mutations tend to start at a younger age than their nonhereditary counterparts. The silver lining in all of this is that according to research, the harmful BRCA1 and BRCA2 gene mutations are relatively rare in the general population. Now, for that small population in harm's way, the question of testing looms larger than life because the management of cancer risk after a positive diagnosis presents new challenges in choosing the "right" options available to them. This is serious business. One of the options available is no walk in the park and that is the Prophylactic (Risk-reducing) Surgery, which involves the removal of the breasts, ovaries and fallopian tubes. I do not know of any woman who would not be absolutely terrified of losing her breasts, ovaries and fallopian tubes whether by choice or by necessity. The other options are less terrifying. There is the enhanced and more frequent screenings starting at age 25 to 35 and the chemoprevention option, which involves the use of drugs, vitamins and other agents to lower or reduce the risk of cancer.

The "butcher knife option" is the treatment that puts fear in woman about testing. As a result, there is a divide when it comes to testing.

The truth is that there are three categories of women when it comes to the question of testing for cancer risk. There is your "**Knowledge is power**" category. This group of women wants to be tested and they want to be in control of anything pertaining to them and their health. Testing puts them in a position of power over their bodies and destinies. They are proactive. They are ready for battle. They want to be in a position of kicking cancer in the rear! This group wants to be in the know. What do they think about the stress of testing? Do they consider a positive result for the gene mutation a death sentence or do they consider it an opportunity to cheat death? You bet they believe it is an opportunity to send death packing on a long, long vacation away from their lives…at least for the time being. Do they weigh quality versus quantity of life? I believe this group does and I think their choice would be living longer (quantity) in the absence of disease

and managing the quality of life the best way they can. This is especially true for women who choose the prophylactic risk-reducing surgery. Imagine opting to undergo surgery to remove breasts and ovaries that are disease free for the moment pending when or if cancer strikes. Removing all the lady bits surely comes with some degree of psychological stress to say the least.

Then there is the group I refer to as the "**Ignorance is bliss**" group. This group has their head in the sand and they pretend that cancer cannot happen to them and so there is no need for such lunacy as testing. They want to live with the notion that whatever will be, will be. They prefer not knowing the odds against them. They hold on to the idea that not everyone who tests positive for the gene mutation is going to end up with cancer. Given that the percentage for cancer in people with the mutation is not one hundred percent, they believe they will rather take their chances and hope for the best. This group is of the mindset that if the chances of getting cancer still exists even after all the risk-reducing treatments, then testing is not worth it. The principle in operation here is, if one tests positive for the gene mutation, they have to go through treatment like a cancer patient and so why not wait and live in bliss until one gets cancer and then begins the tough procedure of mastectomies and salpingo-oophorectomies. The only difference is the word "prophylactic". Do you choose when to get risk-reducing treatment or does cancer choose for you?

The last group I call the "**Should I, should I not?**" kind of people. Women in this category are petrified. They believe they are doomed if they test and doomed if they don't. Any additional piece of information they receive regarding testing makes them more skeptical. They cannot make up their minds because they are afraid their decision to test or not test might be costly emotionally and physically. Remaining in limbo buys them time to ignore reality. It should be noted that mutation testing is only for those who do not have cancer but their family history suggests the presence of the harmful mutation in BRCA1 or BRCA2. So, if relatives are dying of the same type of cancers and you do not have cancer, but cannot help wondering if the same fate awaits you, then testing is an option for you.

What group do you belong to when it comes to testing of any kind that pertains to your health? Are you in the "Knowledge is power" group? Do you research information to the point where you are more knowledgeable than your physician? Are you consumed with the need to know and does the information you gather relieve your fears? Does information empower you? Most importantly, are you able to make a decision based on the information you receive? If you answered yes to any of these questions, then you should consider testing.

Are you in the "Ignorance is bliss" category where you do little or no research? Does the little information you obtain scare you? Does information cripple you to the point that you avoid it altogether? Do you prefer to wait for doomsday as opposed to searching for it and halting it? If you answered yes, then maybe you should consider not testing. It might help your case to know that a true negative test result is only a clean bill of health as it pertains to the mutation gene but not really a clean bill of health for your risk of developing cancer.

For the "Should I, should I not?" people, try to focus on the benefits of testing and of not testing. Which scenario has more benefits as it pertains to you and to people in your life? If testing has more benefits, that should be your choice. If "not testing" has more benefits, then that's the answer to your question…to test or not to test?

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