

BRCA Genetic Testing: Understanding the Physical, Emotional and Financial Challenges

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David: Good evening, everyone. Welcome to today's Sharsheret National Teleconference. At this time, all participants are in listen-only mode. Later, you will have the opportunity to ask questions during the question-and-answer session. You may register to ask a question at any time by pressing the star and one on your touch-tone phone. Please note that this call may be recorded and I will be standing by should you need any assistance. It is now my pleasure to turn today's conference over to your moderator, Ms. Shera Dubitsky. Please go ahead.

I. Introduction

Shera Dubitsky: Great. Thank you, David. Good evening to everyone. Welcome to Sharsheret's National Teleconference, BRCA Genetic Testing: Understanding the Physical, Emotional, and Financial Challenges. My name is Shera Dubitsky, and I am the Director of Navigation and Support Services at Sharsheret. I will be moderating the webinar for this evening and also presenting a little later on. We would like to thank Medivation, Myriad, and the Cooperative Agreement DP14-1408 from the Centers for Disease Control and Prevention for their ongoing support and for sponsoring tonight's program.

Sharsheret is a national not-for-profit organization supporting young Jewish women and their families facing breast cancer. Our mission is to offer a community of support to women of all Jewish backgrounds, diagnosed with breast cancer or at an increased risk, by fostering culturally-relevant individualized connections with networks of peers, health professionals, and related resources. Although our expertise is in young women and Jewish women, we do offer support to anybody who reaches out to us.

As you may know, those of Ashkenazi Jewish descent, meaning from Eastern European Jewish descent, are 10 times more likely to carry the BRCA mutation than those in the general population. As a result of this reality, there is an increased discussion about population-based testing for those of Ashkenazi Jewish descent for the BRCA1 and BRCA2 founder mutations. The recommendations for population testing seemed to be gaining steam. Tonight, we are going to address the benefits and challenges of population testing from both the physical and emotional perspective.

I would like to introduce our first presenter, Peggy Cottrell. She is a graduate of the Sarah Lawrence College Master of Science and Genetic Counseling Program. At Sharsheret, Peggy consults with women and families, and answers individual questions about family histories, the BRCA mutations, and personal risks of hereditary breast cancer and ovarian cancer, and contributes to the development and implementation

of Sharsheret's hereditary cancer resources and programs. Peggy, the floor is yours.

II. Physical Considerations

Peggy Cottrell: Thank you so much, Shera. What exactly is population screening? Population screening means that instead of trying to figure out who is at high risk based on their personal family history of cancer, we would test people just because they have Ashkenazi ancestry. Why might we do that? Because, as Shera said, one in 40 Ashkenazi Jewish will carry a mutation in one of these BRCA1 or BRCA2 genes, and that is men and women. As it turns out, it's not always easy to identify the high-risk families. If we could always count on family history, this would be an easier process.

A couple of years ago, this discussion was first begun in Israel. A study was published, which you can see at the top of here today. In this study, they were trying to figure out, would it actually be beneficial to do population screening? Our concern was, could it be that people who didn't have a family history but carried the BRCA1 or 2 gene actually had a much lower risk to develop cancer because of something different about them, their genetics or their lifestyle. What this study did is gathered a group of people by a method of population screening, and then determined that, in fact, these individuals had a pretty significant chance to develop cancer than if they did not have the strong family history that went along with it.

All around the world, different countries have been looking at this. This was a study published in Canada. Scientists and physicians in Canada seemed to think, perhaps, it is time to consider population screening for Jewish women.

Here in the United States, Dr. Mary-Claire King, who happens to be the woman who discovered the BRCA1 gene almost 20 years ago, proposed, I would say, a couple of years ago, that not only should we consider population screening for Ashkenazi Jewish women but, perhaps, we should consider population screening for all young women in the United States for BRCA mutations.

Now, this is a little more out there because the frequency of mutations in the general population is not as high as one in 40, it's more like one in 300 or 400. The cost of testing in the general population is significantly higher than screening in the Ashkenazi population because we're able to screen for just the mutations that are common among Ashkenazi Jewish. Her proposal has been a little bit more controversial than the proposal to

screen any Ashkenazi community, but has given steam to the proposal in the Ashkenazi community.

This summer, Sharsheret was invited to participate in a research symposium in Israel, and our own Executive Director, Elana Silber, attended to share the patient perspective on the day of population screening. This is an issue that's been greatly discussed, and so we wanted to bring that issue to you tonight.

Right now, if I were to see a patient who had Ashkenazi ancestry, but had no personal or family history of cancer, and I submitted their testing to a lab, this is probably what would happen, denied. That's because right now, insurance guidelines do not allow for population screening here in the United States. What has to change, the way that insurance companies decide what they're going to cover and what they are not, is based on guidelines that come from national organizations. Right now, those national organizations are not saying that population screening is something that's important enough for them, at least, to be saying that insurance companies should pay for it.

One of the problems that we mentioned earlier is that an incomplete family history can sometimes lead to being unable to identify those families who were at high risk. It happens very often when I'm seeing women that come to see me because they are the first person in their family to be diagnosed with cancer, and that cancer may be coming at a particularly young age, and we test them because of that young age of onset, and we're finding something inherited, without any evidence in the family tree.

There are a number of reasons why this can happen. The first reason on the list is one that particularly applies to Ashkenazi Jews and that's the Holocaust. Sometimes, in taking a family tree, when we get back to the generation that was alive during the war, there are numerous people who were killed and murdered, and didn't have the opportunity to live a long life, and have the possibility of developing cancer. Those missing family members can leave a blank, and makes it more difficult to be able to identify something inherited.

Another thing that happens is that families are small. Sometimes, people have only a few siblings. Sometimes, they only have one aunt or one uncle. A small family can make it more difficult to see the evidence of the inherited mutation. Sometimes, families are out of touch. Families may be out of touch in today's society because of increased mobility. In the old days, people tend to stay in similar neighborhoods with their families, and with their children and parents. Nowadays, people take jobs across the country, and they may not be as close they once were, and may not share all the information about their history.

Also, sometimes, people are private. Even though they may see their family members and talk to their family members on a regular basis, they may not want to share the fact that they've been diagnosed with cancer for one reason or another, and this can also prevent women from knowing about the family history that could impact them.

You may belong to a family that has a preponderance of male family members. You may have a big family, but it may be filled with uncles and brothers as opposed to aunts and sisters. Because when men carry a mutation in BRCA1 or 2, they are significantly less likely than women to develop cancer, and this can have an impact of what we see in the family tree. Finally, along the similar lines, paternal inheritance. If you inherit your BRCA mutation from your father's father's father, that is, again, reducing the chance that you'll see evidence in the family tree.

What are the advantages of population screening? Screening more women gives us the opportunity to find more women who are at high risk. Even screening women, studies have shown screening women who have no family history or personal history, we're still going to find about 2% of the time that people carry one of these inherited mutations. When we identify these women, we can offer them our particularly unique medical management that's different from the ordinary screening.

Now, sometimes, women will say to me, "Peggy, I already go and get my mammograms every year. I get a sonogram. I get a breast exam from my doctor. Why do I really need to know about this inherited mutation?" The reason is that we have a higher level of enhanced screening and the opportunity for risk-reducing surgery, and studies have shown that these options save and extend lives.

The biggest difference in high risk screening for breast cancer screening is the addition of an annual breast MRI. A breast MRI is not something that's available to most women for screening. It's available particularly to women who are at high risk to develop breast cancer, and breast MRIs are much more sensitive than mammograms and sonograms. It looks at the breast in a very different way. We're not saying someone should have an MRI, and they don't need the other screening, but each one of these tools has its own way of looking at the breast, and a combination of all of them together can significantly increase the chance that we'll be able to identify breast cancer early in someone at high risk.

This high-risk screening for ovarian cancer includes a pelvic exam, and a transvaginal sonogram, and a blood test for a marker called CA 125, but these screening tools have never been proven to increase survival from ovarian cancer. They do not help us find the ovarian cancer at an early enough stage so that we can increase survival, and that's the way in

which high-risk screening for ovarian cancer is very different from high-risk screening for breast cancer.

A second option for women to consider if they test positive is risk-reducing surgery. For the breast, this is having a bilateral mastectomy before the diagnosis with cancer, and this procedure often includes breast reconstruction afterwards. A woman who has the breast removed significantly reduces her chance of developing breast cancer.

Likewise, for the ovarian cancer risk, we perform what's called a bilateral salpingo-oophorectomy. That's a mouthful, but it means we're taking out the tubes, that's the salpingo part, and the ovaries, the oophorectomy. Some women may also consider having a hysterectomy because of a very small chance for BRCA1 carriers to develop a more aggressive type of uterine cancer. This second procedure, again, significantly reduces the chance of a woman ever developing ovarian cancer. We think that a fair amount of ovarian cancer starts in the tubes and that's why it's always important to remove the tubes along with the ovary.

What are some of the disadvantages? The first one you can see right away based on the picture on this is financial. That's because individuals who don't have that personal or family history of breast, ovarian, or pancreatic cancer won't meet insurance guidelines for testing. Then, the cost of their genetic counseling and testing may have to be paid out of pocket. Most of the time, that's going to be several hundred dollars, but could possibly be several thousand dollars depending on the type of testing that's going to be done. If women, in mass, are going to choose population screening, there will need to be some financial support whether that will come from insurance at some point, from the women themselves, or perhaps from philanthropic sources. It's unclear at this point.

Sometimes when I talk with women about testing and what their options are to consider, I wish that I had a crystal ball, and I could examine each individual, and tell them, "I can see in the future, and I know that you will diagnosed with breast cancer when you're 45. A good time to have a mastectomy would be at 40, and you'll be able to avoid this." Unfortunately, that is a pipe dream. Whenever I talk to women, we have to think about the possibility that since the risk of cancer with a BRCA mutation is not 100%, somebody may end up having prophylactic surgery even though they were never going to get cancer.

The other issue is the age of onset is not predictable. Just because people in your family seemed to get breast cancer in their 40s, then you can wait, and you will also get the breast cancer in your 40s. That's not true. It could come younger. It could come older. Many times, women feel like they're gambling trying to make these decisions, trying to predict what

the odds are of what's going to happen and when it could happen. While, sometimes, we know that we save women's lives by doing the surgery, other times, it's possible that we're just causing them a lot of side effects and not necessarily a lot of benefit.

What are some of the surgical complications that can go along with having a bilateral mastectomy? A bilateral mastectomy is a pretty significant surgery, and most women are going to take weeks or months to recover, especially if they have reconstruction, which most women will want to have. Different kinds of reconstruction can take different amounts of time in terms of recovery. Not every reconstruction is completed at the time of the mastectomy. Often, there are many ongoing steps that need to continue to be taken.

Reconstructed breasts often look fabulous once they're healed, but there's a lot of disruption to sensation as a result of surgery. Nipple reconstruction often involves tattooing, and some women find this problematic because of Jewish law against tattooing. Rarely, even though someone has had a bilateral mastectomy, they could still develop breast cancer.

A bunch of complications from having the ovaries and tubes removed. The surgery to remove ovaries and tubes is much more straightforward than a mastectomy, and many women are able to have the surgery done on an outpatient basis and don't even have to stay overnight in a hospital, but there are significant side effects to the surgery, and the main one is immediate menopause. As soon as we removed someone's ovaries, they will go through menopause, and this is a big downside of the surgery.

Now, guidelines suggest that for carriers of the BRCA1 mutation, the ovaries be removed sometime between 35 and 40, and for those of the BRCA2 mutation, sometime between the ages of 40 and 45. Sometimes, people don't know that you can carry this mutation until they're much older. Then, we would remove the ovaries sometime shortly after we find out that someone is a carrier.

There are risks to early menopause. There's loss of fertility, which goes along with the loss of the ovaries, even with hormone replacement, which may be available to some women who have surgery. These may include vaginal dryness, loss of sex drive, and osteoporosis. Even more concerning to many women are studies that seem to possibly link neurologic or cardiovascular risks to early menopause. While these studies are not conclusive, they are very frightening to women who have to think about these procedures. Finally, while it's rare, one may develop something called primary peritoneal cancer. It is similar to ovarian cancer and can occur rarely even after prophylactic surgery.

Many women are concerned about the loss of fertility that can go along with being a BRCA carrier. Fertility can be lost, as we just mentioned, as a result of the removal of the ovaries and tubes. Sometimes in BRCA1 carriers, that's happening at a young age when women might still be wanting to have children. Sometimes, a loss of fertility comes with the diagnosis with cancer. Being pregnant with cancer is a serious complication, and doctors strongly recommend against pregnancy while being treated because the treatments can have a serious effect on the fetus. Also, chemotherapy treatment can even lead to the loss of ovarian function.

Finally, studies seem to suggest that the BRCA1 and 2 mutations themselves may also be a cause of some women to have what's called premature ovarian failure. Scientists are not exactly sure how this results but it may be that there are fewer eggs available. Someone with a BRCA mutation may go through menopause earlier even if though they don't have their ovaries removed.

Very often, when talking about population screening, we think of the population screening that we do on some level, which is the carrier screening that many couples consider before they get married or before they have children looking for disorders that are recessive like Tay-Sachs.

It seems like it might be a simple solution to this question to just tuck on another genetic test to the ones that are already being done, which might not significantly increase the cost. The problem is that a BRCA carrier has so much more of the significant impact on one's own health than being the carrier, which has no implications for one's own health but only for future offspring. Healthcare providers really believe that carrier testing for BRCA1 and 2 requires a significantly different kind of informed consent before someone would be able to undergo that test.

What do we do? Here at Sharsheret, we want to you know, first of all, that you're not alone. I am here as the Genetics for Life Program, and I'm available to speak with you one-on-one. If you have specific questions about your own history, your own cancer risk, and the implication of genetic testing in your family, you can set up an appointment with me and we can go through these issues.

We also offer free family conference calls, which allow you to communicate with other members of the family that you'd like to discuss testing with who may not be close by. Finally, we have our materials, our booklet, Your Jewish Genes is available to discuss all of these issues as well. Shera.

Shera Dubitsky: Peggy, I want to thank you for your expertise and your insight and shedding light on a very complicated issue.

III. Emotional Considerations

I'd like to reintroduce myself. I am Shera Dubitsky, Director of Navigation and Support Services at Sharsheret. I graduated with two master's degrees in Psychology from Columbia University and earned a third master's degree and completed clinical work toward a Doctorate in Clinical Psychology from Adelphi University Institute of Advanced Psychological Studies. I support and connect newly diagnosed young women and those at high risk of developing breast cancer or ovarian cancer with suitable peer supporters, advanced and develop programs addressing the unique needs of young women and families of Sharsheret, and counsel individual members of the Embrace Program for women living with advanced breast or ovarian cancer.

Using Peggy's very informative presentation as a springboard, I will now explore some of the psychological implications of population testing and genetic testing overall. We have increased calls from women who were curious about their ancestry who took the over-the-counter kit that included a test to discover what their DNA says about their health, traits and ancestry.

One particular call though stands out for me because I think it captures the range of issues and experiences of this model of genetic testing. Debra, which is not her real name, 28 years old, she shared that last year, she received a Hanukkah gift from their friend. The gift was this kit that enabled her to test her DNA to learn more about her ancestry. She excitedly submits her saliva sample, and mails it to the suggested lab, and she received her results.

She remembered sitting with her friends perusing the results, and this is how she described the moment, "We were looking through the results and laughing ourselves silly with interest. Learning so much about my traits and ancestry was completely fascinating. We clicked through so many pages, really in the spirit of just laughing and with interest. I quickly glanced over at the page that read that I carried the BRCA mutation. I didn't have a family history so I didn't pay much attention to this.

October, this year, during Breast Cancer Awareness Month, I was watching a morning news show, and they were interviewing a woman who tested BRCA-positive, and underwent a prophylactic bilateral mastectomy. And I said, 'What? Why would she do that without having cancer?' I immediately made an appointment with my doctor and my world starts to spin out of control."

Debra had many decisions to make, including should she share this information with her only sibling, which was a sister. She decided that she had to share the information with her, and this ended up causing tremendous tension in the relationship. Since then, they have both contacted Sharsheret and received support and resources from this crazy journey.

There are families like Debra's that don't have a known family history. I emphasized "known" because there may be several explanations as to why there is no known family history. The most obvious explanation is that there is, in fact, no family history of cancer, particularly breast cancer or ovarian cancer. Peggy pointed out that families who lost members in the holocaust also won't know their complete histories.

Another reason why people don't have a known family history is because discussing cancer and particularly a woman's cancer was taboo a generation or two ago. The word "cancer" was whispered in corners of the family home. We have asked women to go back, find grandma or the great-aunt's death certificate, and let's see what they died of. When they call us back, they read that grandma's cause of death was a woman's disease. We suspect that it as breast cancer, ovarian cancer, cervical cancer, or uterine cancer. We just don't know which one for sure.

There are certain communities, also currently, who either don't undergo genetic testing or they don't share the results of the genetic testing because they are worried that there will be a taint on unmarried family members, and they may be seen as unsuitable for marriage. As a result, subsequent generations will report no known family history.

Let's first focus on families where there is a known family history. There is a range of emotion that is experienced by each individual. For some women, there is a sense of premonition going into the testing. They know that there is either cancer or a known genetic mutation in the family. Hearing a BRCA-positive result is upsetting, yet, in many cases, expected.

Some women even report feeling relieved because they have been anxiously anticipating testing. Now, they have a clear answer, and can go about planning next steps, and perhaps pursue options that a family member affected by cancer did not have. These women, who have either suffered a death of a loved one from breast cancer or ovarian cancer or watched a loved one undergo chemotherapy and other treatments for cancer, may sooner opt to undergo prophylactic surgery because they don't want to experience what they have seen their loved one go through.

For those women with a family history, testing BRCA-negative is murkier. If the family member diagnosed with cancer tested BRCA-positive and the

caller tested BRCA-negative, she actually can feel relieved because she is a true BRCA-negative and her risk is comparable to those in the general population.

For family members who have a strong family history of breast and/or ovarian cancer, and everyone is testing BRCA-negative, the likelihood is that there is a genetic explanation, but just not on the BRCA gene. Many of these families are now going back and testing other genes. This in itself can raise anxiety and stress because there are no clear answers, and decision making becomes more complicated.

You can also be a person like me where my mom passed away from breast cancer before genetic testing was even an option. I went ahead and did testing, and I tested BRCA-negative, but I can't completely exhale because I don't know if I'm a true BRCA-negative or if I should be maybe concerned about another genetic mutation, just not on the BRCA gene.

Now, let's focus on those individuals who have no known family history and undergo genetic testing. There was a study that was published in the Journal for Genetics and Medicine, and it was performed by the Program for Jewish Genetic Health at the Albert Einstein College of Medicine. The study explored experiences from a prior program bringing BRCA1 and 2 genetic screening to the US Ashkenazi Jewish population.

This study showed that low-risk participants, meaning individuals with no known significant family histories who opted to undergo genetic testing, chose to participate in the study because they planned on using the information and results to better manage their healthcare, to inform their family member of their results, and simply because they had an Ashkenazi Jewish background.

The top fears of testing positive were, obviously, increased cancer risk and also insurance discrimination. Upon them getting their results, those low-risk individuals testing BRCA-positive expressed anger regarding their decision to pursue testing to begin with, and even some disbelief in their results. Many expressed feeling shocked and upset. Some women reported feeling even pressured into surgical procedures by physicians.

It is important to note that not all individuals who carry the BRCA mutation will be diagnosed with cancer. As Peggy pointed out, without a known family history, a BRCA-positive result is confusing and stressful because decision making is murkier. Some Sharsheret callers who used the over-the-counter testing kit and test BRCA-positive have also reported lack of support from family and friends who thinks that it's just ludicrous to remove currently healthy parts of your body regardless of the results from genetic testing. By the way, this is also true for women who do have a family history and test BRCA-positive. They are calling Sharsheret

because they may not be getting support from friends or families about their decisions whether it's to continue high surveillance or to undergo prophylactic surgery.

One last thing with the low-risk individual who tests BRCA-negative, there also may be a false sense of security because there may still be a genetic mutation on another gene and this false sense of security may lead to not scheduling routine mammograms or performing self-breast exams.

Both low-risk and high-risk individuals undergoing genetic testing face some difficult decisions. With the BRCA-positive results, are the next steps to follow increased high surveillance or undergo prophylactic surgery, including, again as Peggy mentioned, prophylactic bilateral mastectomy and/or oophorectomy, removal of the ovaries, or hysterectomy? Many factors have to be considered including intent to have children, breastfeeding, early menopause, and sexuality and intimacy with a partner or a spouse.

This decision can put a strain on a relationship depending on how supportive the spouse or partner is about the decision to undergo prophylactic surgery and the impact on him or her. Based on the calls that we are receiving here at Sharsheret, it seems that those women who don't have a known family history and test BRCA-positive are more hesitant about undergoing prophylactic surgery because they really don't have a reference point. This information is random and decisions feel more elusive and intangible.

For those women who do have a family history and test BRCA-positive, decisions, though difficult, may be approached with more clarity. For those women who have a known family history, and everyone is testing BRCA-negative, what are her options? Should she continue surveillance or undergo surgery? This ambiguity can be anxiety-producing as well. A conversation with her doctor who knows all her unique variables is certainly strongly suggested.

Jewish law may also play a role in decision making. We get calls from women who ask, "Well, is it permissible under Jewish law to undergo genetic testing? Is it permissible to undergo prophylactic surgery? What about reconstruction, is that permissible? Also, we know that it's not permissible under Jewish law to have tattoos. Yet, some reconstruction may include nipple and areola tattooing. Is this permissible?" Again, we encourage women to speak with their Rabbis or other experts who understand the implications of genetic testing under Jewish law.

Whenever we go to a school, a synagogue, or other institutions, we often see a lost-and-found box. It rarely, if ever, says just "lost." In most

instances, it includes "and found." I like this idea because in life, we often experience loss but it's also important to focus on the found as well.

Let's start with the loss. Many Sharsheret callers share that the loss of their breast feels overwhelming for many reasons. The actual loss of breast changes a woman's self-image, her self-perception, and in some cases, her self-esteem. The scars, whether slight or noticeable, are a constant reminder for positive or negative associations with this decision.

Loss of breast also means that women who choose to have children after surgery no longer have the ability to breastfeed, and this can be very upsetting, and they have to look for alternative ways to bond with their children. This also raises fear that the child may not get the physical and healthy benefits that breastfeeding offers.

There's also lack of sensation in the breast after surgery and reconstruction, and this can impact intimacy with her and her partner. It's important for them to communicate openly and explore additional alternative ways to achieve sexual pleasure.

Another loss connected to an oophorectomy or hysterectomy include, as Peggy again said, fertility choices. Specifically, women may have to have children earlier or sooner than planned prior to undergoing the surgery. You can imagine how stressful this is and the impact to some long-term equations such as having several young children in a shorter time span, which in itself, can be very challenging.

Also, once the decision to undergo the surgery has been made, options to have children naturally are no longer available, which can also result in some sadness or grief. Many callers struggle with this and have ambivalent feelings about adoption or surrogacy.

Another result of an oophorectomy or hysterectomy, again as Peggy mentioned, is an early onset menopause. One woman shared that she experiences hot flashes and mood swings, and she can't discuss it with her friends because this is not what they are currently experiencing. She said that her new peer group are women in her mother's age group. She may also experience vaginal dryness or decreased libido, which has an impact on her sexual and intimate experience with her spouse or partner. I once had one woman say that her libido is in the freezer and she doesn't even feel like taking it out at all. This clearly impacts her relationship with her partner and potentially causes increased tension.

Another loss is a sense of security when it comes to the future health of her children. Knowing that she has genetic mutation places a burden on her. Did she pass the mutation on to her children? When should she be

discussing this with them? When should they be tested? This is a loss of innocence for them because of what it may mean to the children later on.

As I said earlier, I also think there are some founds in these circumstances. Knowledge of the BRCA-positive mutation gives women options that were not available to their loved ones. A woman may have more control over her health and choices and timing of her choices. There's also relief that there are interventions that are available to a woman that significantly lowers her risk of ever being diagnosed with breast cancer or ovarian cancer. I once had a woman jokingly share that she had a boob job and a tummy tuck for a copay of five dollars.

There are options to women where they can test these fertilized embryos, as we previously discussed, and only implant those that are BRCA-negative. We have one woman who was diagnosed with breast cancer, had a strong family history of breast cancer, and had her embryos tested, and she shared with overwhelming relief that she broke the chain in her family. I think another found is inner strength that a woman might not know that she had prior to facing these difficult decisions. I think that this is particularly empowering.

Communicating results for family members can be tricky. Things to consider are age of family members, closeness and relationship with family members, the emotional well-being of a family member. Meaning, is there a predisposition to anxiety, for example, and how will they receive the information. As I mentioned earlier, there are some communities that worry if there is a genetic mutation or cancer in the family, it will impact the non-married members of the family. Should she share the information? If so, how will it be received? What will they do with the information? All of this is a tremendous burden on the woman and these are the things that Sharsheret can help you with.

How can we help? In addition to the services we offer through the Genetics for Life Program that Peggy outlined, we can connect you with a peer supporter who had to face similar news and decisions. They can share their experience and help you think out loud about decisions that you may be facing. These conversations are often validating, informative, and uplifting. Our clinical staff can also connect you with additional information and resources that are personalized to your unique needs.

It is now with great pleasure and privilege to introduce Miryam. She is what we call a seasoned peer supporter for Sharsheret and has spoken to many callers over the years. She will share her personal story and offer insights for navigating the emotional impact of testing positive for the BRCA mutation. Miryam.

IV. Personal Story

Miryam: Hi. Thank you so much, Shera. There's been so many things that came to my mind while you were speaking, while Peggy was speaking, but I just want to start by saying that I'm here to speak about my emotional experience having learned that I was BRCA-positive. As with emotions, those emotions that we've got are very highly specific, and they're going to be based on your own personal history, and your own experiences, and your own chemical makeup.

I'm going to take you through my own, the chronology of my emotional rollercoaster that I got to ride as of 2009. That was when I first learned that I was BRCA-positive. What prompted me to get tested was, I guess, based on having heard as I heard from you guys is that I was lucky that I had a family history so there wasn't any doubt. I had lost my mother when I was 14. I'm one of five children. Four of us are girls. Two of my sisters tested positive for the BRCA mutation.

Back in 2009, I was 29 years old, and I had learned that my sister, who had undergone the prophylactic bilateral mastectomy, found that she had a very early stage of cancer that might not have been detected through a mammogram or through an MRI. I had been married for a year and a half. Throughout my life, having lost my mother at a young age, I never really looked at breast cancer as a hereditary thing, which is strange, but maybe that was my denial. Maybe that was how I knew that I was going to be okay and I wasn't going to lose my life at a young age.

By the time my sister went through what she had gone through, and she had a very practical approach to our family history, she encouraged me to get tested as well. I believe that Shera touched on that where it can build family tension. There was definitely a part of me that was rolling my eyes, "We can't control our future. We don't know what's going to be. Why get tested?" but I went ahead and did it, and I knew that I was probably going to be positive because I was told that I was 50% likely to be positive. I joked that, "Well, two out of four sisters are positive. So, maybe I'll be lucky and not be positive," but that's not how statistics work.

When I learned that I was positive, it didn't come as a surprise to me, and it didn't really affect me at any way at that moment. The genetic counselor explains the options to me. She described my situation as being 85% likely to get a very, I guess, a bad, fatal form of cancer. Then, that if I were to do surgery, it could bring me down to 15%. At the same time, she acknowledged that she can't predict my future and it really gave me something to think about.

That was when things started to really tailspin. I did the genetic testing through Memorial Sloan Kettering, and because of my family history,

insurance covered it, and I was guided through the entire process. After learning about my BRCA-positive circumstance, I was brought to a psychiatrist that works within Memorial Sloan Kettering.

The hardest part for me about all of it was making a decision. The reason it was so difficult is because I had to ask myself, "Here I am, married, no children. I plan on having children. Am I about to undergo a surgery, a pretty intense surgery, a pretty dramatic surgery because I might or might not get breast cancer?" I understood from the geneticist that my high risk would range from between ages of 30 and 40. She showed me a mountain in how my risk would peak in my mid-30s. Then, it would start to go down until I'm 40. Here I was, 29, I wanted to have children, I thought I wanted to breastfeed my children, and I didn't want to put myself at a disadvantage.

Really, the hard part was doctors are asking me if I was concerned about losing sensation, if I was concerned about how that would affect my relationship with my husband, who by the way was very supportive, and that made this entire experience a lot easier. It all boiled down to, after not being able to eat, not being able to sleep, constantly crying, did I want to just rush and have children right away, breastfeed them, and then do the surgery? How many kids can I cram into the next how many years? It was really difficult.

I turned to as many people as I could for answers. Each one, I thought I could trust. My mother-in-law, just she didn't know what to say. This isn't anything that she never really lived with. She said, "You have to do what you think is right," and I didn't know what I thought was right. I didn't have the answers. The psychiatrist couldn't help me. My husband couldn't answer this question for me. My sister couldn't answer this question for me.

I finally spoke with, actually, a local Rabbi in my community who knew my parents and who knew me. I really thought that he was going to tell me, "Don't worry about it. God has it under control. Have your children. Breastfeed them. Have a normal life, and don't worry about the science schmience," but that's not what happened.

What he, actually, said to me was, "I know your family and you have a really bad history of cancer, and you sound like you're really distressed. And the bottom line is if you're 85% likely to get breast cancer, why wouldn't you just do the surgery? You don't need to breastfeed your children. I think that by doing the surgery, you're going to separate yourself from a very painful past and you're going to put yourself back in the driver's seat, and you're going to get to decide when you want to have your first child, and then your next child, and then your next child. And you're not going to feel like the clock is running against you."

It was basically that week that I went to speak with a plastic surgeon who I consulted with prior. I remember very clearly, it was a Wednesday, and we scheduled the surgery for the next Tuesday because I just wanted to get it done. It was after that Wednesday that I scheduled the surgery that I was able to finally sleep at night, and I felt great. I was just happy that I made the decision. The anxiety was gone because, for me, that was the difficult part.

Just to be perfectly honest, and I can't say that this is the same for everybody, but the rest was a breeze. Surgery was surgery. I was sleeping. The doctors did the work. The aftermath, percocet can work miracles if you combine it with colace. How did my breasts look? They don't look as great as they did before but I imagine that after breastfeeding children, they would also not look as great as before, and that's carried me through.

After all that, we went on, my husband and I. We now have four children. We have a six-year-old, a four-and-a-half-year-old, a three-year-old, and a one-year-old. None of them are breastfed. They're all amazing and bright, and chunky and healthy. Not only do I have the privilege of speaking with other women who are faced with this decision, and my thoughts with these women is just, "If you can't sleep at night or if you can't live with this knowledge, then just get it done."

I remember a woman who she had decided to schedule a surgery for March time, and this was already September. She was panicking and crying, and panicking and crying. I just asked her, "Why are you waiting? Why are you waiting so long? Once you've decided to do it, just get it done." She went ahead and did it in November, and she said, "I just want to let you know that I'm so happy that I got this out of the way. I knew that I wanted to get it done. Why did I have to have it head hanging over my head for so long?"

It's definitely not for everyone. A question that often comes up is, "Is it hard to not breastfeed?" Logistically speaking, I'd say it's much easier to not breastfeed, but there's definitely something that I feel like I'm missing out on, not with my first three children, to be honest. I was fine with it. I actually felt proud that I did something that was considered to be bold and brave.

I almost found it a little bit comical. Everybody goes through what they go through, but many friends of mine who are breastfeeding would beat themselves up because they couldn't breastfeed or they wanted to, but felt guilty because they weren't doing enough and how long should they do it for." I wasn't part of that discussion because it wasn't an option for me, but I was able to tell them, "I can't tell you what to do, but I could tell

you that my kids are okay, and I love them very much, and I think they love me very much. So, don't beat yourself up."

Today, I could tell you that as far as aesthetics, that's something that I really wanted to just touch on for a minute, for a long time, it just didn't phase me. I guess, I was like in the state of mommying. My breasts looked really great in a bra, and they looked great under a shirt, but without a bra and without a shirt, they don't look amazing. I call it an angry cat because my pectoral muscles pulled back my silicone implants, and they get this like angry wrinkly. I always hiss when I try to describe what they looked like at that time.

I don't love it, but I could tell you that have I not done the surgery, I really don't think I would have been able to go on to have the first child, or the second child, or the third. I was able to raise my children or continue to raise my children with peace of mind, with a calm, with all the regular things that come with raising four small children in a Manhattan apartment, and that is where I am today.

I know that there's probably a lot that I didn't cover in my little soliloquy here but if anybody has any questions, I know that one of the questions that came up was how did I tell my family. My husband was with me when I found out. It did not come as a shock. It was pretty much expected. It was my in-laws. It was a little bit tricky for me because, again, I felt a little bit silly. "Hey, mom and dad. Um, thanks for bringing me into your family. Um, I have this genetic mutation, and, uh, I might just have to undergo a nine-hour surgery, and not be able to breastfeed my children. Ho- how do you feel about that?" Thankfully, they were very supportive also.

I just repeat that once a decision was made, the anxiety really dissipated. Here I am today. I'm pretty close to the age my mother was when she first learned that she had breast cancer, and I feel like I could just enjoy life, and it's one last thing to worry about.

Shera Dubitsky: We are very happy, Miryam, that you are in fact here today. I imagine that many people listening tonight were nodding their heads in unison as they hear your story. I think that your words were very validating, and that I also think when you said that your life went on, it was probably very uplifting to hear. Thank you very much. We will now begin our question and answer period. The operator, David, can you please instruct the callers on how to ask questions.

V. Question and Answer

David: Of course, Ms. Dubitsky. If you'd like to ask a question, please press the star and one on your touch-tone phone. You may withdraw your question

at any time by pressing the pound key. Once again, if you'd like to ask a question today, please press the star and one on your touch-tone telephone keypad.

Shera Dubitsky: Great. Thank you. Please keep your questions broad in nature so that everyone on the call can benefit from the discussion. As you're calling in, we have already received some questions. I'm going to go ahead and throw one to you, Peggy. What type of doctor is best to follow somebody who is high risk or who is even testing positive for the BRCA mutation?

Peggy Cottrell: Somebody who's having breast cancer screening, the breast MRI, should be followed by a breast surgeon because that's the doctor who best knows how to interpret those tests and the does the breast exam. For a woman who needs ovarian cancer screening or surgery, they should follow with a gynecologic oncologist.

Shera Dubitsky: Okay. This is an interesting question. How often are genetic tests incorrect?

Peggy Cottrell: That's a good question. I think we know that there are false negatives in the sense that people test negatively, and we believe that there are other genetic factors that are increasing the risk of cancer in their family. False positives, we really know very little about. There are a small number of cases where a lab has changed their call on a certain finding. They might have called it ovarian suspected deleterious, and then change it possibly to ovarian. This is not something that happens very often. Most of the time, testing is correct.

Shera Dubitsky: Another woman asked that she has a strong family history and everybody is testing BRCA-negative, similar to what we mentioned during our presentation. She wants to know what should she and families like hers do next? What are the next steps in terms of testing?

Peggy Cottrell: If the testing was done only for BRCA1 and BRCA2, you could consider now doing a panel test, which include additional genes that may also predispose to breast and ovarian cancer. Very often, we don't find anything inherited. The important thing is to screen the family based on the family history as if there is something there because we know that a significant portion of the inherited causes of breast and ovarian cancer has not yet been identified.

Shera Dubitsky: When should a male be tested? Also, with that is any prostate cancer connected with BRCA?

Peggy Cottrell: We want women to start thinking about getting tested when they are 25, but men can wait a little bit longer. That's because the cancer risk for men don't occur at ages that are quite as young. Some men want to know if

they test positively before they have children. Yes, prostate cancer is definitely a part of BRCA1 and 2. Studies seem to show about 10% of the time when prostate cancer is at an advanced stage, we may be able to identify an inherited mutation that's causing that.

Shera Dubitsky: We have some question about insurance. What to do when insurance won't cover for genetic testing? What can they do about that? What are their options?

Peggy Cottrell: When insurance doesn't want to cover, it's important to figure out what the problem is. Sometimes, the problem is that your insurance is contracted with a different lab than one that your sample has been sent to. For example, in New Jersey, Horizon has a contract not with Myriad or Ambry but with LabCorp, and this can be a problem.

Sometimes, there's not coverage because your family history doesn't meet with guidelines for testing coverage. Then, you may have to think about the least expensive test that you may be able to pay for out of pocket. I found the best way to try to figure out these issues is to speak with a genetic counselor, have a genetic counselor take care of your genetic testing, and they can usually have the best chance of being able to send your testing to a lab that will be able to get it covered.

Shera Dubitsky: Interesting. Do most BRCA-positive cancers occur before menopause?

Peggy Cottrell: That's an interesting question. Many of the breast cancers that occur in BRCA carriers occur before menopause, but probably more of the ovarian cancer occurs after menopause. I don't know an exact number but maybe half of cancers before menopause and half of cancers after.

Shera Dubitsky: I just want to follow up on an earlier question about the insurance coverage for genetic testing. It was a question about insurance covering prophylactic surgery and reconstruction surgery, and that really depends on your insurance company. Though, I have to say that very rarely do we hear that an insurance company is not covering prophylactic surgery or reconstruction surgery when indicated. Peggy, here's another one. All four of my biological grandparents live into their 80s with no cancer, yet, I am a BRCA1 carrier. How is this hereditary cancer?

Peggy Cottrell: I don't know for sure which of those four grandparents passed along that inherited mutation, but it's very possible that was one of your grandfathers because most men were carrying the mutation through their whole life and never get cancer. Also, many women who have a BRCA1 mutation will go their whole life and never get cancer. If we say that a woman has 50 to 85% chance to develop breast cancer, that means she has somewhere between a 15 and 50% chance of never developing breast

cancer. That can happen as well. I'm not sure where your inherited mutation came from, but those are just some thoughts.

Shera Dubitsky: Actually, one came in for you, Miryam. It was a question about being in the hospital, and how was it for you when you were in the hospital, and maybe lactation consultants were coming in, or women next to you were breastfeeding, how was that experience for you?

Miryam: It's actually a really, really interesting question. With each child, it was a whole new experience, but when I had my third child, I had him at the same hospital where I have had my prophylactic bilateral mastectomy. I was lucky enough with my second child to have my own room just by chance, but in this case I was sharing a room with a young girl who she has had her first child. She was breastfeeding her child, and they brought the baby into the room with this woman.

I guess, I'm just going to be honest here and not politically correct in any way. Her mother came with her, and her mom was sitting with her in the room, and she was breastfeeding in the room. At that time, it really didn't bother me that this woman was breastfeeding and I wasn't. What actually bothered me was that I wanted to get some sleep, and this woman and her mother were going to be sitting there breastfeeding their baby, and trying to figure out, and it sounded really complicated.

I remember going to one of the nurses late at night and saying, "Hey, you know, I can't breastfeed because a doctor in this hospital actually made it so that I can't breastfeed because that was what was recommended. And there's a woman in a room with her mother and her baby, and her baby is going to be in the room all night." The nurse was really sympathetic, and she said, "I'm so sorry but our rooms are all full. And you know, it's our policy that the moms will have to keep the baby in the room with her, and, you know, her mother really shouldn't be there with her."

Then, I just broke down and started to cry. I was postpartum, and not as postpartum. I just had a baby that morning. I said, "I don't have a mother and I don't have a breast, and you're going to make me share a room with a woman who's breastfeeding her child with her mother." They moved me. She said, "Well, why don't I move you to this other room with this other woman whose child is in the, the NICU." I said, "Okay, fine. That's what we'll do."

I started working over, and it turned out it was a woman who had her music blaring, and she was hanging curtains, and she said, "Please, come in. come." I asked her, "Do you plan on going to sleep any time soon?" She said, "No, no. I'm not sleeping at all tonight." I went back to my room, and I just put my earplugs in, and I went to sleep.

The point is, emotionally at the time, it's probably bothering me, not because I felt like she was bonding with the baby and I wasn't, more because I wanted to get some rest and she wasn't allowing me too. Do I feel like sometimes it's unfair? Maybe, but more often than not, I hear stories about how difficult it is to breastfeed, and how women feel like it's very uncomfortable, and they feel very confined. I just take it with a grain of salt. That's basically it. It's not easy. It's not so much fun, but I do get this overwhelming sense of pride that, again, I did something really, really bold and brave to change the narrative of my life and make it different from my mom. That carries me through.

Shera Dubitsky: Yeah.

Miryam: Something to be proud of.

Shera Dubitsky: That's great. Yeah, I agree. Yeah, thank you for that. Peggy, if a woman already had breast cancer, should she get BRCA testing?

Peggy Cottrell: Yes. That's because a woman who already had breast cancer, depending on the way her cancer was treated, could get breast cancer a second time. If she had either with a lumpectomy with radiation, or perhaps a single mastectomy, she still has a lot of breast tissue remaining.

Then, finally, the other concern that we have is that she had breast cancer, she could still develop ovarian cancer. Finally, the woman who had breast cancer is often the best person in the family to test because she is the most likely person to have the mutation, and that can provide the most help to other family members. Should women who've already had breast cancer be tested? Absolutely, yes.

Shera Dubitsky: What surveillance should BRCA-positive men do?

Peggy Cottrell: That's a very good question. What we recommend is that men who test positive get a breast exam by their doctor. Now, that can seem a little funny to men, but men do have breasts. Whether or not they have a mammogram or not is somewhat controversial in terms of the guidelines. A breast exam is a good idea. Then, they should have prostate cancer screening probably starting around age 40 or 45. That would be a PSA and a rectal exam by their doctor to check the prostate, and that for BRCA1 carrier, that probably would suffice.

Shera Dubitsky: There are other questions that are coming in, but we'll have to continue this conversation at another time. We certainly encourage you to call the office with questions. I do want to end on this last question, Peggy. What are some long-term studies and research right now in BRCA testing and what's on the horizon?

Peggy Cottrell: I think for the long-term, women who find out that they have these inherited mutations, and make the tough choices for surveillance or for surgery survive and do very well. I think that in the future, as we find out more and more about other genes that may be contributing.

VI. Conclusion

Shera Dubitsky: You will be receiving an evaluation in your email box. Please take a few minutes to complete the survey. Your feedback is valuable to us as we are committed to staying relevant by enhancing our programs to reflect the growing and changing needs of the women and families of our Sharsheret community. Sharsheret's expertise, again, is a young women and Jewish women. Through our 12 national programs, we are open to supporting all women and men regardless of their backgrounds.

I want to thank Peggy Cottrell for her expertise. I want to encourage all of you to call Sharsheret to set up a one-to-one genetic consultation or a family conference call with her. Peggy has been invaluable to the women and families who have already reached out to her. I believe that tonight's discussion clarifies the issue surrounding population testing. We, at Sharsheret, encourage genetic counseling before genetic testing even if or when population testing becomes standard level of care. I also want to thank Miryam for sharing her story and bringing many of these issues to life.

Tonight's presentation will be available on audio or through transcript on Sharsheret's website, and you can access it by going to www.sharsheret.org/resources/transcripts. I would like to again thank Medivation, Myriad, and the Cooperative Agreement DP14-1408 from the Centers for Disease Control and Prevention for generously sponsoring tonight's teleconference, and for recognizing and supporting the needs of families who are at a higher risk of carrying the BRCA mutation.

I hope that tonight's webinar is a springboard for further discussion with your doctors and treatment teams where you can talk about your own personal circumstances. You can visit Sharsheret's website at www.sharsheret.org or give us a call at 866-474-2774 to discuss tonight's topic or any other concerns that you are facing. Finally, we want to wish all of you a Happy Hanukkah or Happy Holiday Season and a Happy New Year to all of you. Good night.

VII. Speaker Biographies

Peggy Cottrell, MS, CGC, Genetics Program Coordinator, is a graduate of the Sarah Lawrence College Master of Science in Genetic Counseling program. At Sharsheret, Peggy consults with women and families and answers individual questions about their family histories, BRCA mutations, and personal risks of hereditary breast and ovarian cancer, and contributes to the development and implementation of Sharsheret's hereditary cancer resources and programs.

Shera Dubitsky, MEd, MA, Director of Navigation and Support Service, is a graduate of Columbia University with two masters degrees in Psychology, and a third masters degree and completed clinical work toward a doctorate in clinical Psychology from Adelphi University Institute of Advanced Psychological Studies. At Sharsheret, Shera supports and connects newly diagnosed young women and those at high risk of developing breast cancer or ovarian cancer with suitable peer supporters, advance and develop programs addressing the unique needs of the young women and families of Sharsheret, and counsels individual members of the Embrace program for women living with advanced breast or ovarian cancer.

VIII. About Sharsheret

Sharsheret, Hebrew for “chain”, is a national not-for-profit organization supporting young women and their families, of all Jewish backgrounds, facing breast cancer. Our mission is to offer a community of support to women diagnosed with breast cancer or at increased genetic risk, by fostering culturally-relevant individualized connections with networks of peers, health professionals, and related resources.

Since Sharsheret’s founding in 2001, we have responded to more than 60,000 breast cancer inquiries, involved more than 6,900 peer supporters, and presented over 250 educational programs nationwide annually. Sharsheret supports young Jewish women and families facing breast cancer at every stage--before, during, and after diagnosis. We help women and families connect to our community in the way that feels most comfortable, taking into consideration their stage of life, diagnosis, or treatment, as well as their connection to Judaism. We also provide educational resources, offer specialized support to those facing ovarian cancer or at high risk of developing cancer, and create programs for women and families to improve their quality of life. All Sharsheret’s programs are open to all women and men.

Sharsheret offers the following national programs:

The Link Program

- Peer Support Network, connecting women newly diagnosed or at high risk of developing breast cancer one-on-one with others who share similar diagnoses and experiences
- Embrace™, supporting women living with advanced breast cancer
- Genetics for Life®, addressing hereditary breast and ovarian cancer
- Thriving Again®, providing individualized support, education, and survivorship plans for young breast cancer survivors
- Busy Box®, for young parents facing breast cancer
- Best Face Forward®, addressing the cosmetic side effects of treatment
- Family Focus®, providing resources and support for caregivers and family members
- Ovarian Cancer Program, tailored resources and support for young Jewish women and families facing ovarian cancer
- Sharsheret Supports™, developing local support groups and programs

Education and Outreach Programs

- Health Care Symposia, on issues unique to younger women facing breast cancer
- Sharsheret on Campus, outreach and education to students on campus
- Sharsheret Educational Resource Booklet Series, culturally-relevant publications for Jewish women and their families and healthcare professionals

IX. Disclaimer

The information contained in this document is presented in summary form only and is intended to provide broad understanding and knowledge of the topics. The information should not be considered complete and should not be used in place of a visit, call, consultation, or advice of your physician or other health care professional. The document does not recommend the self-management of health problems. Should you have any health care related questions, please call or see your physician or other health care provider promptly. You should never disregard medical advice or delay in seeking it because of something you have read here.

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