

**Y-Me ShareRing Network
October 18, 2006
7:00 pm Central Time**

Tonight's topic is Genetics and Breast Cancer and our speaker is Shelly Cummings. Shelly is a genetic counselor at the Cancer Risk Clinic at the University of Chicago and we're very happy to have her back with us this evening.

Shelly Cummings: Hi. Welcome everyone. What I'd like to do is, my plan for this evening is to give you an overview of cancer genetics and some of the major genes that we know about; some of the options and whose the most appropriate for genetic testing; some of the pros and cons and limitations of genetic testing, and how to really identify whose the most appropriate for it.

So the first thing I'd like to do is first of all describe what genetic counseling is and what we do as genetic counselors. Traditionally genetic counselors are employed in the prenatal setting or pediatric setting where they talk to individuals who are concerned about a risk of a condition that is in the family like Tay-Sachs Disease or Cystic Fibrosis or if there is a family history of birth defects and more and more genetic counselors are specializing in areas like neuromuscular diseases, cardiology and cancer and that's a result of the human-genome project that we're learning about more genes that are related to conditions.

What genetic counselors do is sit down with individuals or couples or families and communicate with them about the genetic risks and issues that might be occurring in the family. So counselors help clarify medical facts. They explain

how inheritance can contribute to a disorder. They inform individuals of available options and genetic testing is not the end-all/be-all of our profession, we primarily, particularly in cancer genetics provide risk assessments so people can know what their risks are and make decisions based on those risks that are appropriate for them. We also provide some psychological and emotional support and evaluate individuals for adverse psychological effects that they might be experiencing and refer them to appropriate professionals. We also act as liaisons between the physicians and the patients and we have the ability to spend a lot more time with a patient. On average a genetic counseling, cancer genetic counseling session can last from an hour to two hours and that might just be the first visit and sometimes they're quicker depending on the question and the issues that are being discussed. Then the counselor can go back and explain that to the physician what's going on in the family and genetics and how that inheritance plays a role. This is really important because a lot of physicians that are out there practicing didn't even have genetics in medical school let alone cancer genetics. So it is a rapidly moving field and it's of no fault of their own, but it's very difficult to stay current and know exactly what's going on and so there are these counselors like myself who have specialized in cancer genetics. We also help families and individuals ease into adjustments of a new diagnosis or new risk status so that they can understand what is going on and ways to modify their risks.

So the vast majority of cancers that occur in the population are what we call sporadic, they just occur by chance. Somebody doesn't have a risk factor for that cancer but they happen to get it and there's a small percentage of the population, less than 10% that we know are due to an inherited receptibility,

something that gets passed down through the generations. The key to identifying which families might be passing an altered gene down is by taking a family history and when we take a family history it allows us to provide more accurate risk assessment, tailor our genetic counseling and give appropriate medical management to individuals. When we take a family history, we ask about males and females, anybody that has had cancer, what type of cancer, how old they were when they were diagnosed and we try to take at least a three generational pedigree as it's called or family history.

So like dogs, people have pedigrees too and we put this in a picture form with the golden squares and lines and draw out relationships and in most families that have cancers coming down through the generations affecting both males and females and a cancer that is occurring at a younger age from what you would expect for that type of cancer, so for example breast cancer if it's occurring in women less than 50 or ovarian cancer less than 50, prostate cancer less than 60, colon cancer less than 50, all of those are features that are a red flag to us that something might be going on in the family other than just chance. We also look for several generations being affected and we ask about everybody in the family because we want to look at the size of the family. So those are the families that we classify as hereditary families and then when we see other family members that just have one or two cases of cancer in the family, but there aren't any young individuals or there aren't multiple cases then we would classify those as sporadic. Clearly there are families out there that don't have many generations of cancers in their families or they don't even know their family history because they could be adopted, but maybe somebody was young when they were diagnosed and they were the only person. We would still be

concerned about that person's risk, but we would be limited to what we could say about whether there is an altered gene or not. So we base our risk assessment on what we see in the family history and try to modify that as much as possible.

So some of the key features to think about when you think about your family history or when you share information with your doctor, and your doctor should update your family history for every condition, like diabetes, heart disease, but in cancer when we're talking about family history, we look for two or more cancers in close relatives on the same side of the family. We look for cancers that are diagnosed at an earlier age or a younger age. We look for an individual that has multiple primary tumors, so a woman that gets breast cancer for example at 42 and then gets another primary at age 52, not a spreading, not a metastasis, but a new primary. We also look for cases of bilateral cancer. So a woman diagnosed with breast cancer in both breasts would be a red flag to us or a collection of multiple rare cancers. So for example endometrial cancer and colon cancer, while they appear to be two very different types of cancers, can be due to the same heredity predisposition gene. Breast cancer and ovarian cancer can be related to the same altered gene. So when we talk about heredity breast cancer in families, we're also concerned about ovarian cancer and I'll talk about that more in a moment. Then we also look for that pattern of cancers coming down through the generations and it's very important to look at both sides of your family, both your father's side and your mother's side because despite what people may think, breast cancer can be inherited from both sides of the family. The genes that are altered in these families are not on the sex chromosomes. They are not on the X chromosome, they are not on the Y chromosome, they are on different chromosomes and those chromosomes can be passed down from

the father equally as they can the mother and that's still something we still hear today when patients will come in and say, "Well I have cancers on my father's side, but I don't have to worry about that" and that's not the case.

The other thing that is very important to do is to update your family history because while your family history might not look significant at one point in time, things change and when you see your physician you should update that history because a different picture might be present that would give a different risk classification to you.

So we know that there are a few genes that can be altered in families that increase the risk for heredity breast and ovarian cancer and the two genes that we know the most about and that we believe are responsible for the majority of heredity breast and ovarian cancer are called BRCA1 and BRCA2. BR for breast, CA for cancer and one because it was the first gene identified and two, they weren't very clever, it was the second gene identified and everybody has these genes. You get one pair, one member of this pair from your father and one pair from your mother, but certain families have these genes that have been altered a little bit and the way to think of this is that like our English alphabet, we have letters that we put in a very specific order and it spells a word and that word can be part of a sentence and that sentence can be part of the story and that story serves a purpose. In the genetic world, we have four basic chemicals that are abbreviated with letters and those letters are put in a very specific order that make up a gene and the message of those letters is to produce a protein and if the letters are out of order or one letter is missing or another letter is inserted where it shouldn't be, then that message doesn't get translated correctly and not

enough protein is produced or no protein is produced. So a genetic test is looking at, it's like a spell-check to see if the genes are in the right arrangement and so BRCA1 and 2 we know can be altered in families where we see early onset breast and ovarian cancer. There are other genes out there that are much less common than BRCA1 and 2 and we would see other types of cancers in the family besides breast, ovarian, male breast cancer, pancreatic cancer, colon cancer and prostate cancer. Those are the big cancers that we tend to see in BRCA1 and 2 families.

So we know that in individuals that have an altered gene, an altered BRCA1 and 2 gene that their lifetime risks for getting breast cancer is higher and I want to emphasize that having the altered gene doesn't mean you will automatically get breast or ovarian cancer or any cancer for that matter, it just means your risk is significantly higher than an individual that does not have that altered gene. So what we know is that individuals that have an altered gene have up to an 85% lifetime risk of getting breast cancer and that compares to the general population risk of about 12%. Now there are other studies that have been published that have a number that's lower than that, 56% in one study, but still that's significantly different than 12%. What we know is that when there's an alteration in these genes, and there can be many different types of alterations in these genes, we can't specifically tell somebody that their risk is 85% or is 44%. We just know that it is significantly higher than the general population's risk. We also know that if an individual has already had a breast cancer and is found to have an altered gene, her risk for getting a second primary breast cancer could be as high as 60% and that number is much higher than an oncologist would quote a woman of having a recurrence or having a new primary. Now these numbers

have nothing to do with a recurrence of getting breast cancer, your chance of recurring is based on your diagnosis, how large the cancer was, if it was in the lymph nodes, the grade, all of that, so I'm not talking about recurrence, I'm talking about new primary.

The other thing about BRCA1 and 2 is that we see a high association with ovarian cancer and in our eyes this is much more concerning because we don't have as good of screening tools for ovarian cancer like we do breast cancer. With breast cancer you can give yourself a breast exam, you can do ultrasound, you can do MRI, there are other tools. With your ovaries you can't screen them, you can't touch them, you can't really do any screening that is truly effective. The other unfortunate thing is when ovarian cancer is discovered or when a woman finally presents, about 70% of the time it's already at an advanced stage and so if we identify these families that are at increased risk based on having an altered gene, they might make different decisions about their ovaries. If they are done having children, if they have already gone into menopause, then they might take their ovaries out to reduce the risk of getting ovarian cancer.

We can also see an increased risk for prostate cancer and colon cancers in these families. However, the numbers that were first published when these genes were discovered haven't necessarily played out true in the population, but we just say, "This is slightly higher risk" and we really don't tailor any medical management based on that slightly higher risk because it's not significantly higher.

Now in BRCA2 families we can see male breast cancer and there have been some reports that have associated male breast cancer with BRCA1 as well, but you still tend to see it a little bit more often in BRCA2 families. Male breast cancer is extremely rare, so anytime we see a male with breast cancer we become extremely suspicious there might be an altered BRCA2 gene in that gentleman. We can also see other types of cancers in BRCA2 families. We can see laryngeal cancer, pancreatic cancer is another one, we can sometimes see thyroid cancers in these families and so BRCA2 tends to have a little bit wider type of cancer spectrum than BRCA1 where we traditionally see breast and ovarian.

The other thing about BRCA1 and 2, the other difference between BRCA1 and 2, in BRCA2 families we frequently can see women that get breast cancer at a little bit older of an age, so around menopause or post menopause whereas in BRCA1 we frequently see them at a much younger age, so in their 30's and 40's. So sometimes looking at these families, in BRCA2 families, you see a women whose post menopausal and gets breast cancer, frequently doctors think less of that because that's more like the average age for a woman to get breast cancer, but what's important is to incorporate the whole family history into the picture and if four women got post menopausal breast cancer that clearly is not normal and so we need to evaluate that for genetic susceptibility. With that said, we can do a great family history, we can do the best test possible that's out there now and you can still have a very strong family history and not identify an altered gene. So we know we haven't discovered all the genes that are related to heredity breast and ovarian cancer, but we are making great advances.

When I became a genetic counselor in 1995, it was before clinical genetic testing for BRCA1 and 2 was even available and now we've advanced just in that period of time to tailoring our medical management to individuals, to offering breast MRI's to women who have an altered gene and we know so much more about preventative measures and different medications than we did. So that's, while it's not as fast as we'd like it to be, it is happening much faster than it ever used to happen, but we still see families that we can't identify the altered gene and last week there was a report that mentioned a third breast cancer susceptibility gene that was out there that was reported in England and that shows great promise. So it's going to take a while before that reaches primetime and before we can actually offer that to families, but there is a lot of research going on in this area.

The other thing that counselors use are risk prediction models and we sit down with individuals and take their family history, their reproductive history into account and we can calculate models for the chance of a person getting breast cancer and the probability that there might be an alteration in the family. Now all of these models have limitations to them, but they're tools that we use to evaluate what's going on in a family and quite honestly even if the tool, if the probability comes back very low, but the family history is striking, then we have to fall back on our clinical judgment and our training to know that, yes, this person is appropriate or there is something more going on in this family than just coincidence.

The next thing that I wanted to talk about is some of the reasons why people might consider genetic testing. Some people consider genetic testing because, many people consider genetic testing for their children or for the next generations

because whether they've had cancer or not, if they're coming forward for genetic testing there usually is a family history of it and they either want to reduce their chances or provide information to the next generation so that they can use that to make decisions. Risk assessment can help that and the next steps would be genetic counseling if it is appropriate and it's interesting in the families that we see there could be two sisters from the same family that have the same cancer family history, but they might have very different perceptions of their risks and make very different decisions. One might be extremely proactive and she might want to take her breasts off preventatively, where another one might not be concerned at all and might not even be getting mammograms where we would kind of work with that one not getting mammograms and try to educate her about why it would be important to at least have screenings, she wouldn't necessarily need to have testing or to have a prophylactic mastectomy. However with that said, we know that women who are at increased risk for having this altered gene, those that elect to have a prophylactic mastectomy can reduce their risk of getting breast cancer by about 92%, which is a pretty high number. It doesn't eliminate the risk, but it does reduce it. Some women also can make the decision to take their ovaries out preventatively or have a prophylactic oophorectomy and we know that prophylactic oophorectomy reduces the risk of getting ovarian cancer by about 96% and in pre-menopausal women reduces the risk of breast cancer by about 53%. So for some women who are considering a surgical route for risk reduction, those are the numbers that we share with them so that they know what we know about these methods.

Then it is also perfectly reasonable for people to make decisions just to be followed more closely or intensive surveillance and there's different ways that

people can do that. There are different studies people can go on that involve screening more closely. Those might include MRI's. They might include getting certain labs drawn, but we always counsel individuals that intensive surveillance is good, but it's watchful waiting, it's not risk reduction, so we just want to make sure that people understand that difference. Then there's also the option for some people who wish to take a medication to reduce their risks. Tamoxifen has been shown to reduce the risks of invasive breast cancer by about 53% and so that's another option that we discuss with individuals, but we really emphasize that there really isn't any one right answer for everybody. It needs to be made in a careful process driven manner and with multiple conversations.

The other thing that counselors do is what's called anticipatory guidance. So if somebody is coming in and if they are going through the testing process we might say, "Well what would you do with your...how would you feel if your test results were positive or how would you feel if they were negative and who would you tell and how do you think they would react and who would you want to share this information and what decisions do you think you would make?" We go through this exercise so that they will know or have in their mind a little bit about what might happen, what might happen in the end. So if they are going along and expecting the results to be positive and then suddenly they're negative, but they've been living their whole life like they're going to get breast cancer that can be a whole new way of thinking for them and it may take a lot of adjusting. We've seen a lot of women who have been getting mammograms at very young ages because of a family history and then they find out that they aren't a mutation carrier based on a mutation being present in the family, but they're not ready to stop getting those mammograms and this is for women who are younger

than the age of 40, for example. So they aren't comfortable in giving up that security blanket or tailoring what they've been doing. We also consider their motivation for testing. Some people come in and they say, "I want to take that test to tell me if I'm going to get cancer" and I usually reply, "I would like to take that test too, but there isn't one yet." What we know is that this test can tell us if individuals are an increased risk for cancer based on a family history. Sometimes their motivation, like I mentioned, is for family members. Sometimes they might have just been diagnosed with breast cancer and only want to go through one surgery and might be treated for that breast cancer with a mastectomy in the one breast and then a prophylaxis mastectomy in the other breast and so that is something that is also discussed at the same time.

We also talk to individuals about how they're dealing with this, who is their support group, who is their support system, do they have people they can reach out to, would it be helpful for them to talk to another one of our patients to know what it's like to make these decisions and they can share their experiences with each other. A lot of patient's perception is going to be based on their personal experiences with cancer. Somebody who has had a lot of cancer in their family and had it diagnosed at a young age, but everybody lived to an older age and died from something else is going to have a very different perception of cancer than the next individual who had people in their family get cancer young and die young and so that all goes into how people think and what they think they should do for themselves. We also spend a lot of time just listening to individuals because frequently they don't have an opportunity to express their concerns in this kind of environment and their concerns about susceptibility or genetics in the family.

One thing that does come up, but it comes up less often thankfully is the fear of insurance discrimination and there is a federal law in place which is part of the HIPAA Law that thinks that genetic information can't be used to determine eligibility and this is for people who have group health policies. It also prevents insurers from charging different individual premiums within a group plan and it states that genetic information can't be viewed as a preexisting condition and that applies to people that don't already have a diagnosis, but if you've already had cancer, you're already considered high risk. It doesn't matter to them if it's related to an altered gene or not, because you've had it once, you're considered high risk. The unfortunate thing about the HIPAA protection is it doesn't prevent access by insurers to the genetic information, it doesn't prevent the insurer from demanding genetic testing as a condition of coverage, it doesn't protect against group health or group rate hikes, but the insurance company is not going to raise the whole group's rate based on one or two people within that population and there is no protection outside of a group policy or a group market or those people that have individual policies aren't protected at all. That's a federal law.

Illinois has a very good law that defines penalties if there is anybody that has experienced health discrimination, employment discrimination, any disclosure or violates confidentiality. My take home message to people about insurance discrimination is that the fear that's in the community, the fear that the media aren't perpetuating is much greater than the reality of it occurring. More and more insurance companies are paying for these genetic tests because they know it's in their best interest to identify people that are at increased risk, get them on a screening management program which will cost them a lot less money than if

that person should get cancer and need treatment, surgery, chemo, radiation. Sometimes we have to argue with insurance companies and justify our cases, but nine times out of ten it's covered.

The genetic counseling process should be done in person. The information should be given to people in person. Usually it's followed up with a letter and the disclosure for test results is always done in person. There's always a plan for follow up for recommendations and depending on the center many times there can be enrollment into research studies and clinical trials.

The bottom line is that all cancers arrive from genetic alterations, but there's only a few families, a small percentage of the population that are due to an inherited susceptibility and like cancer development, genetic counseling is a testing, is a multi-step process that will lead us down the pathway to making, helping individuals make a better decision about their life plans. We hope to one day develop predictive tests to identify genetic predispositions, to develop diagnostic tests to detect cancer at its early ages and then targeted gene therapies. So that's the way the future of cancer genetics and counseling is going and I am ready to answer any questions.

Leslie from Illinois is on line. Please go ahead.

Leslie: Yes, good evening. That was a wonderful lecture. That was the first thing I want to say. **When you're talking about preventing cancer, preventing the probability of cancer by using, would you use something like Raloxifene or Evista for post menopausal women rather than Tamoxifen?**

Shelly Cummings: There's a study that came out relatively recently that showed that Evista also reduces the risk of getting invasive breast cancers. It was less effective in noninvasive breast cancers like (inaudible). So the discussion about using Evista over Tamoxifen is one that should be discussed with your oncologist. It's much more of appropriate drug-related question because some women shouldn't take Tamoxifen because of the risk of blood clots.

Leslie: Right.

Shelly Cummings: And endometrial cancer can be associated with Tamoxifen and sometimes women have some additional side effects, fewer side effects than already post menopausal women, but it certainly is an option and it certainly is one that we discuss at the University of Chicago with women.

Leslie: Okay thank you.

Operator: Nancy from Maine is on line. Please go ahead.

Nancy: Yes, thank you for the lecture; it was wonderful. **I'm wondering about who would get tested. It seems that if there is an altered gene in the family, it's not necessarily inherited. So would it be the person who is concerned that they might get it, you know the younger generation to get the test or the person who has cancer?**

Shelly Cummings: Okay that's a good question. So to clarify a little bit of something that you said, if there is an altered gene in the family that defines a hereditary predisposition, so for example, well let me step back. What we know about these genes is that everybody has them, but people can have these genes and we know that there's people walking around with altered genes that never get cancer, but when you have a family history of say a mom that has had breast cancer at 40 or 45 and her daughter now is 23 and concerned about her risk, the best person to test in the family is somebody that has had cancer because, particularly that 40 year old that had cancer because she had it at a young age, she had it at a suspicious age where we would think it might be associated with an altered gene and I'm assuming there is going to be some other family history to support that. So testing someone who has had cancer increases the probability that we'll get a test result back that we can interpret, meaning that we'll get a positive test result and we can say, "Okay this is an alteration that is likely associated with cancers in the family." The reason why I say that is because if say that woman's daughter was tested and she was negative, she didn't have an altered gene, we wouldn't know for sure if that negative test result was a true negative, meaning that there is an alteration in the family but she just didn't inherit it, because if the mom has the altered gene there's a 50/50 chance that she could pass it down to each of her children. It's like taking a quarter and getting heads or tails and it's truly random, 50/50. So if we test the daughter and she was negative, we wouldn't completely be comfortable that she was a true negative because we'd like to know for sure that there's an alteration in the family and she really didn't inherit it. So we would ideally test mom first. If mom is positive for an alteration and then we test daughter and she is negative, then we can tell that 23-year-old

daughter that she did not inherit the altered gene and she is back to the general population's risk.

Nancy: **Fine, just let me have a little addendum in there. I understand that part, now where the cancer has been in the family, the two people who have had it, the mother and grandmother, both were 65 when they got it. Would you consider that worth testing or is it because they are in the older group that it is not probably the BRCA1 or 2?**

Shelly Cummings: Yeah, so that's a harder question to answer.

Shelly Cummings: So we know that there are mother/daughter pairs out there that get breast cancer and they get them at an older age. We would certainly say that it wouldn't be unreasonable to consider genetic testing, but when we do all of our fancy statistical calculations the probability of us finding an altered gene would probably come up low, but like I said before, we know that we can get a low number and still have somebody have an altered gene. So it really boils down to what the family wants to do, you know how they're going to use that information. So if mom and grandmother didn't get tested or couldn't be tested for whatever reason, we would say that that daughter is at increased risk because there are two women coming down the family history that have had breast cancer and her risk is not as high as if that mother and grandmother had breast cancer in their 40s, but that is much tougher of a call when they're in their 60s. So it's still reasonable.

Nancy: Thank you.

Arline Kallick: **You just talked about if the mother is tested positive what that means for daughter, but I don't know that you really covered if the mother tested negative, and this is a mother that had breast cancer, what does that mean for offspring?**

Shelly Cummings: Right. So if a mother, if a family history is suggestive of a hereditary predisposition and the mother tests negative, then we fall back on our clinical judgment or basically our Mendelian Genetics which regimental other genetics. We look at the family and say, "Look, there are way too many cancers in this family; there's something hereditary going on" and we would still treat that family as high risk even though we haven't been able to put a name to it by identifying an altered gene.

Arline Kallick: **What does that negative test mean to the offspring then? It doesn't mean much then.**

Shelly Cummings: It doesn't mean much.

Shelly Cummings: It kind of checks off the list the genes that we know the most about, the genes that we think are primarily responsible for hereditary breast and ovarian cancer, but we're still going to manage or give recommendations to those people, that offspring, based on the cancers we see in the family. So if like mom had ovarian cancer at 50 or 60 even and if her daughter is concerned, we would screen the daughter for ovarian cancer. We would want to be more proactive than just say,

“Okay we did this test; it’s negative; you’re okay.” You know that that’s not the case.

Arline Kallick: Thank you

Operator: *Julia from Indiana is on. Please go ahead.*

Julia: **Hi. I have BRCA2. I had DCIS when I was 41, I’m 45 now and for the last couple of years I’ve been getting MRI in addition to a mammogram. I read a few months ago about a study from Europe suggesting that BRCA2 women not get mammograms because of the x-ray and only get MRI. I’ve asked a couple of other times on these ShareRings when we’ve had oncologists come on and talk to us about that and they’ve all strongly recommended continuing the mammogram as well as the MRI. Does the genetic counseling community have any recommendation on that?**

Shelly Cummings: So I’m going to tell you what we do at U of C because I’m a little bit at an inability to speak for the whole genetic counseling community, but in our practice we combine MRI with mammography and Europe in general does different things than we do, good or bad, many times good. They have modified care compared to what we offer here, but we know and there’s lots of data to demonstrate the benefits of mammography in addition to MRI. MRI is really good at looking at women that have dense breasts, women that have implants, but MRI is very reader sensitive.

Julia: Right.

Shelly Cummings: So you can do MRI in lots of places, but if the person reading it is not skilled at picking up the nuances they see in the breasts then a woman might have multiple biopsies that they might not need.

Julia: Right.

Shelly Cummings: So MRI is very sensitive, but it's not as specific as we would like it to be and so that's why we combine MRI with mammogram. Some centers in the United States are alternating it, so you would alternate every six months with an MRI and a mammogram.

Julia: Oh I do them at the same time.

Shelly Cummings: Other places are doing it at the same time so that you can compare both procedures together.

Julia: **Right and my DCIS was detected on a mammogram so I'm pro mammogram, I just didn't know if suddenly I should not be getting them.**

Shelly Cummings: No, we wouldn't...the amount of radiation you get from a mammogram is the equivalent of flying from New York to San Francisco. You get radiation when you're on airplanes.

Julia: Okay.

Shelly Cummings: And unless you do that a lot then your exposure is very minimal for the benefit you're getting.

Julia: Great. Okay thank you.

Operator: *Julianne from Illinois is on line. Please go ahead.*

Julianne: Hi yes, thank you very much. Your information has just been fantastic this evening.

Shelly Cummings: Oh thank you.

Julianne: **I was wondering, is there any correlation between being HER2 positive and being BRCA positive?**

Shelly Cummings: Good question. So there's a lot of research going on in pathology to look at which tumors and which cancers might be indicative of BRCA1 or BRCA2 and right now what we know is that when we look at women who have had breast cancer, those women that are ERPR and HER2 negative are more likely to be BRCA1 carriers than not and those are women that have a family history as well, just not a sporadic cancer. We have yet to see in our population anyway HER2 positives BRCA1 and we're kind of waiting because we want to see how that cancer might be different in some way, histologically different. We also know there's some evidence that shows that women who are BRCA2 carriers are more likely to be ERPR positive compared to BRCA1. So there's still a lot of work that

is going on in this area and it's not always consistent and so that's why we aren't to the point where we can see a woman's pathology report and say, "Oh yeah, she's got a BRCA1 mutation."

Arline Kallick: We have to take our last question now.

Operator: Our last question comes from Evelyn from Connecticut. Please go ahead.

Evelyn: Well thank you. **I wonder if you would say something about the company which I believe is the sole company that does this, actually does this testing. Would you refresh my memory as to its name?**

Shelly Cummings: Myriad.

Evelyn: **Is there some reason that there is only one company in the whole country that does all the genetic testing?**

Shelly Cummings: Yeah, so they have a patent right now and I'm not completely familiar with patent law, but I know that as part of this patent, the full sequencing which is the big test that we do to look for all possible changes in these genes that we can technically do, their technology is such that they developed it in a way that everybody has to send samples to them. That's just the way it is and there were huge, there were some legal battles between Canada and I think Europe for a while to try to get outside of that, of those patent agreements, but that's the case and quite honestly, and I have nothing to do with Myriad other than I send my samples there, but honestly they're a good company in that they provide a lot of

educational support; they provide a lot of resources; they provide alternative payment methods for families. They also take a burden off of me in that they look into patient's insurance for me and for the patient in advance to see if insurance will cover the cost of the testing. So the patent, you know their deadlines or their end date to the patent agreements, and I'm not quite sure when that may be and when it would open up, but having the monopoly on the test in my experience with this lab and my experience with other laboratories, I'd have to say that it has been nothing but outstanding from a customer service standpoint, from interacting at my level, from getting questions asked, to even supporting, they even supported a patient education conference that we had a couple of years ago, multiple times actually. But as far as the patent's ending date I'm not quite sure when that occurs.

Evelyn: Thank you very much.

Arline Kallick: Well we have to end our question and answer session right now before we go to a support group breakout. Thank you so much Shelly for speaking this evening. You covered so much material that I know everyone is going to want to go to the transcripts and read them over again.

Shelly Cummings: You're welcome.

Arline Kallick: Thank you. Have a good evening.

Operator: All participants please standby. All parties will be placed on a music hold until the breakout sessions begin. Please do not disconnect. Once again, you will hear music until the breakout sessions begin. Thank you for your patience.



**Y-ME ShareRing Network
October, 2006**