October is breast cancer awareness month and NSGC and the Centers for Disease Control and Prevention want to help women learn the steps they can take like genetic testing and genetic counseling to understand their risk for breast and ovarian cancer. Today Dr. Coles and I are talking about what women should know about direct to consumer or DTC genetic testing and what it can and cannot tell women about their risk for breast and ovarian cancer.

Dr. Sarah Coles: I think this is a really timely and important topic because there is increasing availability and demand for direct to consumer genetic testing. These are really heavily marketed tests. I hear them all the time on podcast advertisements for example, and are available to consumers without involvement of healthcare professionals. That's what makes them direct to consumer genetic tests.

Joy L. Haidle: I agree. There's a lot of presence on social media and for the public to understand about direct to consumer testing, but I think there's some confusion around what is a direct to consumer test, what it can and what it can't tell you and how is that different from the kinds of tests that our physicians or genetic counselors might order. I think an important difference between the direct to consumer tests is they're really meant to be something that's more fun and recreational. It's not typically something that we would use those results to guide medical decisions. However, there's some misconceptions that perhaps those tests actually are medical grade tests that could be used to guide care. How do you use the direct to consumer tests with your population of patients that you serve, Dr. Coles?

Dr. Sarah Coles: Well, typically I don't or I try to avoid it. In general, the United States Preventive Services Task Force recommends particularly for breast cancer genetic testing or
genetic counseling for women who are at increased risk of having a pathologic genetic mutation for BRCA 1 or 2. But they actively recommend against genetic testing for women who are at average or lower risks of these kinds of pathologic mutations. So we spend a lot of time in my practice talking about what are the risks and benefits of direct to consumer genetic tests, how complicated genetic testing really is, how it involves addressing risks for disease states, prevention, diagnosis, disease management, and then what are the implications for you, your family, and your future reproductive health once you get these test results. So typically in my office it's only when they come in and they say, "I have these test results. I went to X company and got these tests. What do I do with it now?"

Joy L. Haidle: I agree. In my practice, I've seen this kind of same comments come in more frequently about people who've already done the testing and thought that they had gotten some really useful information for themselves and the family. It's not really until we sit down and have a really thorough conversation about what the test actually was able to tell them versus what it's not. Depending on those results of the testing, we always recommend that that test actually be repeated in a clinical laboratory before any, before those results are used for medical management purposes.

Dr. Sarah Coles: Yeah, and that's one of the real big problems I think for consumers who are out there purchasing these genetic tests, but also for when they come into a primary care office is there are so many genetic tests available in a direct to consumer market and they're ever changing and ever expanding. It's hard to know which ones are okay, which ones aren't. These companies don't necessarily have to use FDA approved methods and tests. So it can be really complicated to kind of figure out is this a good result or not and what do I do with this information?

Joy L. Haidle: I'm glad you brought that up because I think one thing that would be helpful for the consumers to actually know about is when they're trying to decide on do I want to do a direct to consumer test or not, is to take the time and look at the information that's available on the websites. Because most of the information that's important is actually in the fine print and in the details. But many people don't take the time to read that and they just click through quickly to get to the end game, which is send me the kit. It's important to take a look at what is it that the laboratory is offering, what kinds of genes might you get information about, how good of a job is the test going to do at actually answering the questions that I have, but also take a peek at what resources are available through the laboratory.
Joy L. Haidle: So if I do have questions or if I do find something unexpected on that test result, is there someone available through the laboratory to support me to answer those questions or to do I get in to see my primary care providers such as yourself or a genetic counselor to talk through what those tests are? It's also important to take a look at the fine print to determine who has access to those test results. Is it just me? Are those results perhaps sold to outside markets to be used in additional research? So it's important to take a look at what is the privacy in my information that I submitted.

Dr. Sarah Coles: Yeah, I have a lot of concerns about genetic privacy in some of these companies. A lot of these companies that provide direct to consumer genetic testing are not necessarily bound by the strict healthcare privacy protections that healthcare providers are like the Health Insurance Portability and Accountability Act, lovingly known as HIPAA. So it's possible for companies to claim ownership of your genetic information and to provide that information to third parties without necessarily even giving you that information back, right? Correct me if I'm wrong, but I believe the Genetic Information Nondiscrimination Act, GINA, only protects for certain things but doesn't protect you from discrimination based on life insurance, disability insurance or long term care plans. So your genetic information can be used against you sometimes and we don't really know in the future how much that's going to become a problem as more and more genetic information is available.

Joy L. Haidle: Yeah, it's always important to take a look at the fine print before you click purchase a kit and send in a saliva sample. But I think one thing that's important too is for us to visit about is kind of the difference between the tests that people can do over direct to consumer. Because there is, there definitely is a place for that kind of testing for people who are just really curious and really interested compared to the kinds of tests that you might order or I might order for genetic testing. So for example as a genetic counselor, one of the tests that we might order, we'll take the time to draw out a very thorough family history to get an idea of what are the patterns of cancers that might be present in your family for example. So it's important for us to have a conversation about what kinds of cancer in the family, which relatives have that kind of cancer, how old were they at the time they were diagnosed.

Joy L. Haidle: Conversations, since DNA testing has become more common, it's helpful to ask if some of your relatives have already had genetic testing done and would they be willing to share those tests results. It also can be really helpful to talk with the women in the family about whether or not they had had their ovaries or uterus removed and what age, roughly what age they were at the time they chose to do the surgery and why. Because all of those information comes
together as part of that full risk assessment and that helps the genetic counselor or the physicians take a look at that pattern and determine what is the best test for my family to help answer some of the questions that we have. What process do you use in your practice, Dr. Coles?

Dr. Sarah Coles: Yeah, so very similar. So a very detailed family history with a particular focus on primary cancer site and age of onset or age of diagnosis of that cancer, as well as other potential lifestyle and behavior changes that might be made to reduce cancer risk. Then a big piece of it is also what would you do with this information? Would having this information change something for you? Would you do some chemoprophylaxis surgery? Would it change your reproductive decisions? How would this impact your family? Because if the answer is having this information wouldn't change anything for me, then we probably don't want to run this test. Because there are certainly risks to knowing genetic information, right? So there can be increased anxiety and depression, increased worry. There can be concerns for your family members about what does this mean for my health and future wellbeing and does this mean something about my reproductive health in the future? So I would rather not put somebody at risk without having a clear benefit for them.

Joy L. Haidle: I totally agree. I love that point because it's important that just because we have tests available doesn't mean that every person finds value in them. For those perhaps, would you be willing to share a little bit about, as a physician, when you get such a positive test result for one of these breast cancer genes, how does that test result influence the options that are available to women and their family to either reduce the risk of developing a cancer or what type of surveillance might be a good idea to consider?

Dr. Sarah Coles: Yeah. So it does change kind of how we're going to approach your future risk for breast or ovarian or peritoneal cancers, particularly with like BRCA 1 or 2 mutations. So we talk a lot about surgical risk reduction, so things like bilateral prophylactic mastectomy or bilateral salpingo oophorectomy when you're over the age of 35 and your childbearing decisions are all completed so you don't want to have kids anymore. We might be talking about chemoprophylaxis and medications like Tamoxifen or if you're not interested in that or not a good candidate, even just oral contraceptive pills. Then talking about how do you talk about this with your family and what their risks might potentially be. So it just change a lot of your options. For people who don't want to do any risk reduction, we're probably going to talk about screening more frequently. That's going to be an individualized decision.
Joy L. Haidle: Sometimes the options that are available really change over a person's lifespan. For example, that same data might have a different impact to me when I'm in my twenties versus when I'm moving towards my forties and then up into the older years. So what people pick from might be very different depending on where they are in their lifespan, what their experiences have been with cancer and the family history of those relatives had done well from their treatments or if they had lost members of the family, that can influence what people choose to do with how they want to use that test result.

Dr. Sarah Coles: Oh, totally. It's all about kind of defining what their values and goals are in healthcare. That's why the counseling piece is so important for this and the thing that can be lost in direct to consumer genetic testing. Because you really want to know what is this person hoping to get from this and what do they really value? Is it more important for them to know something and to prepare? Or is the fear and anxiety crippling? That's not really where they want to go. So what is it that is most important to them in their healthcare and so that we can help either with our testing or not testing meet those goals for that person.

Joy L. Haidle: You brought up something a minute ago that I wanted to circle back on because I thought it was a great point. You were talking about how when we get the test results, how is it that we can go about sharing that information with family because that is a time period where some people can be a little bit uncomfortable and it often depends on the dynamics and the relationships and communication flow within a family. Some families are quite open with sharing that information, others are not and it takes a little bit more time to be comfortable sharing that information and some people just never get to that comfort level. I think as a genetic counselor, if I'm meeting with a family and we find a mutation on one of these gene tests, we talk about that at the results appointment to talk through what does the result mean to you, how does that impact the other people in the family because that person becomes an information resource for the other members of the family.

Joy L. Haidle: Often, we'll encourage them to share a copy of the test results to the people that are related to them by blood and we'll send a relative letter that will help communicate that process so that it becomes the family members' choice if they wish to act on that information or not. So the responsibility really is for the person who found the gene changes really just to share it and then it's up to the relatives to determine if they want to find out that information or how they might wish to do that.

Dr. Sarah Coles: Yeah, I think that's a lovely way to do it. One kind of benefit that I have as a family doctor is I often take care of whole families with multi-generations and
sometimes extended families, which is great. So I can also offer if their family members are my patients as well, as I can mediate a meeting. So if they're interested in sharing this information with their family and they're not quite sure how or that people might have questions, I can set up an appointment with multiple members of the family to talk about, "Well, this is the results. Here's what we could do with this for each of you and move forward from there." That's one of the joys of being a primary care doctor.

Joy L. Haidle: Sometimes even as a genetic counselor I can mirror a little bit of what your experiences too.

Dr. Sarah Coles: Oh, I bet.

Joy L. Haidle: We can become a resource for the entire family. Just like you, when we find a gene change, at least in my practice, individuals and their family can come back on a once a year basis to stay current about what do we know about that particular gene and have the surveillance or the risk reduction options changed. The goal of that meeting is to keep them current so that they can have an informed conversation with their physicians to set that management plan. Because personal preference often is a partial driver and in what that plan might be.

Dr. Sarah Coles: Oh sure. One question I had for you in thinking about kind of how you handle families, so a big piece of this is doing a pedigree and looking at family history when determining kind of your risk and whether or not to get the test and which specific tests you get, what do you do for people who don't have a clear family history or your family history is completely unknown?

Joy L. Haidle: That's a good question because we get asked this quite a bit for people who might have been adopted or for just communication patterns in a family, that they've really just lost touch and people move all over the country now and we don't always get a chance to see each other face to face. So in situations where someone may have been adopted, if there is someone who had been diagnosed with cancer at a young age and that's what's driving it, they can still be a candidate to move forward with genetic testing and it's just that it becomes an uninformative family structure. So no family history is not the same as no known family history. So in that situation, it still would be appropriate for that person to consider testing as they might wish to have that information to help determine what do I want to do about it, what are my action steps moving forward.
Joy L. Haidle: For individuals who it's just family dynamics where they might not know a portion of their family history, as a genetic counselor, when I'm taking the family tree, I'll mark a question mark on the family tree from my perspective to remind myself when we're talking through the risk assessment that this piece of the family history is not known and that happens very frequently. So in that case, we have to take a look at does the individual and their family still meet testing criteria that might be billable to the insurance companies? There are other options where it feels like that person would benefit from pursuing testing, but it may not be covered by insurance as some of the clinical laboratories that offer the really thorough genetic testing have options where someone might be able to do cash pay. So it's a way for them to have access to a good quality test, still be able to get that, the test completed at a reasonable cost without there being a barrier.

Dr. Sarah Coles: I have definitely heard of people using the direct to consumer genetics test too as kind of a work around for when they feel that genetic screening or genetics testing rather would be appropriate for that person, but that insurance doesn't cover and it's too expensive. How do you feel about that?

Joy L. Haidle: There are times where it happens and as a genetic counselor who might be meeting with that someone, when we hit that situation, there are times that I will be working with one of the laboratories that offers that cash pay option to keep it at a reasonable price so that they might still be able to have access to that test result. There are times then that they may talk to their insurance company after the fact to see if that medical expense could be applied to their deductible. But there is an option for that workaround to be able to still have access to a medical grade clinical test without, and be able to use those results in your medical management, that's different than the tests that might be ordered on a direct to consumer basis that's meant for recreation.

Dr. Sarah Coles: Well, under the Affordable Care Act, USPDSF grade A and B recommendations are supposed to be covered for the marketplace plans, so access to genetic counseling if they're available in your community should be covered if they meet some of these high risk indications. The problem of this of course is Medicare or Medicaid plans that maybe don't always cover this. So it's something certainly that I could see coming up. I serve a particularly underserved population so I could totally see people saying, "Well, my insurance doesn't cover it or I don't have insurance at all. I'm just going to go online, buy a kit and see what happens."

Joy L. Haidle: Yeah, or we're all super busy. It's online in the evening. This is when I have my free time and I'll do it. But one of the things that you brought up a second ago I
wanted to touch on again is sometimes it's a barrier and just not knowing what resources are available to you either locally or available perhaps over the phone. So NSGC has a website, it's findageneticcounselor.com, which is a great resource for either providers who are looking to refer a patient to a genetic counselor or people who want to find a genetic counselor on their own either because they already did the direct to consumer testing and they have questions about what the results mean and what to do or they're wanting to have that formal risk assessment. So that's a way to find genetic counselors that are close to home, that could see you face to face.

Joy L. Haidle: But there are plenty of genetic counselors that do the risk assessment and the consultation over the telephone. So if you happen to live in a more rural area or that isn't as well served, there's definitely options to still reach someone if you have those kinds of questions.

Dr. Sarah Coles: That's a really good resource to know about. Even I live in Phoenix, Arizona, which is a giant city and even now it can sometimes be difficult to find genetics counselor and there's such amazing resources for our patients and they have so much more expertise than I do in this that sometimes it's very helpful to have that other voice that can come in and kind of help go through some of the nuances of all of this. So I'm really excited about the NSGC website resource. Then of course there's the CDC's Bring Your Brave Campaign, which also has a lot of resources for patients, particularly focused on breast cancer in women under the age of 45.

Joy L. Haidle: Yeah, I really appreciate how you know that the genetic counselors work very closely with the medical community and the physicians to help do the risk assessment and then we work together to help provide the optimal care for the patients. Another option, I think one of the common questions that we get and another option that's available for resources is there's a website through NSGC called aboutgeneticcounselors.com, and on that website there are different resources to click on to help you prepare for what kinds of questions do I need to ask, what is a genetic counseling session, to help flatten that learning curve and feel more confident and prepared going into the appointment. So it's a great resource if people want to take a look at that to get ready and to be more comfortable with that consult.

Dr. Sarah Coles: Yeah. I think kind of added to that is just what risks do these gene changes have? Because like you said, there's a lot of them and we are finding them all the time. So what is the increased risk with this gene change? But also what is the decreased risk if I do some of these measures, if I have a mastectomy, if I do
other chemoprophylaxis, how much does that impact my risk and what is more important to me?

Joy L. Haidle: I think that ultimately will be dependent on what is the gene that's actually identified on that -

Dr. Sarah Coles: Totally.

Joy L. Haidle: I tend to lump things in a couple of different categories, so I tend to reference strong genes. What I mean by that is the chance of developing a cancer over a lifetime with one of those stronger genes like BRCA 1 or 2 for example, is significantly higher than women in the general population. So thinking about starting surveillance as young as 25 or thinking about risk reduction at some point over a lifetime by doing about bilateral mastectomy, those are really big decisions and impacts that thought process and care plan over the lifespan. There's also a bucket of genes that I tend to reference as more of like a moderate risk and what I mean by that is that chance of developing a breast cancer over lifetime hovers right in that 20 to 40% window. But people feel very differently about what that number means.

Joy L. Haidle: Some people feel that 20 to 40% is quite high and they really want to be doing risk reduction. Other people are much more comfortable with the risks in that risk assessment in that range and would prefer to do heightened surveillance. Then there are other times that we'll find that the family history contributes just a slight elevated risk, but that chance of developing a breast cancer over a lifetime isn't quite high enough that we would be changing surveillance based on any particular guideline. It would still be mammogram, but we often will tailor that to what the ages of onset of the breast cancer were in the family. But as a counselor, that's when we will do the risk assessment, we'll talk about those things, but that's when we really work closely with our physician colleagues such as yourself to determine what is the best plan that's right for that person.

Dr. Sarah Coles: Right. That through shared decision making with the patient about where do this matters for them and how this will impact. I think it was a really good point you made earlier too about where you are in your life and how old you are and kind of where you are milestone wise really matters. Because if you're 25 and thinking about having your first kid, you maybe don't want to have that double mastectomy if it's really important to you to breastfeed. Whereas if you're 40 and you're like, "Ah, I've had all my kids, I'm done. Go ahead and take my ovaries." It might be a very different discussion and a very different outcome.
Joy L. Haidle: Completely agree. But sometimes people as they start those conversations in the family, some of the trends that come up is they're afraid to ask their relatives about whether or not they'd be willing to test. Because sometimes I'll have the daughter or a granddaughter that comes in and they're asking the question about should I be the one to do genetic testing? There's sometimes is a misnomer about, I'm the one that should be getting tested because I haven't had the cancer yet versus my mom or my grandma actually had the cancer, they were young at the time they had it. They're the better testing candidate. But the thought is as well, I already had the cancer, we already know my status, we want to know the information for the daughter.

Joy L. Haidle: But to be honest with you, this testing strategies sometimes is in the perfect world is I'd be able to test that family member who had either a breast cancer that they were diagnosed young, they had a triple negative breast cancer, they had cancer more than once. So those are key people in a family, that a test on that person, they're informative and so their test result can provide information to a lot of people in the family. But if I'm testing somebody who is healthy, that their test result give just a little bit of information to them, a little bit of information for their children, but really isn't giving other information to, for example, siblings or aunts or uncles or cousins. So for the same healthcare dollars, if we can pick the strategy to find the best person in that family to test, more people will benefit from that test, and how we use that test result and interpret for a negative test result can be very different depending on who it was that was tested first.

Dr. Sarah Coles: That's an excellent point. It's who your test is key, right? So when somebody comes in and talks to you and says, "My family member, so-and-so," do you encourage them to try to get that family member? Do you kind of feel out what the family dynamic looks like and then kind of piece it out? How do you address that situation with people who are sitting in front of you?

Joy L. Haidle: Yeah, yeah. Really it's all of the above. So I talked through what the ideal situation would be. If we could get these relatives to test, this is the best strategy and why that result would be more useful. So if I'm testing the mother who had had breast cancer young and her test is negative, well, it's telling me that testing the daughter is not going to have a lot of usefulness unless there's something coming from her father's side of the family because the mom's test was negative, but yet she had gotten breast cancer at a young age anyway. So to test the daughter's really not going to be helpful. We need to offer surveillance based on what that family history was instead. That can be a harder concept to understand, but I think once we talk people through that and
throughout this and talk them through the strategy and the why's, then that becomes a little bit more tangible.

Joy L. Haidle: If they are willing to not have their blood drawn that day, to talk to those relatives to see what could happen with the testing, then we'll pause and we'll have them talk to the relatives. But if those relatives are willing to test and willing to share the copy of the results, that's when that person could come back in to finish the risk assessment and put that result in the context of their personal situation, but if relatives have said, "No, I know there's a test out there, I really don't want to do this." We need to respect their personal preferences and then the original person asking the question then becomes a useful candidate to pursue testing, understanding the limitations of what we understand with a negative test result. It's always easier when we find something on the test. It's when we don't that it's a little bit more of a nuanced risk assessment and that's where it's helpful to visit with a genetic counselor or another healthcare provider that has expertise in genetics.

Dr. Sarah Coles: You're right, because we do think a lot about those false positives and what that means to get a positive test result just in general and how that can be anxiety producing, but there's also the risk of false negatives where this doesn't mean you'll never get breast cancer or this doesn't mean your risk is zero, right?

Joy L. Haidle: Right, yes.

Dr. Sarah Coles: It doesn't even necessarily mean your risk is average. We have to look at all of these other pieces that go in. So that's a concern with direct to consumer is I wonder if people take this test and they're like, "I'm great. I'm fine. I don't need to get my mammogram. Everything's okay."

Joy L. Haidle: Yeah. I think there is a purpose for both kinds of tests. It really depends on what the goal was and what someone was hoping to answer with the test. So for those that do wish to do the direct to consumer testing, I think it's fine, as long as you're very clear and you've done your research about what is this test going to tell me, what really is it not telling me and when should I actually involve my healthcare provider with some of these conversations in those decisions.

Dr. Sarah Coles: Yeah. The AAFP policy is that you should always involve your healthcare provider in these decisions because it is just so nuanced and so complicated. It's certainly not our job to tell people not to do something, but we just want to make sure that they're making an informed choice and they know what to do with the results that they get, either positive or negative.
Joy L. Haidle: I agree. Yeah. As a genetic counselor, we'll support that process regardless of how you came about it to come to our office. We're happy to be a resource just like I'm sure you are too.

Dr. Sarah Coles: Oh, of course. I have to say just personally how much I really love working with genetics counselors and what a true benefit they have. I take care of lots of people of lots of ages, including pregnant women, women who are concerned about cancer risks, just all sorts of folks. It's so wonderful to have these experts who can help kind of figure out these really complicated issues and also help kind of piece out family histories and some of these more nuanced or difficult social dynamics that can come to play in thinking about genetics and what are the risks of genetic testing. So it's been, I think, it's been a real privilege for us to have this opportunity to work with genetics counselors. We go so far as to have all of our family medicine residents in my practice actually rotate with a genetics counselor to learn some skills because it is so important nowadays.

Joy L. Haidle: Well, obviously as a genetic counselor that is music to my ears. But at the same point, it is good to have a really well functioning healthcare team and that the genetic counselors play, we come in and we work side by side with our physician colleagues. So we appreciate that feedback and certainly would want to keep that relationship strong. I was just going to ask you, if you had one key takeaway message that you'd want our listeners to take from today's conversation, what would that be?

Dr. Sarah Coles: I think if I had one key takeaway message from this conversation to consumers who are considering a direct to consumer genetic testing is to really do your homework before you purchase or participate in some of these tests. Talk to your healthcare provider and if you're unwilling to do that, make sure you really read the fine print. Look at what is being tested, what is the risks and benefits of that test and how you might use that information to make healthcare decisions. If you're unsure, go talk to your primary care doctor or talk to a genetics counselor to figure out whether or not this is a good test for you.

Joy L. Haidle: I love that. I would piggyback onto that by just adding if you went ahead with testing and you found yourself, you found a test result that you really weren't certain if that was important or not, or there was a mutation that was identified, by all means, reach out to your physician and to a genetic counselor. We're still here to be a resource, even if we weren't involved at the first step of ordering the test.
Host: Thank you for listening to this episode of Genetic Counselors and to You. For more information about genetic counselors and to access tools and resources mentioned in this episode, visit aboutgeneticcounselors.com.