Erica: Welcome to the first episode of Genetic Counselors and You, a podcast by genetic counselors that answers your genetic questions. This season, you’ll hear from leaders in the genetic counseling community as they provide you with information to help you make informed decisions related to your genetic health. On today’s episode, we’re taking it back to the beginning by explaining who genetic counselors are, what genetic counselors do, and why you might see one. My name is Erica Ramos. I was the 2018 President of the National Society of Genetic Counselors, which is the leading professional organization of genetic counselors and I’m really excited to be here today with our 2019 president, my friend, Amy Sturm. Welcome, Amy.

Amy: Thanks so much, Erica. It’s awesome to be here with you today.

Erica: Well, can you get started by telling people a little bit about what you do and what excites you about being a genetic counselor?

Amy: I would love to. In my current role, I work with a team of genetic counselors in a genomic screening and counseling program where we are delivering back to patients and their healthcare providers medically actionable results for genetic conditions that can give risk for common conditions like breast cancer, colon cancer, heart attack, arrhythmias, and other conditions that can be inherited in families. We are returning this information back into the medical system and back to our patients and their families so that they can integrate this into their health care and hopefully prevent the conditions that may have been running in their family for previous generations. Really the goal is to give this information to individuals so they can act on it, prevent genetic conditions in them, and really use this knowledge as power.

Erica: That sounds great. One of your big areas of focus is on preventing disease. What do other genetic counselors do? What types of areas and patients do they work in?

Amy: It’s really pretty amazing to see how far the genetic counseling profession has come. Really much of genetic counseling originated working with the prenatal setting and working with individuals who were planning pregnancies, or who may already be
pregnant and perhaps some type of genetic risk factor was identified in their pregnancy or in their family history. That is still a really important part of the care that genetic counselors provide, but over the past 40 years, our profession has truly integrated in so many different specialties. For example, I've been working in the field of cardiovascular or heart disease genetic counseling really for about 15 years, working with patients who have strong family history of heart disease. One example is a very common condition called familial hypercholesterolemia, so kind of a mouthful, but we like to call FH for short, which is a very, very common condition that most people, in fact, 90% of people with the condition don't realize they have.

Amy: This is a huge deal because this condition, if you know about it and if you can treat it at young enough ages, and it's treated by taking medication, you can completely prevent the heart disease that happens from this genetic condition in yourself and in your children.

Amy: You've heard me talk a little bit about the areas that I've mainly worked in, being cardiovascular genetics, but genetic counselors have really integrated across the healthcare spectrum. One great example is cancer genetics. Almost every single type of cancer can have a hereditary or genetic component. Some of the common ones are of course breast cancer and colon cancer, but other types of cancers: uterine cancer, ovarian cancer, kidney cancer, prostate cancer, many different types of cancer can run in families, can be genetic. There is genetic counseling and genetic testing available that can help identify what exactly is going on in the family, what risks are being passed down, what can individuals who are at increased high risk for cancer in the family do about it. That's just been a huge area of expansion of genetic counseling.

Erica: It sounds like almost anybody could potentially encounter a genetic counselor, which means that we do a lot of different things. How do counselors get trained and how do we learn to address all the different things that we do?

Amy: Genetic counselors come into the field by going to graduate school and getting a master's degree in genetic counseling. The training that we undergo is a full - typically two years - of all types of coursework related to genetics, genetic conditions, genetic testing, genetic risk assessment, and then we also get a lot of training on our counseling skills and psychology. All of the different graduate programs also include a lot of different clinical rotations, and so that's really great because that's when you get exposed to so many different types of clinical specialties, whether it be cancer genetics, or the heart disease genetics, or pediatric genetics - working with children with genetic conditions and their families - you really get a really full in-depth training working across the spectrum of all of the different genetic conditions.
Amy: But really over time many people in our profession have become really specialized, because one of the really exciting things about genetic counseling - and I think what we all love about this profession so much - is how quickly it changes. We really do have to be kind of these lifelong learners to keep on top of this cutting edge field, and so many of us have become very specialized. So for a patient who wants to come in and see a genetic counselor, you might even schedule with someone who is specifically a cancer genetic counselor, or a cardiovascular genetic counselor, or a prenatal genetic counselor depending on why you’re coming in and what your main questions are at that point in your life.

Erica: Is the counseling part of our relationship with patients what you really think makes us different from those other types of appointments?

Amy: I’m so glad you asked that because it truly is. Over the course of my career, I’ve been able to work with so many families who really need that extra support from a health care provider who understands their entire family dynamic, who understands what they and their family have been through living with this condition or living with the fear of potentially being at risk for something that has run in their family. Help them even think through how do they talk about this with their family, sometimes these can be very difficult conversations and we are the health care provider who is uniquely trained to not only communicate the science and the medical issues behind the conditions, but also provide the counseling to them to help them adjust to this condition to make sure that they are able to integrate this genetic risk into their life, not be overcome by it, but really adapt to it and continue to live on and use it for the best way for them and their family.

Erica: I think all of us care a lot about making sure that patients and their families can really understand the information that we’re sharing with them and then use it not only in their care but how also making sure that it of works well and integrates into the rest of their lives. Because this is such important information, when is the best time for people to talk to a genetic counselor?

Amy: You know, there are so many times really across the lifespan that someone might want to interact with a genetic counselor. It might be when you are deciding to potentially have children with a partner and you want to find out if you and your partner carry any genetic conditions that future children could be at risk for. It might be when you have figured out that you’ve collected family history information and you’ve always kind of wondered, "Hey, why did my aunt pass away at a young age?" and you get records on the condition and you want to go seek out, could I potentially be at risk for this? It could be when you undergo a new diagnosis and maybe you've been diagnosed with breast cancer or another type of condition at a young age and you're trying to figure out, "Hey, is this
genetic? Is this something that could give me risk for more cancers? Is this something that my children, my daughters and sons could potentially inherit?“.

Amy: There are many, many more examples like that, so really across the lifespan, there are many reasons that someone might want to go see a genetic counselor and sit down and have a consultation with a genetic counselor. It could also be once you've undergone genetic testing or if you're considering wanting to undergo genetic testing and really wanting to figure out, "Hey, what can genetic testing tell me? What can it not tell me? What's the right genetic test for me?" Or maybe you've even undergone some genetic testing and you want some help figuring out what the results mean for you and what they could mean for your family. So, there are many different examples and you know you might even go on to see more than one genetic counselor over the course of your lifetime depending on where you're at in your life and kind of what's going on with you at that point in time.

Erica: I'm glad you talked about genetic testing, too, because one of the things I think all of us are seeing a lot pop up on TV is genetic test information for ancestry and for other types of health things. Is that something that genetic counselors work with too?

Amy: It sure is and it's been really exciting to see more genetic counselors be available to talk to really the millions of people who are so interested in learning more about their ancestry, and their genetic history, and their genetic risks potentially for certain conditions. It's opening up a whole new group of individuals to really learn more about genetics, learn what type of information it can give to you, and then the genetic counselor can really supplement that type of information with the clinical and medical information that can be brought into the picture to say, "Okay, well you learn that you're from Norway, well what do we want to learn in your family history or your medical history that could also be brought into the full picture for your genetic risk and your genetic health?".

Erica: Yeah, it's really interesting cause I think so many people, even genetic counselors, get really interested in this type of testing. I know several of us have had genetic testing for things like ancestry and it's really fun to find out that information, but it's great to know that genetic counselors are there to make sure that when you find something that could have medical implications or could make a difference in your medical care, that there's a good resource out there for you. With all of those different types of people who might want to seek out a genetic counselor, for people who may be listening who want to know what that experience would look like, what can people expect from a genetic counseling appointment?
Really what's covered in the genetic counseling session is based on you. We make our approaches extremely individualized to make sure that what your concerns are is what we will address. Now, I will say there's kind of some main things that we always will cover with patients when they come in because we want to make sure we have all of the full information we need for the proper risk assessment. We typically start with things like your medical history and your family history, and we really like to look at your family history as far as even three - if not more - generations back. It's really good for you to try to investigate your family history information, talk to relatives, get as much family history information as you can [prior to your appointment].

But that being said, we definitely have situations where individuals who have limited to no family history information come in for genetic counseling. Then depending on what type of information we have available, what your main questions or concerns are, we can review any risks that we might see in your medical history, in your family history, or talk about what genetic tests might be helpful to you, helpful for explaining what's going on with you or helpful to figure out what could happen for you in the future, or for your children, or other generations in your family.

And so there's really a lot of different things that could happen during the genetic counseling appointment, but the main things are really focusing in on you as an individual, looking at your family history, and then if genetic testing is an option, talking through that, talking through the types of results. Then we always follow up with patients, whether it be one time or a few times afterwards, that we're able to work with you to integrate that genetic information back into your care, back into your life, back into your family.

We also work very closely with your health care provider team. One thing I've done over the course of the years working as a genetic counselor is worked directly with my patient's primary care providers. Work with their cardiologists, work with their other types of doctors and nurses so that we can really all be on the same page as a team for you to take care of you when it comes to any of your genetic health concerns.

It sounds like in getting ready for a genetic counseling appointment, the three things that people can do to really prepare are first think about what questions they have and what they might be worried about, think about what they're hoping to learn from a genetic counselor or from genetic testing, or information, and then also really investigate their personal and family history and think about how that might be influencing the conditions that they're worried about.
Amy: Definitely. When we sit down and kind of come together with that first meeting, we will review the questions and concerns, and make sure we're on the same page, and then kind of dig into that information to figure out the next steps we need to take to address the questions and concerns.

Erica: I think every genetic counselor has had to answer some strange questions about what we do and what we don't do. There's a lot of myths about genetics, and we see a lot of Sci-Fi movies, and things like that that talk about genetics and where we're headed about genetic testing. What do people not have to worry about when they see a genetic counselor? What don't we do?

Amy: Well, I think this is a great question. We don't clone you. We don't make designer babies. We don't make you do anything you don't want to do. We don't make you have genetic testing. We just talk about your options. We talk about the way that genetic information could benefit you and that's really what it's all about.

Erica: That last point is really important because I suspect there are a lot of people, maybe even some of the people listening, who are really concerned that they're going to have to do a genetic test or have to find information that they may not want and decide not to come see a genetic counselor. I think one of the biggest things that we want people to know is that genetic counseling without genetic testing can also be very helpful, just having somebody who can talk to you about all of the conditions in your family, all of the things you're concerned about, and the best way for you to move forward can be a really empowering experience.

Amy: Absolutely. It's been interesting working with families over the years where you can see sisters, mother and son, any grouping of different relatives within a very close knit family really think about their options so differently. I remember a mother daughter pair that I was working with and we were talking about a type of genetic tests that can look at almost everything in your DNA and come back and give you information. Not only about a specific disease that might be running in your family, but can also screen you for other conditions like cancer and heart disease. This is something called whole exome sequencing where really the entire DNA code is sequenced and can look at the genes that we currently know about that are tied to different genetic conditions, and a piece of that process is asking patients, "Well, if something is found that can also come back and tell you incidentally that you might also have an increased risk for, for example, breast cancer, would you want to know that?"

Amy: I remember the daughter who was probably in her thirties at the time felt very strongly, "Yes, I would definitely want to know that. I want all of the results. I want to know..."
everything I'm at risk for." Her mother had a much different reaction and she kind of sat back and said, "I'm not so sure, you know, I don't know if I would want that information, and what would the impact of that beyond me, and my care, and my health insurance. And even kind of my psychological response. What would that be to kind of know that I have this risk." That's the exact types of situations that we help individuals through, and every single person is different and how they think about this, and so we can kind of walk down the different paths.

Amy: Well, you know, if you want to get the information, here's what that might look like, and here are the types of things you would want to think about if you do get that information, and here's how it could be empowering to you to know that information. But for someone who has more concerns, to really listen to them and talk through all of those concerns, and where we can be reassuring, provide that reassurance, but where perhaps there might be some unanswered questions, really talk through that. Let people have the time to figure out how do they want to receive genetic information, what do they want to learn about themselves, what don't they want to learn about themselves? We always are there for our patients as resources and every single approach, again, can't stress this enough, is just completely unique to what that person's needs are.

Erica: When people have differing opinions within the family, genetic counselors are people who can help them figure out what the best path forward is for each individual and also for the family then. What other types of ongoing support do genetic counselors offer to patients?

Amy: You know, we're really there as a lifelong support for our patients and it might be something where individuals get a genetic test result that might be something unclear and where we might not completely know all of the important information about it at that point in time, and so oftentimes when we have kind of one of these results that can be described as in the gray zone, we tell our patients, "Check back in with us every six months to a year. We will continue to research this for you. We will continue to look into this for you and what it means for your health, what it means for your risk."

Amy: We're also there for patients as they continue to try to integrate new information into their care and into their lives. We are also there to help them continue to deal with different feelings and emotions that they may go through as part of the genetic testing or genetic evaluation process. I've had some patients who have had to deal with grief and loss, and whether this be due to losing a close one, due to something like a sudden heart attack or an early diagnosis of cancer, or some other type of genetic condition that's been in their family, and maybe they need several consultations with a genetic counselor to really talk through their grief and to adjust to that information. Also, genetic counselors

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do coordinate a lot of care and help patients really access all of the different resources they need.

**Erica:** So whether somebody needs to see a genetic counselor just once or ends up having many appointments with the genetic counselor, what's the best way for people to find that person to work with?

**Amy:** There are multiple resources, but I think the main one that I would recommend is the findageneticcounselor.com website. This can be a really easy tool where you can log on, you can put in your zip code, and you can basically specify do you want to meet with someone in person or would you prefer telephone? Then you plug that in and you can get a list of different genetic counselors who would be able to see you for an appointment.

**Erica:** That's great. And working with your physicians, they may also know genetic counselors in your area, especially if you want to sit down with a genetic counselor who they've worked with in the past and who they may have a good relationship with. What about insurance? A lot of people wonder if genetic counseling and genetic counselor time is paid for by insurance?

**Amy:** Great question. I think the best recommendation is that once you get an appointment with your genetic counselor, you can talk with their clinic directly, have them walk you through the process to check with your insurance and whether your insurance can be used to pay for that visit. Oftentimes insurance will pay for your genetic counseling visit and it might just be something like a copay like when you pay to go see another type of specialist.

**Erica:** We've covered a lot of ground today and hopefully we've answered some important questions for some of the people listening and got I think our podcast series off to a great start. One question that I want to ask you Amy to wrap everything up, why did you become a genetic counselor?

**Amy:** I actually went into research at first and I was working in a laboratory. For me, I realized very quickly that I wanted to have a connection with patients. I wanted to be seeing how my work was directly influencing people, and people's lives and impacting health directly today. Very luckily, I learned about the field of genetic counseling and immediately I had an ah-hah moment like, "Where had this been my whole life? I've been waiting for this career!" It really is that perfect mix of science, but also working with patients, working with people, and seeing the direct impact you can have on individual's lives is just a really, truly amazing. I think for anyone who loves science but who also just loves working with people, this is the career for you.
Erica: I couldn't agree more. Thank you so much for your passion and sharing your enthusiasm for genetic counseling, and everything we do, and taking the time today to talk a little bit more about what patients might want to know.

Amy: Well, thanks Erica. It's been great fun sitting here with you and looking forward to hearing more from the podcast series.

Host: Thank you for listening to this episode of Genetic Counselors and You. For more information about genetic counselors and to access tools and resources mentioned in this episode, visit aboutgeneticcounselors.com.