

Genetic Counselor Interview

Madeleine Jepsen



(Introduction: Nancie Petrucelli is a genetic counselor with the Karmanos Cancer Institute in Detroit, Michigan. She specializes in genetic counseling related to cancer, and also serves as an associate professor of oncology at Wayne State University School of Medicine. She holds a master's degree in genetic counseling from the University of Cincinnati.)

You mentioned that you had wanted to be a veterinarian for a really long time. How did you find your way to becoming a genetic counselor?

I thought I wanted to be a veterinarian, so I declared my biology major shortly after arriving at Hillsdale and really focused my attention on biology and my goal to become a veterinarian. But when I took genetics, I was just really fascinated by genetics. At the same time I was also taking psychology 101 and loved that class equally as much. So it became apparent to me that I at that point really was interested in finding a profession that was a marriage of those two disciplines. The professor had introduced me to the field of genetic counseling, which I had never heard of at that time, and I ended up doing my thesis work on sex chromosome anomalies, which is related to genetic conditions and genetic counseling. So that's how I became aware of the profession and interested, but it was really genetics and psychology, taking those two courses really was a lightbulb moment for me. I was passionate about those two disciplines, and I wanted to work in a field that would combine them.

You found out about it through the professor who introduced you to the class, but are there any other ways to get to the field of genetic counseling?

I know some efforts we take — I'm associated with the Wayne State Genetic Counseling program — and some of the steps we take to try and expose students to this field perhaps at a younger age than college is through talks at local high schools to try and introduce the field to them. There are some efforts to educate local students in high school about the field of genetic counseling.

The program here, they have open houses where students can come visit and learn more about the program. I'm not sure if other career paths would necessarily introduce someone to genetic counseling. I feel like it was just serendipitous myself when I was exposed to it through Professor Platt because at that time, it was a relatively new field.

There's a national society of genetic counselors, so there's a national organization, and they have a very helpful website that has information about the career and the field and the resources, and genetic counselors that you can contact who live locally to you to reach out and learn more about the field.

You're also a professor — what is it like to balance the teaching and the counseling?

Well, right now I'm pulling my hair out because I'm teaching right now. It is a little bit of a difficult balance. Luckily for me, I do have the rank of professor, but that's not my full-time job. My full-time job is providing counseling to patients and families. My involvement in teaching is just sprinkled throughout the year, and right now I'm involved in a course that I teach at this time every year. Luckily, I have colleagues that can help balance out the clinical responsibilities that I have, and cover more of the clinic while I'm teaching.

It is difficult to juggle those things, but I also enjoy that aspect of my work. I like the diversity of not only seeing patients but teaching and having exposure to students who are going into the field of genetic counseling. I often learn just as much from them as they do from me. So that's a very gratifying experience that I have with teaching. It gets crazy, but I enjoy it, and luckily I have good support here who can help me balance some of my other responsibilities while I'm actively teaching.

What classes do you teach?

The one I'm teaching right now is called Practical Applications for Genetic Counseling, and it's for second-year genetic counseling graduate students. It's a two-year master's program for genetic counseling, and they'll shortly begin their clinical rotations, where they go and spend about seven weeks in a clinic, and they spend time in the various specialties and they rotate.

This course is designed to provide a good foundation for these second-year students, so once they get to their clinicals they can hit the ground running and start to see patients pretty soon after arriving. I'm a co-director with two other directors of this course, and we each split it up — I have the cancer section. I lecture over the course of a month, four different times. My first lecture is all about cancer terminology and statistics and making the diagnosis of cancer and what chemotherapy is what radiation is because quite honestly, when you're in genetic counseling, you're learning genetic terminology and modes of inheritance and those types of things. Cancer terminology is really a foreign language for many genetic students, so I spend my first lecture just educating them about the terminology and treatment and what all that means. The second lecture that I give is all about pathology and interpreting pathology reports for a cancer diagnosis because there are some clues in the pathology report that can indicate to us that the cancer might be hereditary in nature. We spend that whole second session on pathology reports and what to look for.

The third lecture is taking a detailed personal and family history and ultimately how to assess that family history to determine whether or not your patient might be at risk for hereditary cancer. Only 5-10% of cancer cases are the the type that run in families. We're kind of like detectives in a sense. When patients come to our clinic, we're trying to figure out whether or not they are at risk for the type that is inherited. So many of them are sporadic, and there's an art to assessing that history. SO my third lecture is all about risk assessment, what clues to be looking for, what questions to ask while taking the family history, and putting it all together in a sense to provide your patient with an estimate of whether cancer in the family might be running through their genes.

I also teach an advanced medical genetics course. It's just one lecture that I give on hereditary cancer syndrome, so it's going through various cancer syndromes and lecturing on those.

On an annual basis, I give a lecture at Madonna University for their nursing program on genetics. I've been doing that about ten or fifteen years now.

What kinds of situations merit genetic counseling?

In general, most cancers are sporadic, so as I mentioned, we're detectives trying to figure out when the cancer could be hereditary, and that's based on some red flags we might see in the family.

There are some general red flags we look for, and it depends on what kinds of cancers we're seeing in the family that may dictate another specific set of red flags we might look for. In general, what gets us concerned is if an individual has several relatives who have developed the same type of cancer. If there's more than one relative that's developed breast or colon or thyroid or kidney — whatever type of cancer it is, seeing it in more than one relative starts to suggest a possible pattern.

We also look for age of onset. That's really important. One in three of us will develop some kind of cancer in our lives, and those are normally the sporadic ones. Those are just as we age, our bodies become more susceptible to cancer. So it's not uncommon to see people developing cancer in their 60s, 70s, 80s, maybe in their mid to late 50s, but when we start to see cancers like breast and colon which typically are diagnosed when we're older, and we see those developing in individuals who are in their 20s, 30s or 40s, that starts to make us suspicious as well.

There can also be some rare cancers or unusual cancers that in and of themselves. For example, male breast cancer. It's pretty rare in the general population, so we don't see it very often, but it has been linked to a number of cancer genes. So when we see that type of cancer in a family, even if there's no other cancer going on, we get highly suspicious.

Another type of cancer, similar to that would be ovarian cancer in women, that is highly suggestive of something hereditary.

We also look for the same kinds of cancer in relatives, but we also look for related cancers. Normally patients do not come in knowing what a related cancer would be — that's where our expertise comes in. Many of these cancer syndromes predispose to a spectrum of cancers. Examples would be breast and ovarian. Those

are related and have been related to a number of hereditary cancer conditions. Another example would be something like colon and uterine cancer. Those two have been linked to another hereditary cancer syndrome.

So we're looking for red flags in the family, the ones I just mentioned, and that kind of gives us a sense of whether these cancers are just occurring coincidentally in the family, or if it is possible that there is a genetic link causing the pattern we're seeing.

Do you mainly deal with cancer cases, or are there other hereditary diseases you deal with as well?

I specialize in cancer genetics, but there are genetic counselors who work in a prenatal setting, so they may be counseling couples who are currently pregnant who maybe have a family history of cystic fibrosis, and they're concerned for their child or offspring or fetus. There are pediatric genetic counselors who may see a patient who is a child with multiple congenital anomalies, and the child is coming in to be evaluated to see if all these are related to some genetic component. I specialize in cancer, but there's also cardiac genetics, so there are genetic counselors focused on heart-related issues. Those are the main specialities. There are also genetic counselors working in what we call industry, so they're not working in a clinic at all, they're working for large laboratories and providing genetic counseling or really more interpretation of genetic test results. They're the genetic counselors on staff at the lab, and if someone has a question about the interpretation of a genetic test report they've received from that lab, they can call the counselor on staff there and have a conversation about that.

In your teaching and the work you're doing, you must have conversations with grad students and PhDs, but also people who don't have much background in science — do you have any advice for communicating advanced ideas to people with a variety of backgrounds?

That is one of the challenges I think genetic counselors face, and I think you naturally get better at it over time. The challenge really is to tailor and customize your conversation to meet the educational background of your patient. We use a variety of tools — I've gotten better in terms of the language choice I make, depending on my audience. Instead of using the word mutation, for example, I may use the word change. It can be just very simple changes in language you're using. We also use various visual aid tools that help illustrate the concepts we're discussing with patients, and I think that can be very helpful if someone's more of a visual learner than a verbal learner. Sometimes I have to explain things a few different ways as well because the patient may not be understanding the concept the first time around. Using analogies can be helpful. I think naturally you get better at it with experience. I work in Detroit, so I do see a variety of educational backgrounds, but we also counsel patients in some of our suburbs. When I'm counseling in a different setting, it's usually a different clientele, and they have very good questions and the level of conversation, the level of detail, is very different. I try and keep it simple for people who

may have difficulty understanding, but if people are asking very specific questions, I'm able to have a very different conversation — ultimately conveying the same information, but maybe at a different level of detail.

Are there any common misconceptions that people have about what you do as a genetic counselor?

Well, I think one of them, at least in the prenatal realm, which I don't work in, but the idea of eugenics, or that genetic counselors are proponents of pregnancy termination. I think this has been one of the biggest misconceptions about what we do.

Our job as genetic counselors is to promote our patients to make informed decisions and make the best decision for them. Our job is not to impress upon them our own biases or beliefs. That can be challenging for some people that have very strong beliefs one way or the other, but genetic counseling really isn't about the genetic counselor, it's about the patient. Our job, again, is to arm them with information so that they can make the best choice for them. I think that's one of the biggest misconceptions about what we do.

In cancer, and I think this is a little bit more minor, but one of the biggest misconceptions when we're talking about female-related cancers — ovarian, breast — there's a big misconception out there that men cannot carry and transmit these genetic mutations, that it's only the maternal side of the family that matters. That's a complete misconception. Most of these conditions are autosomal dominant, and that means men and women have an equal chance of carrying and transmitting these genetic mutations to their children. The paternal family history is just as important as the maternal family history.

Are there any state certifications that you need to obtain to be a genetic counselor?

You do go to school, and there are accredited programs around the country — I think 32. Once you've completed your masters, you do have to sit for a national certification exam to become a board-certified genetic counselor. There are nurses, even physicians, that provide genetic testing to their patient. They are not genetic counselors though by training, and are not usually certified in genetics either.

Some states also have licensure, where you are a licensed genetic counselor. I think there are maybe 12 or 18 states that have licensure. That's not the case in Michigan, although we're working toward it.

What would you say is the most challenging aspect of your job?

I think one of the most challenging, but is also one of the things I love about it the most, is that it is a very rapidly-evolving, extremely dynamic field. I'm committed to lifelong learning, and I feel like this profession requires you to constantly be learning. They are discovering new genes all the time, so what I was coun-

seling patients and families about just a couple of years ago has already changed because we know of more genes related to cancer. My job as the genetic counselor is also to keep abreast of all this new information. I attend national conferences and participate in webinars. A lot of continuing education goes on beyond your degree. I think in some ways, that's very challenging, but it's also one of the things I love the most about it.

I think one of the other challenging things, because you're dealing with patients and families, is to keep that boundary up. You are there to provide counseling, but it's hard not to get emotionally involved sometimes. That became apparent to me when I started counseling patients when I started to approach the age of my patient population. When I first started out, I was a lot younger than all of them, so I couldn't relate to them in terms of where they were in their life. As I started to become around the age of my patients, and I have my own children, it strikes a different chord with you when you are sitting face-to-face with someone who's your age, who has children at home, who has a stage-four diagnosis of cancer. Their cancer isn't going to be cured, they're just in a position to try and obtain helpful information for their family members. I think sometimes it can be difficult to keep that boundary and not get psychologically and emotionally attached to your patients and involved in the case. You get better at that with time, and I think it's ok to become emotionally involved sometimes — that's not necessarily a bad thing — but you don't want to become so emotionally involved that you can't provide an unbiased conversation and presentation of the information. I think that's where you have to be careful.

What are the job prospects in the field of genetic counseling?

There is a high demand for genetic counselors right now. If someone is looking for a field where you are involved in lifelong learning but also a pretty good opportunity for employment and growth, genetic counseling does offer that.