**The Uncertainty of Genetic Testing**

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counselors, including what they do and why you might see one, visit aboutgeneticcounselors.com.

**Deepti Babu:**

I'm Deepti Babu, Certified Genetic Counselor and NSGC member. A quick note that I'm very excited to

share with you. For 2020, I'll be hosting the Genetic Counselors and You podcast. So every two months,

we're continuing to bring experts in to help break down complex topics in genetics that are making

people curious, with the goal of turning that curiosity into knowledge that you can apply in your own

lives. If you have an idea for a topic that you would like us to explore, please email the show at

nsgc@nsgc.org.

**Marleah Kruzel:**

And that for me is what's really so interesting as a researcher and a patient, is that ultimately,

uncertainty is inherent in genetic testing, but it's inherent in our lives.

**Deepti Babu:**

Hi, everyone. Welcome to our next episode of the Genetic Counselors and You podcast. When it comes

to genetics and health, there are many things we know, and with such a fast moving science there are

things we don't fully understand yet.

**Deepti Babu:**

Genetic testing can help us understand and improve our health, but it can also bring about uncertainty.

Maybe it's uncertainty of whether you'll have a genetic risk factor come back on a test, for a health

problem, or maybe it's uncertainty about if that health problem will even happen, and if so, when? This

can be difficult, but there are steps that we can take to navigate and manage, and ultimately live with

this uncertainty. Given the current Coronavirus pandemic, maybe these are skills that we can apply

elsewhere outside of the realm of genetic testing.

**Deepti Babu:**

Today we're talking with Dr. Marleah Dean Kruzel. She's an Associate Professor at the University of

South Florida. Her research focuses on how communication can help health outcomes, often in the face

of uncertainty. Dr. Dean Kruzel has been through the genetic testing experience herself, and is also part

of NSGC's Digital Ambassadors program. Welcome Marleah, and thank you for joining us today.

**Marleah Kruzel:**

Thank you so much for having me. It's a pleasure.

**Deepti Babu:**

Well, I'm excited to dig into this topic with you, because I think the topic of uncertainty as it pertains to

genetic testing is one thing, but I feel like in the context of a public health pandemic and other things

that are going on in the world right now, uncertainty is something that's becoming more and more in

the forefront of our lives. Well, right off the top, I'm wondering maybe if we could define what we mean

by uncertainty, so that we have a shared understanding of it as we discuss things. How would you define

uncertainty for us?

**Marleah Kruzel:**

There are lots of different ways I think you can think about uncertainty, but for me, a helpful

conceptualization or a definition is that, uncertainty exists when we believe aspects of our situation are

ambiguous, they're complex and/or they're unpredictable. So definitely, all those facets would relate to

COVID-19 right now. In the case of genetics specifically though, uncertainty often arises when obtained

information, so like genetic test results, are unperfect or they're unknown. This can create uncertainty

about a clinical diagnosis and risk management.

**Deepti Babu:**

I think, if I think about my time in the clinic talking to patients and families, because the human genome

project completed and we have the genome sequenced, we think that we've got this all figured out. And

uncertainty, and the element of uncertainty, is surprising in the context of genetic testing. Would you

say that, that's true?

**Marleah Kruzel:**

Yeah. Absolutely. If you test positive for a pathogenic or likely pathogenic variant, you come to learn

that you are significantly at an increased risk for developing hereditary cancer in comparison to the

general population. But that doesn't necessarily mean that you're going to be diagnosed with cancer.

And then sometimes, there's also the possibility that you could test negative. If that happens, but you

still have a family history of cancer, there's lack of context. It might be confusing or overwhelming

because you expected to test positive. And then of course, just in the genetic testing technology, you

could also receive a variant of uncertain significance. Here, it looks different than what's normally

expected, but it's unclear whether the variant is harmless or has a risk factor. So in all these different

scenarios, uncertainty exists, it just exists in different ways. And that for me is what's really so

interesting as a researcher and a patient, is that, ultimately uncertainty is inherent in genetic testing, but

it's inherent in our lives. We can't get away from it.

**Deepti Babu:**

Yeah, as much as we try to.

**Marleah Kruzel:**

Yes.

**Deepti Babu:**

Yeah. And I think that sort of leads to what I wanted to talk about next, which is a bit about the genetic

testing experience that you went through. I think about seven years ago? Correct me if I'm wrong, you

went through genetic testing yourself. And just for the listeners Marleah's experience circles and your

expertise circles around cancer genetics. But much of what we're talking about, there are parallels to

other genetic diseases and conditions. But anyway, back to you, you went through genetic testing. So

you've definitely dealt with uncertainty as it relates to genetics and your personal health. Can you tell us

a little bit about your experience?

**Marleah Kruzel:**

Yes. So my mom was diagnosed with breast cancer when I was eight years old and she was the first one

in our family. So it was shocking. It was devastating really to the family. At the time, it was 1997 so she

didn't undergo genetic testing until many years later. But in my case, I actually contemplated genetic

testing for a long time before I did it. In fact for about a year, I think it was, after seeing a genetic

counselor with both my mom and my husband, the genetic test kit that my genetic counselor gave me

sat in my safe, in the back of my closet. And as I reflect back on that, I think I took such a long time to

decide because I knew if I tested positive, like my mom did, it was going to change my life. And so I

wanted to be absolutely sure that I was ready to receive those results, whatever they were.

Marleah Kruzel:

But I think, going back to uncertainty, after I tested positive for a pathogenic variant in BRCA2, I quickly

realized that my uncertainty did not go away. I think many individuals, including myself, who have a

significant family history of cancer, we undergo genetic testing to confirm what we already expect or

worry about, right? That we're also at risk for developing hereditary cancer. But while knowing one's

genetic predisposition can be empowering to make informed health decisions, undergoing genetic

testing does not necessarily decrease or alleviate cancer related uncertainty. In fact, it often produces

new forms or types of uncertainty. And then that forces us to acknowledge and therefore manage

uncertainty over time.

**Deepti Babu:**

Yeah, just for clarity. So, you talked about your mother being diagnosed with breast cancer. So she

ultimately had genetic testing, which showed that she had a change, a pathogenic variant in the BRCA2

gene, that's associated with an increased risk for breast cancer, ovarian cancer and others. So then you

were then offered the option for that testing. And you took a while to decide. That test kit sitting in the

box represented your uncertainty about whether you wanted to-

**Marleah Kruzel:**

Absolutely [crosstalk 00:08:36].

**Deepti Babu:**

Or not. And you finally felt comfortable enough, I guess, that you had explored all the different ways in

which you would receive that information and decided to go for it. And so when you talk about, that the

uncertainty doesn't go away, the uncertainty then it transforms into something else then I guess. When

you get the result saying, okay, in fact you have this genetic risk factor. Now you're moving into another

type of uncertainty.

**Marleah Kruzel:**

Yeah. Before you undergo genetic testing, you're wondering if you have this change, which would put

you at risk for hereditary cancer. So it's a maybe, depending on what your family tree looks like, it might

influence you more or less. But once you test positive, it brings up new uncertainties. Now you're

dealing with, okay I have this result, I know my genetic test results and I have an increased risk, but it's

not for sure that I'm actually going to be diagnosed with cancer. It's just a lifetime risk. And so the very

nature of testing positive, from my perspective, and a lot of what my research focuses on, is you become

certain about the uncertainty. So before you're uncertain about if you have it and then you test positive,

and now you're certain about the uncertainty that you had originally. And so then you have to start

coping with that in different ways.

**Deepti Babu:**

Wow. That's kind of making my head explode thinking about it, seriously. But I haven't heard it

described that way before. So, when you begin to shift from you're certain about your uncertainty, and

then you have to become comfortable with that, what does that look like?

**Deepti Babu:**

I'm imagining it's an evolution, it's a process there's twists, turns, pivots, life happens. You're also...

You're doing things to reduce that risk is, hopefully, the opportunity that the genetic test results give

you, a chance to get in front of things.

**Marleah Kruzel:**

Yeah. I think, well you really have to take it one day at a time honestly. And it really depends on the

person. Some people might try to reduce, to use that term, reduce the uncertainty as much as possible.

And then other people react in different ways, and so they're more managing uncertainty in the sense

that, well some days they might just be okay with the uncertainty they're experiencing and then other

days they really need to reduce it. And other days maybe they'll actually enhance the uncertainty to

make them feel better. So I think it does depend on the person. And ultimately just over time, you have

to figure out how you work, how you think, what makes you feel comfortable, drawing on lots of

support from others can be helpful.

**Deepti Babu:**

Can you tell us a little bit about how you managed during that process? And then living with that

uncertainty today?

**Marleah Kruzel:**

Yeah. That's a good question. I definitely would say from my perspective, it's managing or living with the

uncertainty. It's definitely... I'm not able to reduce it completely. In general though, there's a few things

that I do to try and cope with my uncertainty. The first is I really try to tap into this part of my identity,

my family history, the connection between my genes and my family, and really try to help others while

continuing to challenge myself. So in a different way, my research really helps me cope with my own

uncertain future because ultimately what I'm trying to do is to improve patients and families health

experiences, by developing different types of communication tools to improve health outcomes. So that

helps me cope in one way.

**Marleah Kruzel:**

Another thing that's helpful, as a professor I also teach. And so anytime I teach a class I'm always trying

to teach my students uncertainty management skills. So that could look like teaching them how to

effectively seek credible, reliable health information. When we're turning to Dr. Google, for example. It's

helping them learn how to identify the source of uncertainty that they're experiencing and then

acknowledging that not running from it. And what does that look like?

**Marleah Kruzel:**

And then ultimately learning and teaching them how to self manage their health journey. I think, so

much of a patient's experience happens outside the clinical encounter, and we're never taught how to

be patients. Healthcare providers, genetic counselors, physicians, they're taught for years how to be

experts on this particular subject, but we're patients all our lives. And so I really try to help my students,

giving them specific skills.

**Marleah Kruzel:**

I think probably one of the most important things I try to draw on when I'm trying to manage the

uncertainty in my own life is, I'm frequently reminding myself that I'm never going to have all the

necessary information to make a decision. That's just never going to happen. And so somehow

acknowledging that makes me feel better. And then I can ultimately... I try to embrace the uncertainty

really. And I didn't really realize this, now looking back though, when I finally decided to undergo genetic

testing and take that genetic test kit out of my safe, in the back of my closet. My mom said this phrase,

which has stuck with me so much, and I tell my patients and my students a lot, which is "you make the

best decision you can with the information you have at that time."

**Deepti Babu:**

That's pretty powerful. It just gives you permission, right? Forgiving yourself that it's not perfect. And

like you said, it's so contextual and it's so dynamic. So, a day later you might have different information.

You might feel differently about it. Kind of going back to that model about the uncertainty that you were

talking about, where sometimes you reduce it. I might have said compartmentalize it, but reduce it is a

better description. Because you just kind of want to get it out of your face. And then sometimes you

want to let that come towards you a bit more and embrace it. Did you find a particular moment through

that experience that led you to your decision to pursue a PhD?

**Marleah Kruzel:**

Actually, really the pursuit of my PhD goes all the way back to my early childhood experiences. Living

in... Or at least what it felt like living in hospitals as an eight year old, watching my mother undergo

chemotherapy, radiation, reconstruction, breast surgery, and then all the side effects that came with

that. Even though I was eight, I knew what was going on. And I tried to be involved as much as they

would let me as an eight year old, because that was how I cope with things is I want to know what's

going on. I want to know the information and be a part of it. So, that's really why I decided to go into the

field of health communication. And then the main reason why I decided to become a professor, going

further was really to give back to the medical community who I feel like really saved my mom's life in a

variety of different ways.

**Deepti Babu:**

Right. Yeah. And I think that that is certainly one reaction. And then I definitely know there are people

who more information is definitely not something they want. Those might be the individuals who never

really want to pursue genetic testing, that maybe that's a reason, they just don't really want the

answers. And so sometimes it's easier if we just don't ask the questions. I guess you've talked about this

a little bit already. What have you learned through your research about how people cope? And I think

what I'm hearing is just, there's so many different ways and there isn't one right way or one wrong way

and your way can change. [crosstalk 00:16:50]

**Deepti Babu:**

I remember even for myself early on, when I was younger and had fewer dependents, so to speak, in my

world, I did not really want to ask too many questions. I wasn't... And I didn't even know the questions

to ask. And now I feel like more information does help me. It helps me make decisions that impact other

people. So are there common experiences or moments that you're seeing though, even in all these

different stories, or ways that you're researching how people communicate about health?

**Marleah Kruzel:**

Yeah. Sometimes we avoid. Because I think that's often... Before I answer your question, I'll just say,

avoidance is an effective coping mechanism. It's one that has consequences, right? So if we avoid

information such as not wanting to undergo genetic testing because of trauma... But that is a legitimate,

that's an effective way to cope. Now the impacts of that is what then we, as patients and genetic

counselors have to deal with and try to figure out how to support the patient, but also be careful with

what could happen to their health. But in answer to your question, really the most common experience

is to seek information. So though some do avoid information, most seek information. That's an effective

communication uncertainty management strategy. So for example, in the case of previvers, so people

like me who they've tested positive for a pathogenic variant, but they haven't been diagnosed with

cancer.

**Marleah Kruzel:**

We tend to seek information by turning to different types of sources. So our healthcare providers as

being probably the number one source, and then also cancer related nonprofits. And then lastly, the

internet and in particular, Facebook and Twitter, these are all different sources of information. And we

can get information, which then enables us to feel better by seeking, by organizing by then utilizing the

health information that we find or that we learn. It helps us make that informed health decision,

whatever it is, and ultimately making that decision I have found helps previvers feel empowered. They

feel in some, I will preface it, in some control of their genetic health risk.

**Deepti Babu:**

Yeah. I think you're getting into the nice takeaways that we want to leave people with is there's the kind

of professional organizations, there's your healthcare provider. And then there's the internet. And I

think coupled with that, and this is what I would tell families, is really placing a very strong filter and pin

your brain there as much as you can, because not everything that you read is accurate and true. But I

think using those resources to find trustworthy places, such as, bouncing something off your healthcare

provider, your doctor, your genetic counselor, or other family members who have gotten a great

curated list of something already, I find they can be really great resources.

**Marleah Kruzel:**

And I think you made a really good point is that, ultimately you want to turn to the experts, right? So if

we're thinking about genetics, I mean genetic counselors, they can really... They helped me, my genetic

counselor helped me. Even though it took some time to make a decision, but she really did help me

come to terms with the certainties and the uncertainties really that arise before, during and after the

genetic testing process. But we also, again, have to keep in mind that patients aren't just going to see

healthcare providers as their main source because our lives exist outside of the clinical encounter. So as

much as possible when you turn to other types of information sources bouncing that back to your

healthcare provider, sharing what you found, it can be really helpful to help them interpret the

information and tailor it to your specific experience.

**Deepti Babu:**

And this whole concept of becoming comfortable with uncertainty. I mean, that gives me discomfort.

I'm just going to be really honest-

**Marleah Kruzel:**

You're in good company.

**Deepti Babu:**

Having said that. So I'm feeling that a little bit. I think people are feeling that a little bit more right now

with the pandemic layered on top of everything else. And I think they're very important skills and tools

that you have probably taught your students. And you've done your research on that we can use and

apply in many different ways. Some a little more often now than at other times. So you've described

gathering information. Can you think of some other ways that we can become more comfortable as you

articulated earlier about living with that uncertainty? I think, I just had to come to terms with the fact

that there's never going to be a perfect time for this, or enough information for me to finally say I have

hit my threshold. I'm ready.

**Marleah Kruzel:**

Yeah, it's a great question. And one I'm really not an expert on still even given my experiences because

it's something I'm continually struggling with, but I kind of pushed through. I think uncertainty, it

changes over time. So it kind of ebbs and flows. And so I think first off, we're thinking about trying to

manage longterm uncertainty, whether we're talking about someone like me, who's at risk for

hereditary cancer, or just thinking about COVID-19 and the effects that that could have on you and your

family. So this is a longterm process.

**Marleah Kruzel:**

So I think acknowledging that, identifying the type of uncertainty. So is it medical, uncertainty you're

experiencing? Is it familial uncertainty that you're experiencing? Is it personal? Is it financial? There are

different types. And without going into all theory on you, there are different types of uncertainty. And

so if we can identify what we're experiencing, that is a good way to then figure out, okay what do I need

to address it?

**Marleah Kruzel:**

Is it talking to my mom about it? Is it talking to my friend about it? Is it finding a social support group? Is

it having a conversation on Twitter? So figuring out what it is, acknowledging it. So building on what

we've discussed already. I think one thing I would add is that we are often more resilient than we think

we are. And so when we're faced with genetic information that is probabilistic, complex, ambiguous, we

can actually focus on our ability to deal with it based on past experiences that we have. So we might not

have actually received genetic test results before, but we have dealt with uncertainty in other contexts.

So what career to pick, who to spend our lives with. We often times don't think of these moments in our

lives as times of uncertainty, but they are, and they can be stressful at times.

**Marleah Kruzel:**

And so engaging in self-reflection to think about what did we learn in those experiences and then apply

them to our present uncertain situation. And I think that can be really helpful and definitely something

that I have been applying in other uncertain experiences that I've had since learning about BRCA2 and

my family.

**Deepti Babu:**

Right, because as you said, the uncertainty doesn't really stop. So it's a matter of responding to it and

living with it. I like that idea that we have done a lot with uncertainty that we don't even really

necessarily recognize as uncertain at the time. So...

**Marleah Kruzel:**

Exactly. I think we're more experts than we think.

**Deepti Babu:**

Well, that seems like a good place to conclude our conversation because I think you've left us with some

hope and some realization that we probably are managing a lot better than we think. It's just perhaps

these new circumstances are making us question that a little bit. So thank you for sharing your time and

your expertise with us.

**Marleah Kruzel:**

Absolutely. Thank you so much for inviting me here. And I really enjoyed our conversation.

**Deepti Babu:**

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