Transcript Details
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Family Matters: The Link Between Family History & BRCA Mutations

Announcer:
Welcome to the Clinician's Roundtable on ReachMD. On this episode, Dr. Jen Simmons welcomes Dr. Susan Domcheck, who's the Director of the McDonald Women's Cancer Risk Evaluation Center at the Hospital of the University of Pennsylvania. She's also the Executive Director of the Basser Center and a Basser Professor in Oncology. Together, they'll be reviewing the importance of family history in the context of BRCA mutations.

Let's tune into their discussion now.

Dr. Simmons:
So, Dr. Domchek, if you're a primary care provider, are there certain populations that you should be looking at that are at particular risk? Or certain populations who you don't want to miss for genetic testing?

Dr. Domchek:
It's a great question. the family history really matters on both mom's side and dad's side, because 50% of the time, if an individual carries the genetic mutation, they have inherited it from their dad. And oftentimes this sort of gets lost when people are thinking about their family history. Another crucial component is that certain ethnicities, and particularly individuals who are of Ashkenazi Jewish descent, have a much higher chance of having the BRCA1 or 2 mutation than other ethnicities. So to give you specifics, 1 in 40 individuals of Ashkenazi Jewish descent has the BRCA1 or 2 mutation, and that's compared to approximately 1 in 300 in the general population. For that reason, there has been a lot of discussion about whether or not we should just test everyone who is of Ashkenazi descent, but at a minimum, it should be a question that's asked because we have a really low threshold to test people of Ashkenazi descent. And to emphasize the point about men, people often ignore the dad's side – or the family history on the dad's side, so it's really important to get that component. But it's also important for men to get their own genetic testing. They often think, well it's just about ovarian cancer, it's just about breast cancer, it doesn't matter to me. But again, men with specific mutations, and specifically BRCA2, have an increased risk of male breast cancer, an increased risk of lethal prostate cancer, and pancreatic cancer and melanoma. And, in addition, if a man has a BRCA1 or 2 mutation, or other gene mutation, 50% of their kids will have this mutation. So, it's a 50/50 chance for each kid. So, men are not off the hook even though they often want to be.

Dr. Simmons:
Yeah, I think it's a common misconception that the BRCA genes relate to breast cancer and men think that they are exempt. And I'm glad you mentioned that there are other epithelial cancers that men need to worry about. And I just wanted to reiterate, are you in favor of testing everyone of Ashkenazi Jewish descent for the Ashkenazi mutations?

Dr. Domchek:
I think it's a real implementation problem right now, and it's going to give us some insight as to what will happen in the future, so we currently have a study going on right now called the Before Study, where we're offering genetic testing to anyone of Ashkenazi ancestry after doing a digital health platform education, and then having the results disclosed to them, either by their own physician or by medical professionals within the study. So we're trying to do a hybrid model because there are these direct-to-consumer approaches where people have taken out their medical care, their medical providers completely from the equation, and at the end of the day, you're getting genetic testing to help improve your medical care. And, for that reason, I feel pretty strongly that it should be embedded somehow in the medical model, but we've got to figure out how to do it. So I think its' reasonable to consider – it's fine to consider it, but if we don't know how to actually get it done, it's not very helpful. So that's what we're working on right now.

Dr. Simmons:

Or what to do with the information once you've received it.

Dr. Domchek:
Exactly. And if you have a BCRA1 mutation, but you haven't told any of your healthcare providers, they can't help you, right? So we need to figure out a way to do this in an efficient way where healthcare providers are in the loop and yet at the same time not be overly burdensome. One of my primary care doctor colleagues once said that if he did everything that was on the checklist for a primary care doctor to do from, you know, seatbelts to cholesterol, that it would take him 8 hours. And the idea that we're going to add genetic testing to that checklist is hard to get our heads around unless we make it super easy.

Announcer:
That was Dr. Jen Simmons and Dr. Susan Domcheck talking about genetic testing in populations who are at a higher risk of BCRA mutations.

To access other episodes in this series, visit reach-m-d-dot-com-slash-clinicians-roundtable, where you can be part of the knowledge.