Genetic Counseling: Benefits for Cancer Treatment

Claire McDonald MS, MPH, CGC discusses the main types of genetic testing in oncology. She shares somatic/tumor testing for treatment versus germline testing for hereditary cancer risk. She examines the question of whether people should consider direct-to-consumer testing if they are concerned about hereditary cancer risk and how the field of cancer genetics and genetic counseling is evolving.

Featured Speaker:
Claire McDonald, MS, MPH, CGC

Claire graduated from the University of Pittsburgh in 2020 with an MS in genetic counseling and MPH in public health genetics. She has been a genetic counselor at the University of Alabama at Birmingham since August 2020. She currently sees patients in cancer and pediatric neurology clinics.

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Melanie Cole (Host): Welcome to UAB Med Cast. I'm Melanie Cole and today, we're talking about genetic counseling, the benefits for cancer treatment. Joining me is Claire McDonald. She's a Genetic Counselor in Cancer Genetics at UAB Medicine. Claire, it's a pleasure to have you with us and what an interesting topic we're discussing today. And you know, we've heard about BRCA testing and providers are referring their patients for this. But tell us about some of the other types and the main types of genetic testing in oncology. And I'd like you to tell us about somatic tumor testing for treatment versus germline testing for hereditary cancer risk.

Claire McDonald, MS, MPH, CGC (Guest): Right. So, there are really two main types of genetic testing in oncology. The two you just listed. There is somatic or tumor testing, and then there's germline testing. So, somatic testing looks for gene changes in a sample of the tumor itself. And by definition, all cancers have gene changes that are not found in the rest of the body. Those changes have happened by chance as the DNA replicates. And they're

what allow the tumor to actually grow uncontrolled and to be a cancer. So, the somatic testing can have treatment implications because there are plenty of drugs that are known to be effective only if certain gene changes are found within that tumor.

And somatic testing is continuing to become more important over time for cancer treatment, as research about targeted therapy progresses. And then separately there's germline testing, which is done on blood or saliva and can look for inherited gene changes that we'd expect to find in all cells of the body. So, if someone has a gene mutation identified on germline, think that indicates an increased risk of cancer. And each gene has specific cancer types that are more prevalent for people with a mutation in that given gene, whether it's BRCA or a different gene that's been identified. And then there are medical management recommendations such as increased cancer screening or preventative surgeries that can be used for patients who are found to have one of those germline mutations.

Host: So interesting and how somatic tumor is really helping to pave the way for various treatment options. Right? So, tell us a little bit about a genetic counselor's role in oncology and the benefits of seeing a cancer genetic counselor for other providers, Claire. They're telling this to their patients, tell them what you want them to be telling their patients.

Claire: Yeah, absolutely. I think a lot of times patients are told they're being sent for genetic testing and that is some of what we do, but it goes beyond that as well. So, genetic counselors take a detailed, personal and family history in the context of cancer. We focus on cancer and we assess familial cancer risks. And we determine whether genetic testing is appropriate for the patient and which tests should be ordered. But like I said, we also do more than that. One important part of our job is discussing the risks and benefits of testing with the patient and helping them decide whether they want this information, whether it could be beneficial for them because not all patients will want the information that genetic testing can provide.

And we actually have training in counseling in addition to the science of genetics and facilitation of that decision making is definitely a benefit of seeing a genetic counselor. We also assist their healthcare teams in developing medical management plans if a genetic diagnosis is made. So, if someone is found to have an increased risk for cancer, based on a genetic test result, plans can be made for screening that help detect cancer early, when it's easier to treat, or in some cases there are medications or surgeries that can reduce the risk of cancer.

We can also make recommendations or refer to other specialists based on family history, if the patient tests negative or chooses not to have the genetic testing. So, we offer, some insight beyond just what a positive genetic test could show. So, for instance, females with a family history of breast cancer may consider having an evaluation at a high risk breast clinic to see whether they qualify for increased breast screening.

An example could be having MRIs every year, in addition to mammograms, to try to increase detection of breast cancers. And then another example of that is individuals have a first degree relative with colorectal cancer should have colonoscopies every five years starting at age 40, or maybe even younger, depending on the youngest age of diagnosis in their family. And that's different from recommendations for colon cancer screening in the general population. So, since only five to 10% of cancers are hereditary, where we find one of those germline mutations, these recommendations based on family history are important for a lot of our patients.

Host: Well, one of the things I find most interesting when you said that you're also trained in counseling, because I think for providers, that's one of the big questions they get, they meet with you, they get their test results, then what, then they get the inevitable questions. What to do with this information. Speak about some of the practical aspects of the genetic test and how you counsel your patients on whether they go and work in their medical home. Do they work with you? How does that whole referral process work once they get the information and they're trying to figure out what to do next?

Claire: So, a lot of times I'll make direct referrals. So I'll send a message to the Prevention Clinic here at UAB, which is a high risk breast clinic. And they will contact the patient and set up an appointment. And then that patient gets to see one of the nurse practitioners over there and get that personalized evaluation. Or I can refer them to a GI doctor that I know is familiar with family history of colon cancer. So, they can start talking about getting their colonoscopies, things like that.

Host: Now, some people think that it's easier to do these direct to consumer testing, like 23andMe, if they're concerned about hereditary cancer risk and maybe the whole family does it, what do you think of those?

Claire: So, I think it's a complicated topic, but the short answer is that it depends on the company. Some companies like 23andMe test for very limited health information. For instance, you mentioned BRCA, many people have heard of the BRCA or BRCA one and two genes. There are thousands of different mutations or misspellings in those genes that can cause the gene not to work properly, which is what leads to that increased risk of cancer. And 23andMe's tests only actually looks for three of those variants and they're founder mutations in the Ashkenazi Jewish population. So, this means that a negative result for someone who does that test, doesn't actually rule out a mutation in one of those genes, because it's not a fully comprehensive test.

And so it will be more useful for individuals with Ashkenazi Jewish ancestry than those who don't have that ancestry, but any person can still have other mutations in those genes or in the many other genes that are not included on that test. And then in addition, if someone does get a positive result in a BRCA gene on 23andMe, medical grade testing is also recommended to confirm that positive test result. So, they would still need to get another genetic test to make sure that's accurate. Then there are some other companies that offer what I'd refer to as consumer initiated medical grade testing, and those include Color Invitae and JScreen, and they do offer more comprehensive genetic testing.

These companies require approval from an ordering provider, such as the patient's primary care doctor, or they also have third-party doctors, that partner with the company and they usually offer a call with a genetic counselor who's employed by that company to discuss the results as well. So, that's a little more comprehensive than something like 23andMe, and does allow access to a genetic counselor.

But my biggest recommendation would just be for any patient who's concerned about hereditary cancer risk to have a discussion with their primary care doctor or a genetic counselor or other healthcare provider before ordering one of those tests or deciding what might be the best route for them.

Host: One of the things that I think keeps people from wanting to do this and myself included are the insurance implications of hereditary cancer testing. Tell us a little bit about possible insurance discrimination concerns and how you counsel your patients with these concerns, especially if they have a positive reason.

Claire: If someone tests positive for a gene mutation, there's a law called GINA that protects against discrimination from large employers and health insurance. So, this means that for someone who tests positive, their health insurance company can't raise their premiums or deny coverage based on genetic testing results.

But GINA does not apply to other types of insurance, such as life, long-term care or disability insurance. And for those types of insurance, companies can legally ask whether a person has had genetic testing and can raise premiums or deny coverage based on their response. And also other factors such as personal and family history of cancer or other health conditions.

So, even sometimes if a person has a strong family history, they haven't had a genetic test that still might impact

those things. But some patients like to establish these plans before having genetic testing, if that person tests positive, but they already have a plan in place, the company can't change that plan retroactively, and they're not required to call and send and update.

And so when I'm counseling patients, I think this is an especially important consideration for those who've never had cancer where testing positive might really impact the way these insurance companies think about their risk. And also for those who know that there's a gene mutation in their family and therefore they have a higher chance of testing positive than some of those other patients. So, particularly for patients in those situations, I make sure to let them know about GINA, what it applies to, what it doesn't apply to. And then sometimes they'll elect to go, put those plans in place before proceeding with tests.

Host: It's complex. It's an interesting aspect of this amazing field of genetic counseling. As we wrap up Claire, how is the field of cancer genetics, and genetic counseling evolving? And what would you like to see, or where do you see it going in the future? Please offer your best advice, referral information, any other final thoughts that you have.

Claire: So, the field is evolving constantly. One thing that comes right to my mind is that the cost of testing has greatly decreased in the past 10 years. So, some labs will actually offer a $250 self-pay price. So, whether either insurance won't cover the test or there's a high out of pocket cost due to something like a high deductible plan, patients can forgo their insurance and just pay $250 out of pocket for the test. And a test this cheap, really would have been unheard of in the recent past. So, I sometimes see patients who come for updated testing because there are a lot more genes to look at since the last time they had testing. And they have stories about paying thousands of dollars for themselves or other family members to have testing in the past.

So, while $250 is still a lot of money, many patients are reassured to hear that there's financial assistance. And even without financial assistance, there's no chance they'll end up paying thousands of dollars out of pocket, $250 is really the worst case scenario. There are also other new genes being discovered and described.

So, 10 years ago, a patient with early onset breast cancer would have only had BRCA or BRCA one and two testing. Whereas today I offer a 23 gene panel for that person. There's also a new type of testing called polygenic risk score. That's being done mostly on a research basis. We often see what we call familial cancers, which describes families with more cancers than we would expect by chance, but we can't find any gene mutation in that family. These families likely have an increased risk of cancer due to a combination of genetic and environmental factors that are shared within families. But we can't really pinpoint anything specific for them right now, because there's not a change in a single gene that we can identify. A pologenic risk score looks at hundreds of genes where each individual variant may increase or decrease the risk of small amount. And the variants themselves likely don't have a significant impact on cancer risk individually, but they may have an added effect when they're combined or added up together. So, the question is how to interpret these results and translate them into action, such as what would we recommend for increased screening for certain cancers?

We're not quite there yet, but if the interpretation improves in the research setting, I'm hopeful, this type of testing or other updates in technology may shed some light on the apparent increased risk in some families who have negative genetic testing today. And 20 to 30% of cancers are familial as opposed to the five to 10% that are hereditary. So, useful updates in technology has the potential to provide answers for many patients.

Finally, one last thing, I think genetic counselors have really been at the forefront of Telemedicine, particularly in cancer because a physical exam is not usually part of a cancer genetics evaluation. We have more flexibility to be able to talk with patients over the phone or on video chat. And this has been useful, not only during the current pandemic, but also before then, and will continue to be helpful for expanding access to our services for patients who live several hours away from the nearest cancer genetics clinic, because there aren't a lot of cancer genetic

counselors. And so being able to offer that flexibility is one way to expand our services.

Host: What an interesting topic and a fascinating field that you're in Claire. Thank you so much for joining us today and really giving us a nice update on cancer genetics in oncology. And a physician can refer a patient to UAB Medicine by calling the Mist line at 1-800-UAB-MIST. Or you can visit our website at UABmedicine.org/physician. That concludes this episode of UAB Med Cast. I'm Melanie Cole. Thanks so much for listening.