Living Beyond Cancer Podcast Transcript

Understanding Genetics Counseling and Testing

Guest: Dr. Nadia Falah

Lauren Hixenbaugh (00:00):

Welcome to Living Beyond Cancer. I'm Lauren Hixenbaugh, the Coalition Manager for Mountains of Hope. Living Beyond Cancer is a series of podcasts created for cancer patients, survivors, and their caregivers. This series is sponsored by the West Virginia Cancer Coalition, Mountains of Hope, and is produced for the WVUCI's Cancer Prevention and Control. Today we are recording in multiple locations, so please forgive any of our tonal differences.

Lauren Hixenbaugh (00:26):

Today's focus is genetic testing that is available to patients, survivors, and their families. This topic was chosen to help us better understand what is available and how it could be helpful to you and your family. I've invited Dr. Nadia Falah, a Medical Geneticists and Assistant Professor for the WVU Department of Pediatrics in the section of genetics to share her knowledge and expertise with us. Thank you so much for being here today and fitting us into your busy schedule. I'm excited to dive into today's topic and learn a bit more. Let's start off with something pretty basic. What is a medical geneticist?

Dr. Nadia Falah (01:01):

All right. Thank you so much for having me today. A medical geneticist is a doctor who is specialized in diagnosing and treating genetic conditions. Medical geneticists, along with a genetic counselor, counsel individuals, and families at risk for certain genetic disorders or cancers. Cancer survivors face unique challenges that impact not only the survivor but their family members. Cancer genetic counseling can help them and their family understands possible inherited cancer risk. Inherited cancer risk may be passed from a parent to a child.

Lauren Hixenbaugh (01:41):

So if somebody wanted to do a consultation with you, what would a genetic consultation look like for a cancer survivor?

Dr. Nadia Falah (01:48):

A genetic consultation with a cancer survivor is a health service that provides information and support to a survivor who has a possible hereditary cancer disorder. During a consultation, the genetic professional meets with the cancer survivor or their family members to discuss cancer genetic risks, confirm or rule out hereditary cancer.

Lauren Hixenbaugh (02:14):

What happens during that consultation?

Dr. Nadia Falah (02:17):

During that genetic consultation, the genetic professional will review with the cancer survivor, their history, their family history of cancer. We analyze the family tree, also we call it pedigree. We provide a

personalized cancer risk assessment. We discuss whether genetic testing is recommended or not. We also provide recommendations for increased cancer screening and preventions and provide psychological support and follow-up.

Lauren Hixenbaugh (02:49):

I know a lot of people may or may not know their family history. So what do families do if the survivor doesn't know that family history?

Dr. Nadia Falah (03:00):

Having limited information about a cancer diagnosis in the family members, or if you have a small family size, may make it a little difficult to accurately assess the chances of hereditary cancer. However, that shouldn't stop the survivor from seeing a genetic professional.

Lauren Hixenbaugh (03:20):

You mentioned the term earlier about a genetic disease. How do you define that? What does that mean for folks?

Dr. Nadia Falah (03:29):

Yeah, so cancer is a genetic disease, which means that it's caused by certain changes to genes that control the way of our cell functions, especially how our cells grow and divide.

Lauren Hixenbaugh (03:46):

And what kind of this genetic testing can be offered to cancer survivors?

Dr. Nadia Falah (03:53):

There are two types of genetic changes that are related to cancer. The first one is that gene changes that is acquired and is located in the cancer cells or the cancer tissue. The testing, or the genetic testing for that involving the testing the tumor or cancer through a biopsy, that's completely different from what we call the hereditary cancer testing, which is testing the DNA that represents a change or an abnormality that's located in every single cell in our body. That is done by testing the blood, mouth swab, or saliva test. Abnormal genetic finding from the blood or saliva is the one that is increasing the risk for cancer, and that is what is contributor associated with hereditary cancer syndrome.

Lauren Hixenbaugh (04:50):

So this testing of the genetics with the tumor is different? I think I'm getting this right. It's different from testing the genetics of the blood?

Dr. Nadia Falah (04:59):

Yes. Testing the tumor or the cancer tissue are increasingly useful for therapy selection. Many cancer therapies are targeted at the particular gene functions. The oncologist may require information about tumor genetics to use it to target therapies effectively. In these cases, the National Comprehensive Cancer Network, which is the NCCN guideline, as well as the FDA have outlined tumor testing that is recommended for specific cancer and the associated treatment implications. These results could also qualify patients for clinical trials.

Lauren Hixenbaugh (05:44):

Let's talk a little bit about heredity, and what is the percentage of cancer that's actually hereditary?

Dr. Nadia Falah (05:53):

So only five to 10% of all cancer is hereditary, meaning that there was a change in the gene that can be passed from one generation to generation. And that could be contributed to the family history of cancer. Knowing if a family member carries genetic abnormalities allows for better cancer risk prediction and gives an individual an opportunity to look for a cancer earlier or take steps to lower the risk.

Lauren Hixenbaugh (06:29):

Many people are going to be listening to this, and they're going to say... There's this different definition of a survivor for everyone it seems, but they're going to be listening to this and they're going to say, "Well, I already have cancer." Or, "I've already survived cancer, so is hereditary genetic testing, is that still recommended for me?"

Dr. Nadia Falah (06:49):

The cancer survivors with hereditary cancer syndrome have a higher than average chance of developing another different cancer. The US Preventive Services Task Force recommends that breast cancer and/or ovarian cancer survivors should be offered genetic counseling and genetic testing for BRCA1 and BRCA2. The NCCN guideline as well, recommends genetic testing for the cancer survivor who meets clinical criteria for hereditary cancer syndrome. Their guidelines and criteria or updates almost yearly. NCCN guidelines, for example, recommend that an individual with colon cancer or uterine cancer that was diagnosed before the age of 50, even without a family history of cancer should receive genetic testing for possible hereditary causes.

Lauren Hixenbaugh (07:44):

When folks are in this appointment or having these genetic tests done, some possible results for them are positive, negative, or inconclusive. Can you give us an example of how positive genetic testing could be useful for somebody that's a survivor?

Dr. Nadia Falah (08:00):

If you have a positive test indicating possible hereditary breast and ovarian cancer syndrome, you may receive a different approach in future clinical care. For example, you may be recommended to have a double mastectomy compared to a partial mastectomy or removing part of the tumor. You may also be recommended to have preventive surgery and remove the ovaries, giving the higher risk of ovarian cancer associated with hereditary breast and ovarian cancer syndrome. It will definitely change your treatment and make it a more personalized medicine. These social studies have evaluated the survival rate of women who tested positive for BRCA mutation and who treated for stage one or stage two breast cancer. Studies revealed that women who are treated with bilateral mastectomy are less likely to die from breast cancer than women who are treated with unilateral mastectomy.

Dr. Nadia Falah (09:12):

Another example is colon cancer. Individuals with hereditary colon cancer like Lynch syndrome have up to 70% chance in their lifetime to develop secondary primary colon cancer after the first diagnosis of colon cancer. Lynch syndrome also increases a person's risk for endometrial, ovarian, stomach, and

other cancers. Prophylactic hysterectomy, or removing the uterus and removing the ovaries could be considered after childbearing is completed. Because of the increased chance to have more than one type of cancer for those individuals, people with these or other hereditary cancers should be following a specific guideline for early cancer detection, such as breast MRI or yearly colonoscopy, or cancer prevention such as surgery and removing some of the organs that are giving a high risk for cancer.

Lauren Hixenbaugh (10:15):

So how do patients find the guidelines for their results?

Dr. Nadia Falah (10:22):

The medical guideline can be provided by your doctor. Usually, during a genetic consultation, we talk to you based on your genetic finding and which guideline meets the criteria of your genetic results. Every patient is unique and every patient is different. There is no guideline that can be generalized to everybody. It's more specific to what is your genetic abnormality and what we should do about it. We're treating you as of who you are, not as part of the general population or general patient care.

Lauren Hixenbaugh (11:01):

Thank you. That's really helpful. One term that I came across that I don't know much about is chemo prevention, I think I'm pronouncing it correctly. So can you tell me a little bit about that?

Dr. Nadia Falah (11:14):

Yeah. Individuals who do not elect for preventive surgery when it comes to an abnormal genetic finding that suggested a hereditary cancer syndrome or a higher risk for cancer, maybe offer it chemoprevention, which is our, which are medications to prevent cancer from happening. An example for that is Tamoxifen, which had been used for breast cancer prevention, but it has limited information or not have been assessed by randomized trials in a high-risk woman. Interestingly, that aspirin has been shown to decrease the risk for colorectal cancer in individuals with Lynch syndrome.

Lauren Hixenbaugh (11:58):

Wow. That's interesting. You want to tell us a little bit more about aspirin and Lynch syndrome?

Dr. Nadia Falah (12:03):

Yeah. Aspirin therapy has been shown to decrease the risk for colorectal cancer in individuals with Lynch syndrome, based on combined several consensus statement and expert reviews, including the NCCN guideline and the US multi-society task force on colorectal cancer suggests that aspirin can be considered in the management of individuals with Lynch syndrome, of course after taking into account an individual's personal health and co-morbidities. And studies have shown that aspirin decreases the chance of developing colorectal cancer for those people who test positive for Lynch syndrome.

Lauren Hixenbaugh (12:50):

That's great information for our folks, especially with those with Lynch syndrome.

Dr. Nadia Falah (12:54):

Yeah.

Lauren Hixenbaugh (12:57):

We'll move into... In this podcast series, we typically talk about the survivor and their family. So I kind of want to focus in on that family aspect. What does genetic testing, how does that affect the survivor's family?

Dr. Nadia Falah (13:16):

The genetic test result from a person who has had a diagnosis of cancer provides the most accurate risk assessment for other family members. If we find an abnormal change in the gene from the affected person, we take that as a biomarker. We recommend that we test it for all first-degree relatives who are at risk. Individuals who test positive, they will follow the specific guideline for early cancer detection and prevention. And those who test negative will be cleared from having high risk from their family standpoint, and the risk will go back to the general population's risk. Everybody is at risk for cancer, but if they do not have an abnormal gene, then they are not at a higher risk, and they've just followed the general recommendation. If they test positive, we will include them into a higher screening or early detection, or early cancer detection like MRIs, colonoscopies, and may offer them even preventive surgery.

Lauren Hixenbaugh (14:28):

Could you tell us a little bit more about some common family history that people should kind of red flag? What history may suggest that they have that hereditary cancer?

Dr. Nadia Falah (14:45):

Usually or typically consider that the number one risk for cancer is age. And if a diagnosis happens at a young age, which is defined or considered to be below the age of 50, we start thinking about the possible hereditary component. Now, a diagnosis, if you have cancer that was diagnosed before the age of 50, you may qualify for, or you may have a possible hereditary cancer syndrome. If you have a family history of cancer that was diagnosed before the age of 50, then you also could qualify for genetic testing and you should seek a genetic consultation. Now, the number of family members who are affected, that also plays an important role of defining or examining the family history. If you have more than two first-degree relatives with cancer or a family history of both endometrial and colon cancer, male breast cancer is also an alarm sign. Two first-degree relatives with pancreatic cancer increases the risk for pancreatic cancer. Ovarian cancer at any age is also something to consider getting a genetic consultation, cancer diagnosis, and you don't know anything about your family history, or if you have limited information about it, if you are belonging to a particular ancestry or ethnicity like Ashkenazi Jewish, and you have a family history, or you have a history of breast-ovarian cancer. Those are all features that suggests a possible hereditary syndrome, and we recommend that you seek a genetic consultation to examine this further and see if you qualify for genetic testing.

Lauren Hixenbaugh (16:48):

So these folks should definitely get the testing you're saying?

Dr. Nadia Falah (16:52):

Not particularly testing, but they should seek a genetic consultation to address the question and see which syndrome that you may qualify to. We have hereditary breast and ovarian cancer syndrome, we have Lynch syndrome, we have multiple endocrine neoplasia. We will take a detailed family history. We

will look into what criteria that that particular person meets for us to determine what type of genetic testing and how that person would benefit from the genetic testing.

Lauren Hixenbaugh (17:28):

Okay. Well, as we've talked about the benefits, the flip side of that is what are the barriers? In regards to genetic testing, what are the barriers for patients?

Dr. Nadia Falah (17:42):

I think that the number one barrier is the lack of knowledge about genetic testing, the genetic risk assessment, and how the results could impact your health. I always tell my patients that when we see you in the genetic clinic, and if we offer you genetic testing, our goal is to treat you as of who you are. We are tailoring your medical care to you as if we are not applying to every single patient or generalize it to the patient. Some of them, come with a lot of anxiety. They don't know what the visit is for. They do not understand how that testing could help them. So those are the main barriers. I think also lack of time and the availability or access to a genetic service could also be a barrier for having genetic testing done.

Lauren Hixenbaugh (18:46):

When we work in the community, we get lots of folks who mention fear. The fear of the unknown is often something we hear.

Dr. Nadia Falah (18:55):

Right. So again, with the genetic consultation, we do not just evaluate and offer testing, but we also provide them with detailed education support, psychological support. They do not have to do the genetic testing in the first visit. They can decline it and then think about it. We always are there for our patients whenever they decide to do the testing again or to change what they thought about at first. We go through the education step-by-step, and I think that we help them understand how this test will change their clinical care and how it could benefit them in the future.

Lauren Hixenbaugh (19:45):

Okay. In addition to those more common barriers, are there additional emotional barriers that patients face?

Dr. Nadia Falah (19:54):

Yeah. I mean, genetic testing should be proposed along with the proper support and counseling. Decliners may make more informed decision after tailored health education, including adequate risk information. I inform my patients that everybody is at risk for cancer. Receiving positive results means you will want to consider cancer prevention and early detection strategies that are best for you.

Lauren Hixenbaugh (20:26):

Certainly, we want patients to make the best choice for them. You did mention a term that I'm not familiar with, decliners. What does that mean?

Dr. Nadia Falah (20:33):

Decliner is an individual who comes to the genetic clinic and we offer them genetic testing, we give them education and they may not be interested to pursue the genetic testing because of maybe fear, barrier, emotional, fear of anxiety, thinking about insurance coverage. We do have very few number of them after receiving the counseling and the genetic support, we do see a few number of decliners. But usually, decliners, if you do not give proper genetic counseling, you will see a higher percentage of them.

Lauren Hixenbaugh (21:16):

Great. Thank you. So, one of the things that I saw on lots of frequently asked questions and things, as I was researching this topic to talk with you today is folks are really worried about the risk of losing their health insurance coverage. And then the other question that was posed is what does insurance cover? Can you talk a little bit about that?

Dr. Nadia Falah (21:42):

Right. And that is part also of our genetic consultation. We talk about insurance coverage and how genetic testing could affect your life in the future. The Genetic Information Nondiscrimination Act or GINA was passed a little over 10 years ago to address this question specifically. It is a federal law that prevents medical insurers from raising your rates or dropping your coverage based on the genetic test results. It also prevents people from employment discrimination based on genetic testing. The law however does not prevent people from discrimination related to life insurance or long-term care or disability insurance. Different states have other laws related to that, but that is not covered by this specifically.

Lauren Hixenbaugh (22:41):

Okay. And I think, like I said, people are really concerned about that. So that's great for them to understand. As we talk about this, most people know West Virginia is the only state that lies entirely in Appalachia. And so that makes us pretty rural. Let's talk about that from two sides. Where can folks find a genetic counselor, and then specifically in West Virginia, what if the counselor is really far away or not accessible to them?

Dr. Nadia Falah (23:10):

You always have to reach to your doctor for a referral, your primary care doctor, or your oncologist. They can help you find a genetic provider in your area. If you have health insurance, you can also contact your insurance company to find a medical geneticist or a genetic counselor in your area who participate in your plan. And as I stated earlier, tele-genetics is a key to access genetic service to all patients. Before COVID, we offered tele-genetic through an outreach center. After COVID, in addition to our in-person visit and outreach service, we offer virtual video and virtual telephone visits. We offer testing remotely. We order a saliva kit that the patient will receive in their mailbox. They will have to complete all of the instructions and they can send, or they will have a shipping label inside that saliva kit, and they can mail the kit back to the lab. Tele-genetic helps significantly with genetic access to those who are residing in underserved communities, especially West Virginia.

Lauren Hixenbaugh (24:30):

Great. That's great information. Certainly, telehealth has definitely helped us during the pandemic. But now to know there's tele-genetics that's pretty... So another question that had popped up often is, of course, we've all seen the commercials for different types of "genetic testing" such as like 23andMe and

I believe Ancestry is one of them. What are your thoughts on those types of tests and are they beneficial?

Dr. Nadia Falah (25:06):

The 23andMe or even the Ancestry are direct to a consumer test. You don't have to have a genetic order or a genetic consultation to complete those testing. Those testing, or especially specifically the 23andMe doesn't test cancer. It only includes three genetic variance in the BRCA genes. It's a screening test and it only includes three variants or three changes in the gene. There are more than thousands of BRCA variants that are known to increase cancer risk.

Dr. Nadia Falah (25:48):

Now in the genetic testing that we offer in the clinic, it does not only include the BRCA gene, but we have a multi-cancer panel, which includes 84 genes. So the test does not include neither the thousands of the variants that are included in the BRCA gene or the gene of the other, let's say 82 genes that we test commonly for cancer. And based on that, if you have a 23andMe testing that comes back negative, that means nothing. And if you have a 23andMe test that is positive, then you still have to come to the genetic clinic to confirm that positivity. Because if you have a screening test, you always have to do complimentary testing. So either way, you will be coming to the genetic clinic and have the evaluation completed.

Lauren Hixenbaugh (26:50):

Okay. So certainly not comprehensive those tests, it sounds like. So genetic testing is definitely at the cutting edge right now. What do you see the future looking like?

Dr. Nadia Falah (27:05):

There was a great advance in genetic testing in the past five years, and our expectation that would be advanced technology, there will be more labs that offer genetic testing, more providers, and more covered by insurance. The cost of the test is going down. I think when I started genetic in 2014, genetic testing used to be in thousands and not affordable to patients. Now, in addition to coverage by insurance, we can do panels that are including 84 genes. If insurance does not cover, or the patient doesn't have insurance at all, it can be as low as 250 for the full panel. Genetic is not only important to understand the cause of the disease, but also recognizes the manner in which an individual responds to a particular therapy and how to change or how to tailor your management based on your genetic information. Not as based on the general guidelines.

Lauren Hixenbaugh (28:19):

I look forward to seeing how this continues to grow. This has certainly been a great conversation. I would like to add, is there a story, a patient story, or a personal story that really is close to your heart?

Dr. Nadia Falah (28:35):

Yes. I lost my father and my brother because of cancer at the age of 16. So when I was a child, I was observing them fighting for cancer. I think that they fought for a couple of years before their death. Now I have a brother who is in his early 40s who has stage four lung cancer, and he is struggling with the treatment and care. So I mean, cancer is a very devastating disease. If there is anything that we can do to stop it or to learn more about how we can manage it.

Lauren Hixenbaugh (29:28):

Thank you for sharing. If there's one tip from today that you hope listeners would remember, what would it be?

Dr. Nadia Falah (29:36):

Well, I know that I spoke about cancer and genetics, but as a physician, I know that cancer is not just caused by genetics. There are a lot of other multi... I would say it's a multifactorial environment, its lifestyle, some genetic component. And one of how we... Actually, are we seeing doctors at the right time? Are we doing screening? So I think that we may not be able to change our genes, but we can work on a lot of environmental aspects, lifestyle change, healthy diet, exercise, smoking cessation, and take action. The most important is that we have a time where we can say yes, we want to make that change and move forward in making that possible.

Dr. Nadia Falah (30:32):

Now, if you change all of your environmental aspects, the other thing that you can also do is to make sure that you follow up with all of the screening recommendations. You have to know your family history. What is your risk? You need to be on top of your health. Nobody is going to knock the door and say, "Hey, you have to come here." But you have to be on top of your health. You have to be very aware that when it happens, medicine is limited. We can offer treatment management, but there is no magic pill that we give in medicine that will cure you. Everything has some complications, even chemotherapy surgery, and all of those make your life difficult and it's not the same. The quality of life won't be the same. I have patients who come to my clinic and they will be going to other centers trying to find a cure. It's very important for everybody to know that you have to act before it happens. You need to start changing lifestyle. You need to start working on your health, make sure that you do your screening and you do your part. Because again, there is no magic pill that we give that will cure you when the situation will be difficult.

Lauren Hixenbaugh (32:04):

Great, thank you. Where do patients find... We talked a lot about some different resources, but is there anywhere specific that we should lead people that have additional questions?

Dr. Nadia Falah (32:18):

Usually, when we have a patient who have positive results, we tell our patient that, "We're not meeting with you every day. There are some updates that happen or clinical trials that could be available." So we give them the name of the support group and the address, and we recommend or we advise them to sign up for that so they can receive some information, some leaflet, they can be up-to-date about their diagnosis, clinical trials. And one of the associations that we recommended is the National Breast and Ovarian Cancer Center. There is also the National Breast Cancer Coalition, and the Breast Cancer Information Core. Those, we print it out, we give it to our patients. There are also some websites that are associated with those associations, but every patient is having a different association that we recommend. There is for the Lynch syndrome, which I can add...

Dr. Nadia Falah (33:26):

Again, Lynch syndrome is an individual with colorectal cancer, a family history of endometrial cancer, ovarian cancer, and they may get genetic testing that comes back positive in confirming the diagnosis of Lynch syndrome. We usually, if the patient tested positive for it, we give them information about the

supported group and I'm looking at it right now. We have the Lynch Syndrome International, we have the Colorectal Cancer Coalition, the International Society of Gastrointestinal Hereditary Tumors. All the information that we provide, has a website associated with that. Some of them, have the phone number. If they are in the United States, there is an address associated with it. A lot of patients have said good things about those societies. They provide them with updates, opportunities for clinical trials, and they answer questions if they can't reach us to the provider.

Lauren Hixenbaugh (34:37):

Okay. So let me clarify for the patients and make sure I have this correct. So for a patient, they need a referral from their primary care provider to get to you and do a consultation, and then you will provide them with these resources at their consultation.

Dr. Nadia Falah (34:51):

Exactly. We will do the evaluation. We'll see if they will meet the criteria for having a genetic diagnosis, or if they meet the criteria for genetic testing. If so, we'll proceed with testing. Usually, we'll receive the results in about six weeks, and in that time, if they test positive, then we will bring them to the clinic, we discuss with them the results, the implication of the testing, how it is important for the first degree relatives, who should be tested. What is the recommendation for cancer prevention, screening, recommendation? Are there options for preventive surgery? So we actually spend an hour with every patient, just discussing all the details. And we go over the resources that we have and the resource of information that we go through it, at the end of it has some of the associations that we recommend that every patient should sign up for those or subscribe through their website, and make sure that they receive any updates that may not be provided by us.

Lauren Hixenbaugh (36:04):

Okay, great. So Dr. Falah has provided lots of resources. But then to find out more information about living beyond cancer or the Mountains of Hope Cancer Coalition like Dr. Falah mentioned earlier, you can visit moh.wv.gov or wvucancer.org. Also, if you wanted to join our Facebook support group, visit Facebook and search for Living Beyond Cancer. And I would really like to thank Dr. Nadia Falah for joining us today, as well as our listeners. We hope that you will join us in the future.

Dr. Nadia Falah (36:40):

Thank you so much for having me.