Precision Medicine Podcast, Season 6 Episode 67

#2 in the Series "Bringing Precision Medicine to Everyone"

Inherited Cancer Risk: What Every Patient and Clinician Needs to Know with Ellen Matloff

May 21, 2025

Ellen Matloff:

She said to me, "My grandmother didn't make it to my mother's high school graduation and my mother didn't make it to my high school graduation. I have two little girls at home. I'm going to be at their high school graduations. I'm going to watch them graduate from college. I'm going to dance at their weddings. I'm going to know my grandchildren. This is the easiest decision I've ever made in my life."

Karan Cushman:

Welcome to Season 6 of the Precision Medicine Podcast, proudly sponsored by Trapelo. This is the podcast where leading voices in cancer explore how to bring precision medicine to patients everywhere.

Welcome back to the Precision Medicine Podcast. I'm Karan Cushman, your co-founder, producer and host, and we are continuing our series *Bringing Precision Medicine to Every Patient.* We're beginning with understanding inherited risk for cancer. In other words, what we're born with, and how genetic testing can empower patients to make informed decisions about their health and treatment plans.

My two guests today are authorities here yet in very unique ways, but together, we are going to explore how patients and clinicians alike can navigate the complex and growing world of genetic testing and how this knowledge can literally save lives. My first guest is a certified genetic counselor and the CEO of My Gene Counsel, a digital health company that provides scalable genetic counseling solutions for health systems, pharma and precision medicine partners. She founded the Yale Cancer Genetic Counseling Program and served as its director and a faculty member at Yale School of Medicine for 18 years. She was also an individual plaintiff in the 2013 Supreme Court BRCA gene patent case. She is the Forbes.com contributor on genetic counseling and testing and digital health. Ellen Matloff, welcome to the Precision Medicine Podcast.

Ellen Matloff:

Thank you for having me.

Karan Cushman:

Thanks for being with me and Angie. My second guest, and more of a co-host today really, is a physician expert in molecular diagnostics and chief medical officer for Palmetto GBA, where she writes policy that helps move the field of precision medicine forward. She is associate professor of pathology at Loyola University Chicago and board certified in clinical pathology, medical microbiology and molecular genetic pathology. Her passion is education around the appropriate use of genomic testing. Dr. Angela Charnot-Katsikas, welcome.

Angela Charnot-Katsikas:

Hey, thank you.

Karan Cushman:

Before we get started, I'd like to give a shout out to our good friend and collaborator on this series, Dr. Kashyap Patel and his No One Left Alone initiative. Our combined goal for this series, *Bringing Precision Medicine to Everyone,* is to empower patients, caregivers and care teams with the knowledge and tools to bring the best of modern cancer care to every corner of the country. Thank you, Dr. Patel for inspiring this collection of episodes and for your continued leadership and devotion to patients everywhere.

So being the queen of prep that I've been so lovingly labeled, thank you, Dr. Juneja, I'd like to start us off with a little bit of an orientation. We work really hard on the Precision Medicine Podcast to distill down the complexity of precision medicine into everyday terms that all of our listeners, patients and clinicians alike, can really understand and put into action. So just a few stats off the bat. According to the American Cancer Society, 1/3 of people in the U.S. will be diagnosed with cancer in their lifetime. And while only five to 10% of all cancers are considered hereditary, they include some of the most common types, breast, colon, prostate, pancreatic, melanoma and ovarian just to name a few. Inherited mutations increase a person's chance of cancer, of course, and potentially at a younger age.

The problem that we face right now is that we aren't doing a great job of capturing a patient's family history and determining that risk. Even when there's a cancer diagnosis, genetic testing rates are low in some of those key disease areas where we know that hereditary mutations exist. One example report that I found recently, it was published by Stanford University in 2023, studied 1.3 million patients diagnosed with cancer in California and Georgia. Genetic testing was highest in those patients with male breast cancer where 50% of patients were tested, and it was lowest in prostate cancer with only 1.1% being tested in the first two years of a diagnosis. What's worse is study have shown that testing results often come in too late to inform cancer management, but when genetic testing is done at the right time, it can absolutely save lives.

Just a little bit more background, I want to take a second just to circle back to two episodes previously on the Precision Medicine Podcast where this will give us a really good understanding of the landscape of what we mean by a pre-vivor and then also what we're talking about in terms of genetic testing and forming treatments. So, Ali Rogin, she joined us in Episode 51 of the Precision Medicine Podcast where she shared her in-depth story from learning that her father carried a gene mutation for breast cancer, male breast cancer. Ali as some of you may know, is an award-winning journalist at the PBS News Hour and she's now an author, but she found out in her early twenties that she carried the BRCA1 mutation. This genetic mutation gave her a very high risk of developing breast and ovarian cancer. So armed with that knowledge, she made the decision to undergo a preventive double mastectomy.

That proactive choice made before any diagnosis dramatically reduced her cancer risk and potentially saved her life. This is why she's called a pre-vivor. So, this was all because, like I said, her father carried the genetic mutation, and her parents sat her down at the age of 18 to inform and encourage her to get tested. And this is a message she carries with her today. This was almost 20 years ago, so many years before Angelina Jolie had made her same news public. On the other side of this spectrum is my good friend Trevor Maxwell who shared his story in Episode 60. Trevor was diagnosed with stage four young onset colorectal cancer at the age of 41. Statistically, the odds were not in his favor. The five-year survival rate from metastatic colorectal cancer is just 15%. But Trevor, he calls himself biologically lucky.

After diagnosis, genetic testing revealed that he had Lynch syndrome and an MSI high tumor, a combination made him a strong candidate for immunotherapy. Today, more than six years later, Trevor is still here and he's using his story to advocate for better access to testing and personalized treatment. So those are just two stories that highlight both sides of the precision medicine coin, prevention and treatment, and both began with genetic testing. So, Ellen and Dr. Katsikas, you both have really unique personal and professional stories here when it comes to precision medicine and the role of genetic testing that have shaped your personal journey and your career focus. Ellen, I was hoping you might take us back to that case to when you were in your late 20s working as a genetic counselor, what was going on at that time and then after, why has this work become so important to you in your career that it became the impetus for you founding your own company?

Ellen Matloff:

When I started in the field of cancer genetics, it was brand new. We were all really starting at baseline with very little information about how to move forward. And before the genetic tests became available for BRCA1 and 2, we did have several genetic tests for some types of Lynch syndrome, some types of thyroid cancer, but it was really when BRCA1 was discovered and then made public in terms of testing that referrals started pouring in. And at this point, I'd been at Yale for like two months. And I received a referral of a woman who was about 35 years old, very well-educated, very smart, who marched right in, sat down and said to me, "I want to have this test." And at the time, we had all of these professional guidelines and recommendations about all of these steps we needed to go through in terms of educating people because we really wanted to be careful.

So, I said to her, "Let me take your personal and your family history." And when I did, she had an extensive maternal family history of cancer, and she was of Jewish ancestry. And she very politely listened to all of my risks, benefits, limitations of testing, and then said, "Yes, I want to have the test." So, we had her blood drawn and I brought it back with me to and dropped it off at the DNA lab. And about 10 days later, the director of the lab knocked on my office door, which was a converted storage closet where I could sit in my chair and touch all core walls. And he knocked on the door to my little storage space and said, "You know that sample you brought me?" Cause this was one of the first ones, "Your patient came back positive for a BRCA1 mutation." And I was just for some reason blown out of the water because this was one of the first.

Karan Cushman:

Wow.

Ellen Matloff:

And this woman was a provider. She was healthy, she had never had cancer. And so, I was seeing her for a result disclosure at Greenwich Hospital. And the night before I had to go down there and give her this news, I was doing all of these scenarios in my head of, “She’ll come in, she’ll sit down. I’ll say this, she’ll say this. I’ll say this, she’ll say this.” And I was so nervous about it that I could not sleep. So, by the time I reached her the next day you can imagine I looked like I’d been dragged behind a bus from New Haven to Greenwich, and she must have sensed how nervous I was because she, the patient, reached across the table and put her hand on mine and said, “It will be okay. The patient counseling, the genetic counselor.”

Karan Cushman:

Wow, that’s amazing.

Ellen Matloff:

I said to her, “We found that you carry a mutation in the BRCA1 gene.” And she said, “This is what I expected, and I’ve preventatively made an appointment with a plastic surgeon. I’m seeing him in two weeks and I’m having both breasts removed.” And I did one of these, “Oh, hold on. Wait, let’s slow down. Let’s see a psychologist,” which that was best practice then is like-

Karan Cushman:

Sure.

Ellen Matloff:

... “Take three months, see a psychologist. Let’s make sure we know what we’re doing.” And she said to me, “My grandmother didn’t make it to my mother’s high school graduation and my mother didn’t make it to my high school graduation. I have two little girls at home. I’m going to be at their high school graduations. I’m going to watch them graduate from college. I’m going to dance at their weddings. I’m going to know my grandchildren. This is the easiest decision I’ve ever made in my life.”

Karan Cushman:

Wow.

Ellen Matloff:

And it stopped me in my tracks and brought me a kind of clarity about what these patients, many of them have lived through, what their lived story is and the opportunity we had to help them change the story moving forward and how powerful that was.

Karan Cushman:

Powerful, definitely. It seems such a trite word, but that is really amazing. And how many years ago was this now?

Ellen Matloff:

So, this would’ve been in the late ‘90s, and when I was leaving Yale, we had an event that was a screening of a patient movie and invited all of our past patients. And she and her daughter, her daughter, who was like a beautiful woman, walked in the door and she started to tell me her name and I said her name. And she laughed and she was like, “You remember.” And she has never developed cancer. Her sister, who was also positive, had never developed cancer. She’s living a healthy cancer-free life. It’s had trade-offs, believe me. But in terms of what she had experienced with her mom and her grandmother, she’s getting to choose a different path.

Karan Cushman:

Well, I think one of the important points that maybe I missed in my intro a little bit is that when it comes to the BRCA mutations that women who do have that hereditary risk confirmed have as much as a 70% chance of developing breast cancer by the age of 80, I believe, if that statistic still stands. Well, thank you. That’s quite a moving story. So, Dr. Katsikas, let’s fast-forward from Ellen’s patient story, which occurred in the late ‘90s to 20, 25 years ahead. It’s 2018 and you are working as a pathologist, so a bit closer to the field of precision diagnostics, if you will, than the average person. And you begin a very complex journey with genetic testing and screening to discover that like Trevor, who I mentioned, not once, but twice, you discover that you’re lucky to be unlucky. Why did you initially have genetic testing and where did that perhaps fall short in today’s terms, and how were you able to continue to make early, proactive, informed decisions about your health through your journey with cancer?

Angela Charnot-Katsikas:

Sure. So yeah, as you mentioned, Karan, certainly have a couple of lucky to be unlucky situations that have happened throughout my journey. So, I was tested because my maternal grandmother passed away at a fairly young age with ovarian cancer, and that really was my ticket for testing. Meanwhile, I had this awareness that if I were to be positive that my positivity would’ve come from my father’s side, my paternal side. They were the, quote, unquote, “cancer family” that I called them. Although I had limited information about all of the different types of cancers because in a large family, there were various deaths that happened sometimes over the course of many years, many of which occurred when I was a young person myself in my teens and earlier. But I knew enough to know the likelihood was higher on that side. Nevertheless, fast forwarding to a positive test, so I was positive, and the testing itself that was performed was the standard BRCA1, BRCA2 test.

Now for somebody that comes from a family history of a hereditary breast ovarian, and I’m going to add prostate and pancreatic cancer syndrome, the HBAC plus syndrome, the BRCA1, BRCA2 test is not a sufficient test, and we know that today. The guidelines have evolved over the last few years to the point where it is crystal clear now that there are a number of additional genes that should be tested in somebody with that history. In my case, however, I was positive, and so that’s my lucky hit, so to speak. I was also lucky, again, so to speak, having been tested as a result of my maternal grandmother’s history. Had I not had that, I may not have been tested ‘cause again, oftentimes the information elicited from a patient is regarding the maternal history and less often the paternal history. So that information was very helpful because I was then able to have my first risk-reducing surgery.

Karan Cushman:

Sure.

Angela Charnot-Katsikas:

So, I had my prophylactic surgery and prophylactic oophorectomy and hysterectomy, and then intensive screening, which led to when I did end up having a breast cancer diagnosis, it was diagnosed at an incredibly early stage because of that intensive screening. So again, I was lucky in that regard. And so, this really enabled me to really have less treatment than I otherwise could have had, had this been diagnosed at a more advanced stage. So, surgery took care of it, and I didn't require any kind of chemotherapy or anything like that. So that's my journey, and I feel like it's a very empowering message because when you do get the information from a genetic test, there are so many things that one can do to mitigate their risk.

Karan Cushman:

Well, Angie, you really are both a pre-viver and a survivor and really represent why we're doing this episode and this series. You were able to take proactive measures so that you never develop one type of cancer and then remain steadfast in screening for another form of cancer, breast cancer, and found it early. And so, for you, you really represent, you're really a supermodel for precision medicine and personalized medicine. And I know it took incredible courage and determination and so appreciate you sharing your story with our listeners. So, let's take a step back, I think. Ellen, I wanted to just... so we were just talking about testing, and I think there's a step that comes before that, which is, is doing a thorough risk assessment and getting to potentially a risk score.

It's not really happening in practice unless you're lucky enough to be informed like we just talked about in some of those patient scenarios, and with Dr. Katsikas, she obviously has incredible knowledge. But if you don't have that, what does that look like? And I found myself this morning being a mom and realizing, I think I've realized this more recently, that both my kids, male and female have prostate, breast and pancreatic cancer on both sides of their grandparents and they're approaching adulthood. And so, as I think about me being a parent, at what point do I talk to their pediatrician even, right? But it is my responsibility as a parent to go back and I'd have no idea what the hereditary risk is from that. But again, this is where it starts. And so how do you go about doing a risk assessment? What does that look like, and how do you get to a score in clinical practice today?

Ellen Matloff:

Great question, Karan. And by the way, we can circle back and talk about your family history another time. I'm happy to help you with that. But for the average listener, I think that a lot of people, as we were just discussing before, know that family members died, they're not really sure what they died of. So, one thing we can all do is parents and as adult children even is to make sure that you're sitting down with your parents, the family elders, and getting that family history because it's really not that long ago that people didn't even say the word cancer. They felt like it was bad luck, and they didn't say it and it's not captured.

So it's really the responsibility of all of us as parents and as adults to take your family history, and if you're lucky enough to still have your grandparents alive or your parents alive, to really ask them enough information that you can draw a family tree and to keep that pedigree in your safe deposit box or in your wherever you keep your essential documents and to send people an electronic copy of it or else they won't know about their history. And where does that interface with medicine? I'd like to say that a really good history is taken by every pediatrician, every primary care physician and gynecologist that it's taken when you go for a mammogram or colonoscopy. But what I have found as a patient myself as well as a clinician is that that doesn't always happen even with very well-meaning clinicians.

It was only a few years ago that I went to get my own screening mammogram at a major medical center near me. And right on the form it said, "Do you have a mother or a maternal history of..." and they didn't even take the paternal history. And I was horrified. I contacted them and let them know as a genetic counselor, you're doing it wrong, and they fixed it. But even recently, going back for again my annual mammogram, the very well-meaning mammography technician, who by the way, was trying to already do 10 jobs, said to me, "I see that your mother had breast cancer. Has anyone else had breast cancer?" And I said, "No." And she goes, "Okay, good." And that was the whole history, and that is not adequate.

What we need to be doing is getting a standardized family history on both sides of the family that include cancer but other conditions as well, of course. So, we really need to capture the ages of diagnosis, what those diagnoses are, have people in the family had genetic testing? And frankly, there just isn't enough time in most primary care gynecology visits, pediatrician visits for them to do that. And so really looking at tools that can be part the practice so that we don't miss that because a patient is two minutes late to their appointment or because that clinician that day is running a little behind, which, let's face it, that happens often. We don't want to miss this really important moment to ask about family history and to potentially catch things before they happen.

Karan Cushman:

The Precision Medicine Podcast will return right after this. With the explosion of new discoveries in precision medicine, how can clinicians keep pace to know which biomarkers will guide cancer treatment decisions? Trapelo knows. Trapelo is the only decision support platform used by oncologists, labs and payers to resolve the complexities of precision medicine in real time, eliminating treatment gaps for patients. Trapelo knows who to test, when to test, which tests to order, the preferred labs to use, and how to connect biomarker results to the right therapies. It also knows which tests and treatments align with health plan policies, streamlining prior authorization, reducing delays in costs, and unlocking the full promise of high-quality personalized cancer care. Visit trapelohealth.com to learn how you can give cancer patients the most appropriate evidence-based treatment options when time matters most.

Well, and I think what we're talking about here too is something that's often talked about when someone's sick, that you have to be your own advocate and that starts before you're even sick. And so, it really is up to patients to own this pedigree, if you will, own the family history, carry it with you. Patients come in and out of institutions, change health insurance all the time. And so, it's really difficult for clinicians, your primary care doctor, unless you've had them your entire life, it's really hard to put that responsibility into their hands only. So, I think where I wanted to go next is what's interesting is that we're getting to this place of knowledge and consumer ability to act on some of these things. Of course, we are not experts.

That's what genetic counselors are for, helping us get to that score, if you will. And I'll come back to that in a minute. But where I was going with that is we've gotten to this interesting place right now where genetic tests, consumer-facing genetic tests from companies like 23andMe hit the market and reduced the cost, which maybe was one of the barriers to people taking that next step. If you had the fortitude to know, "I need to get that family history, I have the family history, I have a risk, I just want to know more information," and so that's the next practical step is to go do that. "I'm going to do it myself." What is the difference between a test like that from a company like 23andMe and a medical-grade test that consumers really need to understand?

Ellen Matloff:

So 23andMe had a really interesting novel idea, which was to put genetic testing in the hands of the consumer, and they brought a whole new market in and a new way of thinking about genetics. But it really has been genetainment, meaning genetic entertainment rather than a medical assessment and giving you the full range of testing that you should have based on your family history. So, I'll give you an example. I have had so many patients over the years who've said, "I've already had BRCA1 and 2 testing and it was negative." And I say, "Oh, where did you get that testing?" "23andMe." And what they hadn't realized is that within BRCA1 and 2 there are thousands of different mutations that occur in people from every ethnic background from around the globe.

But until very recently, 23andMe was only testing and reporting on the three common Jewish mutations found in people of Jewish ancestry. They weren't reporting the other thousands of mutations, and they weren't allowed to report them by the FDA. So, most of the patients who reported that they were negative to me weren't even of Jewish ancestry. So, they thought they were having genetic testing for BRCA1 and 2, and they were not. And they were also not having testing for the other cancer genes that we talked about that can be part of a comprehensive panel. And so, it can be a starting point, it can be interesting, it can get you interested in genetics, but is it the same as medical grade testing? No.

Karan Cushman:

Got it. Well, that's super helpful. Of course, accessibility is something that's super important. And I think what we were talking about earlier, too, is just the strain on clinicians because that's where we go for information like this. And so, it's really this delicate balance of enabling, empowering that patient to take on some of this, but recognizing that maybe there's not enough genetic counselors to go around and where do I go for information to find one? Where I'm headed next is yes, we get the genetic counselor, right?

Super important, because the most next important thing if you get the testing, if you get the right testing, is in a consumer environment, you're certainly not going to have that professional there for interpretation, and this is another difference in the medical grade of testing and interpretation, is that key component of having a counselor help you interpret those results. Many studies have shown where clinicians themselves, whether it's family practice or in another setting, are not comfortable interpreting genetic testing, even though they may be on the front lines of that. So, what can you tell us a little bit more about the importance of having that genetic counseling professional there as part of just the interpretation process? Why is that so important, Ellen?

Ellen Matloff:

In a perfect world, it would be great if every office, every medical office had access and ready access to a genetic counselor so that that person could take their family history, could order the genetic testing and interpret the results. But what's happened since I started out in the field when there were only a few genes available for testing and the criteria were very, very strict is now we know that every single person diagnosed with ovarian cancer is automatically a candidate for testing. Every person diagnosed with pancreas cancer, automatically a candidate. Probably the majority of people diagnosed with breast cancer, all men diagnosed with breast cancer, many people diagnosed with colon, uterine, prostate cancer are now candidates for genetic testing.

And this will open up so that in a few years, every patient diagnosed with cancer will absolutely be a candidate for testing. And many people with a family history of cancer will be candidates for testing. And so, unless you have a genetic counselor in your pocket at all times, it's pretty hard to make that happen that you've got someone right there and accessible. And so, I think using best-in-class genetic counseling information and knowledge and building it into digital tools that can be used as part of the workflow in these offices so that we're not missing people and that we're not only talking about this to people whose first language is English and who are white and who are wealthy is critically important. And the way that you do that, and we do it with a lot of things these days, is with digital tools.

Karan Cushman:

Great. So, Ellen, as we think about maybe a little bit more on the pre-viver side of the house, what are some of the questions that patients and clinicians alike should be asking in order to determine a patient's risk and whether they should be candidates for genetic testing?

Ellen Matloff:

When you're looking at a cancer history, you want to ask not only about the mom's side of the family, but the dad's side of the family as well, and you want to focus on relatives who are your blood relatives. So, we're not talking about your stepfather or a relative by marriage, we're talking about your blood relatives. We're also not just looking at one type of cancer. So for example, if you have a cousin who's been diagnosed with colon cancer, in the past, sometimes people would focus on who else in the family has colon cancer, instead of asking who else in the family had cancer, because we know that the same genetic mutation that increases the risk of colon cancer, some of those mutations increase the risk of uterine and ovarian cancer and a cancer of, for example, the genitourinary system, things like bladder cancer and cancer of the ureter or urethra. So, asking about all cancers in the family and the ages of diagnosis.

So, we know that people diagnosed at any age can be part of a cancer family history, but particularly people diagnosed at young ages and people diagnosed with more than one primary cancer. So, it's not someone who was diagnosed with breast cancer that spread to the brain. It's someone who was diagnosed with two separate breast cancers or a breast and a pancreas cancer. Those are people diagnosed with more than one cancer. We also want to pay attention to ethnic background because we know that certain genetic mutations are prevalent in different ethnic backgrounds. And so, I would always ask patients, "Are you of Jewish ancestry?" Because they may say, "Oh, my grandfather was Jewish, but I am not religious." It doesn't have anything to do with that. It has to do with the ethnic background and are you at increased risk? Those are some of the questions that clinicians and patients should be focusing on when they're looking at a cancer family history.

Angela Charnot-Katsikas:

And I would just add that it's important to remember that a lot of these same cancers also happen in a non-hereditary way. So, most breast cancer is not hereditary. Most prostate cancer is not hereditary. However, there are these common threads, and it's about putting the pieces of the puzzle together and connecting the dots. And that's where a relevant comprehensive, in so much as it can be, approach is important because again, if you're asking somebody, "Who else on your mom's side had breast cancer?" Well, then you just missed everybody who had prostate cancer, ovarian cancer, anybody on the dad's side who may have had other cancer types that are all related. There are syndromes that are just more commonly expressed as a breast in women or ovarian in women and as a prostate cancer in men, but that doesn't mean they're restricted to those things. So, at a minimum, it's important to ask about all of those things, and it is tough to do in a 15-minute appointment.

Karan Cushman:

For sure. Let's talk about closing some of the gaps here. We've talked about obviously the need for the risk assessment to determine whether or not genetic testing makes sense for the different ways they can go about that, but we don't have the infrastructure in place, for every patient to have this seamless experience. And there's a variety of reasons for that. Even in the largest oncology practices with teams of 30 genetic counselors, it's not enough. And so, what are some ways that we can maybe standardize this process and approach and scale it so that becomes attainable for more patients to get the exact assessment they need and the testing they need, let alone the right interpretation? What do you think, Ellen?

Ellen Matloff:

I think the most rudimentary way is the way that my gynecologist's office operates, which it used to be that I would go in and they would hand me a clipboard with a piece of paper, and I would check off about my cancer history, and they did a pretty good job of covering most things. Now when I go in, they have it on an iPad and they collect that information. That's a heavy lift for the clinician because it means that they're in charge of going through and deciding if that history is relevant enough to take it to the next level. But I would say that's the most rudimentary way if an office, for example, didn't have the resources to use other kinds of tools, but there are also other tools available that can drop into the workflow so that before a patient even comes in, they're filling out this family history on their phone.

Karan Cushman:

Awesome. And so, what about when it comes to the actual testing and then the interpretation, how can we make that a little bit more seamless built into the workflow, if you will? What kind of solutions and tools exist today for that?

Ellen Matloff:

So again, there are some clinicians that are lucky enough to have ready access to a genetic counselor, and they can refer the patient to go and see the genetic counselor. Or in a perfect world, the person can even see a genetic counselor in that office space. That doesn't happen for probably 95%, maybe even 98% of patients in this country. And so, they can either do a telehealth visit with a genetic counselor, or again, there are digital tools that can drop into the workflow so that if a patient qualifies for genetic testing, they can learn the risks, benefits and limitations of that testing and start the genetic testing flow that way.

Angela Charnot-Katsikas:

And it's important, too, to incorporate the correct genetic testing. So, as we mentioned earlier, not all clinicians will be familiar with, "Okay, I think my patient has a heightened risk of a hereditary cancer. Now which test do I choose? Which is the correct test?" I think we've established medical grade is important. There are quality controls. There are regulations that we don't have time to go into right now in the medical-grade testing sphere, but there are so many tests out there, it's really difficult for someone to know which is the correct one to choose. And I will say there are still tests out there that are just for BRCA1 and 2 that are still offered in 2025 to patients with a history of a hereditary breast, ovarian, prostate syndrome. And I've purposely excluded some additional cancers there. But that is happening even in 2025, and I think it's important to keep that in mind. It's the questions that the physicians and patients have to ask, and it's the selection of the appropriate testing that has to be performed, and then, of course, the interpretation of the results, which comes later.

Karan Cushman:

So, Dr. Katsikas, one thing we haven't gotten into yet, but one of the barriers here in all of this, of course, can be cost. And so, from your perspective, I was wondering if you could give listeners just where are we today in looking at genetic testing being reimbursed? And as we talked about on the diagnosis side, we're going to get to a place where every patient that's diagnosed does receive testing. That's a future that will be wonderful, but this all only happens if patients can pay for it. So where are we today in the landscape of health plan policy as far as genetic testing on what gets reimbursed, what doesn't? Can you just give us a view into that and what it might look like in the future?

Angela Charnot-Katsikas:

Sure. So, the landscape has evolved along with the guidelines for the acknowledgement that precision medicine matters and is important to patients and their healthcare journey. So, I think you're going to see across the board more and more uptake of that acknowledgement, which translates then into reimbursement. Having said that, every plan is different, and so I certainly cannot speak to them all. However, that is the trajectory of the science, and typically health plans tend to follow however slow or quickly after that. But certainly, precision medicine is here to stay. It's a standard of care, and an uptake of biomarker testing is becoming more and more significant as the screenings and interventions for patients have increased as well as the treatment availability.

Karan Cushman:

Great. So, Ellen, I'm curious, what are you writing about next in Forbes on this topic?

Ellen Matloff:

Good question. I recently wrote an article about tests that we should be ordering in addition to cancer genetic testing. So if you have a patient who's diagnosed with cancer sitting before you, are there also some pharmacogenomic tests that should be ordered at the same time to see if that's going to be the patient who has excess toxicity to some of the chemotherapies that we use on a regular basis, or who's going to react to other medications that are often used in patients diagnosed with cancer. So, I think that's what we're going to start seeing is that when we have that patient in front of us and we have the opportunity to order genetic testing, does it make sense to take advantage of that and to pair it with other things that are going to be critically important in managing that patient safely and effectively moving forward?

Karan Cushman:

I love it.

Ellen Matloff:

So that's what I wrote about most recently.

Karan Cushman:

So, one more question for you. If folks, listeners want to get in touch with you or follow you, what are the best ways, whether it's LinkedIn or where are you most active on social media? And obviously, folks can find your articles through Forbes, but what are the best ways for folks to follow you and get in touch with you with questions?

Ellen Matloff:

You can come to mygenecounsel.com and follow us there. You can also follow us on LinkedIn, Instagram, Facebook, X and also Blue Sky.

Karan Cushman:

Awesome. Everywhere.

Ellen Matloff:

Yeah, everywhere. You can also write to us at info@mygenecounsel.com if you have questions, although we clearly don't answer personal medical questions. But if you're interested in solutions or you're looking for a referral to genetic counseling, we can certainly refer you to some of the telehealth genetic counseling companies out there.

Karan Cushman:

Awesome. So far, we have talked about patients who are very proactive, eagerly seeking out testing, Ellen, and your first patient that we talked about, and obviously, Angie, very open to getting testing. But I think it's important to recognize those patients who are candidates but may be hesitant to get testing or have family members who are hesitant. Ellen, what do you find are some of those common reasons they may doubt whether to pursue testing, and then how do you help guide or counsel them through those decisions?

Ellen Matloff:

I'm glad you brought this up, Karan, because not everybody wants genetic testing and thinks it's the greatest idea in the world. In fact, we have a lot of patients who hear about genetic testing and feel reluctant or scared. They may be afraid that their insurance company will discriminate against them. They may be afraid of what they learn and what they can do with that information or what they may be forced to do with that information some people feel that will then cause undue stress on them to make decisions that they don't want. And so really, as genetic counselors, our job is to present patients with all of their options and to help them understand the risks, benefits, and limitations of genetic testing so that they can make the most informed decision possible. And some of them get that information and decide not to have testing. It's not for everyone.

Karan Cushman:

Well, thanks, Ellen. That's super helpful. I know I've been living through those decisions in different ways, whether through my mom who has sisters that have both had breast cancer and her navigating her testing on her own because they didn't want to, or as I was saying in the green room, one of my really good friends just recently has been wrestling with this and decided to do it. So, I think it's important to recognize that folks have to come to it in their own terms, whether it's for themselves or for their family members. So, thank you.

Ellen Matloff:

Yeah, of course. And I think it's important that they have the opportunity to really speak to someone about those risks and benefits and limitations of testing, because sometimes they're afraid of things that are not accurate, and sometimes they really just don't want more information. So maybe they're not a good candidate for testing. So, listening to them and giving them the chance to make a decision without judgment is really a critical part of the process.

Karan Cushman:

Absolutely. Absolutely. Back to the interpretation part, so important. Angie, did you want to add anything?

Angela Charnot-Katsikas:

Well, I think simply, just to echo Ellen's sentiment, that everyone has a different idea sometimes about what constitutes genetic testing, what that will mean for them personally, professionally even. Will they struggle to have to have more doctor's appointments? Will have to, will they be able to pay their bills? Will they have to take time off of work? What does all of this mean? And it can be very overwhelming and very scary, but I agree with Ellen 100% in that at a minimum, a conversation with a professional in this space can be so helpful to at least lay out what the reality is of testing and then the person can make a truly informed decision.

Karan Cushman:

Ellen Matloff, we really appreciate you being with us today, genetic testing counselor extraordinaire and expert. Thank you for being with us today on the Precision Medicine Podcast. And Dr. Katsikas, thank you for being my honored co-host today.

Angela Charnot-Katsikas:

Thank you. It was a pleasure.

Ellen Matloff:

Thank you so much, both of you.

Karan Cushman:

Yes, hopefully we have left folks with a little bit more information. And of course, you can reach out to Ellen at My Gene Counsel as we talked about earlier. Thank you both. You've been listening to the Precision Medicine Podcast sponsored by Trapelo. Trapelo is the only decision support platform used by oncologists, labs and payers to simplify precision medicine and eliminate treatment gaps for patients. To learn more, visit gettrapelo.com, we invite you to join us on social media. You can find us on LinkedIn at the Trapelo Company page, on X at PMP by Trapelo, and on YouTube at the Precision Medicine Podcast channel. You can also connect with me your host, Karan Cushman on LinkedIn, X, and Instagram. If you know someone who'd enjoy the Precision Medicine Podcast, please share it. Thank you, and we hope you'll tune in for the next episode.

About Our Guests

Ellen T. Matloff, MS, CGC
CEO, My Gene Counsel

Ellen Matloff is a certified genetic counselor and the CEO of My Gene Counsel, a digital health company that provides scalable, updating genetic counseling solutions for health systems, population and research studies, pharma, and precision medicine partners. Matloff founded the Yale Cancer Genetic Counseling program, served as its director and a faculty member at Yale School of Medicine for 18 years, and was an individual plaintiff in the 2013 SCOTUS BRCA gene patent case. She serves as the Forbes.com contributor on genetic counseling, genetic testing, and digital health.

Angella Charnot-Katsikas, MD
Chief Medical Officer, Palmetto GBA

Dr. Angella Charnot-Katsikas is a physician expert in molecular diagnostics. She is board-certified in Clinical Pathology, Medical Microbiology, and Molecular Genetic Pathology. She served as an Associate Professor of Pathology and director of various clinical laboratories at the University of Chicago.

In 2020, Dr. Charnot-Katsikas joined the MolDx program as a Chief Medical Officer for Palmetto GBA, where she writes policy that helps move the field of precision medicine. She is also a past president of the Academy of Clinical Laboratory Physicians and Scientists. She maintains an academic position as an Associate Professor of Pathology at Loyola University Chicago.
Her passion is education around the appropriate use of genomic testing. And as a breast cancer survivor, her hope is that more patients will benefit from the revolutionary tools that exist in the world of precision medicine today and in the future.