

Yale CANCER
CENTER

answers

WNPR Connecticut Public Radio



Hosts

Edward Chu MD

Chief of Medical Oncology

Francine Foss MD

Professor of Medical Oncology

Understanding Your Genetic Risk for Cancer

**Guest Expert:
Ellen Matloff, MS**

*Director, Yale Cancer Genetic Counseling
Program*

Yale Cancer Center Answers

is a weekly broadcast on

WNPR Connecticut Public Radio

Sunday Evenings at 6:00 PM

Listen live online at

www.wnpr.org

OR

Listen to archived podcasts at

www.yalecancercenter.org

Welcome to Yale Cancer Center Answers with Dr. Ed Chu and Dr. Francine Foss, I am Bruce Barber. Dr. Chu is Deputy Director and Chief of Medical Oncology at Yale Cancer Center and Dr. Foss is a Professor of Medical Oncology and Dermatology specializing in the treatment of lymphomas. If you would like to join the conversation, you can contact the doctors directly. The address is canceranswers@yale.edu and the phone number is 1888-234-4YCC. This evening I will be sitting in for Ed and Francine and I am joined by Dr. Ellen Matloff. Ellen is Director of Cancer Genetic Counseling at Yale Cancer Center.

Barber Ellen, I think a great place to start is how you became interested in genetic counseling.

Matloff I had never even heard of genetic counseling as a career until I was a sophomore in college and taking a genetics course as a prerequisite for a biology premed major. We had a genetic counselor who came and spoke to the class and when she described the combination of medicine and science with counseling, psychology, it seemed like such a great blend to me. Even back then when I graduated from college it really seemed like genetics was the future, and so that's how I started researching it from there.

Barber I had not thought about it in those terms, but you are exactly right, you do have amazing science that is going on right now but you need the ability to interact with human beings, it is not just in the lab.

Matloff I think that's what makes genetic counselors unique to be honest with you, is that we do have to do the counseling psychology piece as well as interpret very complicated laboratory information. Also there is a medicine side of it that comes into play and even most physicians don't have a background in counseling psychology, and so the fact that we have all of these elements really makes it a unique career.

Barber When a layperson, such as me, hears the term genetic counseling, there would maybe be a tendency to go blank, like what is it, and probably a lot of people don't think about it until they are in a position where they have been told they need it. So for someone such as me, what does the field really entail?

Matloff It is funny because when people here genetic counseling, I have heard now all sorts of permeations on what they think it is. One of my favorites is, well I am not old, why would I need geriatric counseling? Actually it is genetic counseling and a lot of people think you are a social worker, you are a psychologist, no, genetic counseling is really a process by which graduate level professionals take an entire family history and we usually take a four generation category and we interpret and do a risk assessment to figure out if the disease in the family appears to be hereditary, or appears to be sporadic. If it is hereditary we are the people to decide which genes are most likely mutated

3:16 into mp3 file <http://www.yalecancercenter.org/podcast/aug2910-cancer-answers-matloff.mp3>

and what testing would be most appropriate, and then we can order the testing, we can interpret the testing, which is very complicated and we can talk about which medical management options would be most appropriate based on a test result and the impact on other family members.

Barber That's interesting when you talk about the fact that you are looking back generations, so again, it is not just science, it is not just reading a chart, and it is not just knowing the latest science, but it obviously also involves some detective work.

Matloff It is a lot of detective work and I would say that one of the greatest changes in genetic counseling since I entered the field is that no one knew what I was talking about, they didn't even know what genetic testing was, and this really was not that long ago that people just gave blank stares and now genetic testing is much better known, which is a good thing, but the hazard is that a lot of these testing companies are marketing either directly to the public or directly to physicians not trained in genetic science saying "Hey, this is a piece of cake, order one for yourself." People don't realize this is not a pregnancy test that you are getting a yes or a no. It is quite complicated.

Barber I would imagine that if you don't know a lot about it you would think, well if this is available and I have the money why not just get tested?

Matloff Sure, and in fact, I don't know if you saw this but last year the New York Times did this article about these cocktail parties in Manhattan that you could go to and the entertainment for the evening was spitting in a vile and having your DNA analyzed so that you could see what track you have and what diseases you are risk for, almost like having a tarot card reader at a party for entertainment, but this is really not a tarot card reading.

Barber No, this is not a Botox party as what it sounds like.

Matloff It is really not.

Barber Who do you usually end up seeing?

Matloff We see a variety of people. Because I work in cancer genetics sometimes we see a person who already has cancer. They have been diagnosed on Monday, today is Wednesday, and they have learned that genetic testing could help guide their management and help them pick which surgery to have and whether or not they should have radiation. So we see those patients that are newly diagnosed. We also see patients who have never had cancer but have a strong family history and are concerned about getting cancer. We also see people that have been treated for cancer, but they are worried about their children, their grandchildren, their siblings and they want to do

6:07 into mp3 file <http://www.valecancercenter.org/podcast/aug2910-cancer-answers-matloff.mp3>

something to either prevent them from getting a second cancer, or to help their family members.

Barber We are speaking with Ellen Matloff Director of Cancer Genetic Counseling at Yale Cancer Center. I am also intrigued, everybody in their job has favorite stories, a great day of work, a great outcome. What springs to mind for you recently that was just like “Wow this is really why I love doing this.”

Matloff My favorite type of day, and it does not happen very often, but my favorite type of days is when we see a family with a known genetic mutation that causes cancer and we test someone and they don't carry the mutation. One story that comes to mind was I had a patient who I had been seeing for years, she had two breast cancers, she tested positive for a mutation and her biggest concern was her daughter who at the time was like 16 and was really too young for testing. So we corresponded over the years and kept in touch and when her daughter was about 22 they came in together and her daughter, comes from a different generation, she has not been through cancer, she is the daughter, she is not the mother, so her level of stress about the whole thing was quite a bit lower than her mom and she had testing and she was a true negative. She didn't carry her mom's mutation and they came in for those results and I gave them to her in person and certainly she was happy and she had a big smile on her face, but when she walked out of the door her mom stayed behind for just a moment and she gave me a hug that could have broken my back and just sobbed for like three seconds; her daughter never saw it. I could feel in that hug the relief that her daughter would not have to go through what she had been going through now for about 15 years, it was just amazing. I felt oh if only this could happen every day.

Barber The other side of that, and that's a wonderful story, but obviously part of your job is delivering difficult news and working with people you oftentimes going through one of the most difficult periods in their life.

Matloff Yes.

Barber How do you do that?

Matloff It is challenging not only for the patient but for the care providers, because so many of our patients are diagnosed with cancers at young ages or they are diagnosed with many cancers. I saw a 76-year-old gentleman recently on his seventh primary cancer. This poor guy has been through hell and although the work is challenging, I think to be able to deliver the information in a sensitive, warm, caring, manner and to give them accurate information that could really help them prevent developing another cancer, that's how we get through it. Also, as a team, and this is an unusual model, we have a counseling meeting once a week where we vent all this stuff and we even have

9:21 into mp3 file <http://www.yalecancercenter.org/podcast/aug2910-cancer-answers-matloff.mp3>

a psychologist comes once a month to help us, her name is Rebecca Behrends. She is here in New Haven and she comes to help us talk on all this because otherwise you burn out.

Barber This got to be so stressful and so difficult because I would imagine you get so involved with your patients and then you have to deliver that tough news.

Matloff People may be surprised to hear this, but I can't tell you how many times I have stared at the fax machine as a result is coming though and you can see the patient's name come though first and you are standing there like God please let this be whatever and I am sure people would be surprised to hear that, but we really care about these people and we are just as hopeful that we will get good news.

Barber If someone is referred to you to get cancer genetic testing, what should they expect?

Matloff They should expect to come in and have us ask them all about their personal history and their family history and the more information they bring us the better our risk assessment and we have patients all the time who will say things like, I do not know I am not close with my family, I don't have a lot of information, but in this day of the Internet, and Facebook, and Goggle, you will be amazed how many relatives you can track down if you really try. We have had patients contact family members they have not spoken to in 30 years and the family member says, you know what, since we last spoke I had breast cancer, I had genetic testing, and I carry this mutation, so really even half an hour of detailed detective work, as you mentioned, can really give us so much more information.

Barber Are people getting better about maintaining their health history, or is that still an area where you would love to see some improvement?

Matloff There is a lot of room for improvement there. Ironically, people that are into genealogies do this really as a pastime and there are genealogy groups and genealogy web sites, but when you get the genealogy it will have peoples full names, their dates of birth, if they got married, who they got married too, and date of death, but it would not often say what they died from, or did they have cancer, how old were they at diagnosis, and really by just adding a little bit more information it could go from an interesting hobby into an essential piece of family history information that you could pass down to generations and it could save the lives of your future family members.

Barber How much of cancer is hereditary?

Matloff We believe that it is the minority, that about 10% of cancer is passed down in families, but I will

12:18 into mp3 file <http://www.valecancercenter.org/podcast/aug2910-cancer-answers-matloff.mp3>

tell you that as genetic research unfolds on a daily basis we are learning that more and more things are hereditary, for example, everyone knows that if you use tobacco you increase your risk for these 10 different cancers. However, if you carry a genetic susceptibility to tobacco, you may be the one who develops an early onset head and neck cancer or an early onset lung cancer, so even the things that are clearly environmental may also have a thread of hereditary in them.

Barber What do you think about the future of genetic counseling? I have read about how now they have started to, and correct me if I am wrong, identify some of these differences that will put you at a higher risk. What role do you think more genetic counseling will have going forward?

Matloff I am not sure because we are never going to say to someone, you do not have the mutation for tobacco, so please smoke a pack of Marlboros a day. We are never going to say that. We are never going to say, you don't seem to be at high risk for melanoma, so please jump in a tanning bed. Because even baseline these things do harm to everyone and I don't know if it may influence treatment at some point because that's really where the field of cancer is going. Instead of giving every single patient with colon cancer the exact same treatment, we are doing genetic testing on these patient's either on their tumor or even on their blood and saying based on your genetic profile we can tell that you will respond to this therapy most effectively, personalized medicine, that's where it's headed.

Barber When we come back let's talk a little more about the science and what is going on. We are speaking with Ellen Matloff Director of Cancer Genetic Counseling at Yale Cancer Center.

*Medical
Minute*

There are over 11 million cancer survivors in the US and the numbers keep growing. Completing treatment for cancer is a very exciting milestone, but cancer and its treatment can be a life changing experience. Following treatment to return to normal activities and relationships may be difficult and cancer survivors may face other long-term side effects of cancer including heart problems, osteoporosis, fertility issues, and an increased risk of second cancers. Resources for cancer survivors are available at federally designated comprehensive cancer centers such as one at Yale Cancer Center to keep cancer survivors well and focused on healthy living. This has been medical minute brought to you as a Public Service by Yale Cancer Center. More information is available at yalecancercenter.org. You are listening to the WNPR health forum on the Connecticut Public Broadcasting Network. I am Bruce Barber, and my guest is Ellen Matloff, Director of Cancer Genetic Counseling at Yale Cancer Center.

Barber We promised we would get into the science a little bit but I want to just back up briefly because

15:25 into mp3 file <http://www.yalecancercenter.org/podcast/aug2910-cancer-answers-matloff.mp3>

we touched on this before, those direct-to-consumer kits that you are very concerned with, as someone who is very well respected in this field.

Matloff Now, consumers can either go online and order their own genetic testing, and there were even some kits that were available in Walgreens, which have since been pulled at least for the time being, in which people could just with a simple saliva sample send their DNA for testing and the results would come back and you are left to interpret that yourself. When the field of cancer genetic counseling really launched in the late 1990s, our greatest concern was insurance discrimination, that my goodness, if you find out you carry a mutation you are never going to get a job, your health insurance will drop you, bad things will happen, and there are laws in place, both locally and nationally that protect people and we have 1400 families, we have only had two people who have ever reported any type of genetic discrimination and even those claims are still washing out. The greatest risk of genetic testing we have learned over the years has been the risk that your genetic test will be misinterpreted, and we are seeing this again and again that well-meaning providers who are trying to do a service for their patient's and have been told by these companies that you can order this yourself, they are ordering it, but sometimes ordering the wrong test or ordering the right test but misinterpreting the results. We have had women both locally and nationally who had prophylactic mastectomies only to learn later that they did not carry a mutation, women who have had their ovaries removed preventively and later learned that their information was misinterpreted, and this has become such a hot-button issue that some collaborators of mine across the country have written a paper collecting these cases and we are going to be publishing those data in August in the journal called Connecticut Medicine, so I guess what I would say to people, I understand that it is convenient to get on the internet and order you own genetic testing and it sounds like fun, but if you care enough to order it at all and care enough to have it interpreted correctly, go to a genetic counselor who is certified.

Barber What I find, as the spouse of a physician, and having watched her go through medical school and now practice medicine, so much of it is an art as it is science.

Matloff It is, and keep in mind also that these medical schools, and I teach here at Yale which is a great medical school one of the best in the country, but you would not believe how much stuff is packed into these kids schedules, and so genetics is only a tiny part of what they learn and you cannot even expect the best minds in the country to be able to pick up an entire field after a few hours of training in a specific field, that is why we have specialties.

Barber: And it is so new, I am mean when you say just in what, the late 1990s that the field emerged, so this is brand new stuff.

18:48 into mp3 file <http://www.yalecancercenter.org/podcast/aug2910-cancer-answers-matloff.mp3>

- Matloff It is brand new, in fact, one of my first responsibilities when I came to Yale was to write a book chapter on cancer genetic counseling, one did not exist, and I remember piecing together stuff because it just did not exist, we were making it up as we went along, and as these genes were discovered and as we began doing testing.
- Barber It does seem like now, getting into the science piece of this, I do not know what impact this had, but since mapping the human genome it seems as if, to this lay person, the amount of science that is emerging is just amazing.
- Matloff It is breathtaking. It is all I can do to keep up on the literature in cancer genetics, now that's cancer genetics, that is not prenatal genetics, cardiac genetics, the genetics of neurology. There is no way that if you came to me for genetic counseling, quite frankly, that I could counsel you on anything but cancer genetics, and this is my job.
- Barber So one of the things in your job description has to be balancing your clinical obligations with whatever publishing you are doing and you mentioned the chapter and staying up on the literature.
- Matloff It's crazy. I will say there are some good things about that. It keeps you fresh, you are always learning, you are never bored, but when faced with things like, I just had a baby; can I take 5 years off and return to the field? It is a little daunting because in five years, the field will be completely different.
- Barber What are the things that the science is shouting at us right now?
- Matloff The big news in science since the human genome was sequenced is we knew about all of these genes, but the testing was so expensive that no one was going to get tested for all of the genes in your body. Since that period of time, the technology has gotten so much better and so much cheaper in such a short period of time that, believe it or not, to sequence all of your genes, right now it is estimated to cost in some labs between five and six thousand dollars. That's especially impressive when you consider that right now when I order just BRCA1 and BRCA2 testing, the breast ovarian cancer genes, just those two genes cost four thousand dollars for all the testing. So, it gives you a sense of Wow, two genes for four thousand, I might as well have the whole genome for five thousand, that is amazing. The dilemma is that instead of getting a one-page report people are going to get a report that looks like a small phonebook and interpreting those data is going to be incredibly challenging, even for those of us who know what we are talking about.
- Barber If you had unlimited resources, unlimited financial resources, and you could just go out and do any kind of research or any clinical work, anything you wanted to do, what would that be?

21:55 into mp3 file <http://www.valecancercenter.org/podcast/aug2910-cancer-answers-matloff.mp3>

- Matloff Well first of all, I think I would train more people. We have a dedicated summer fellowship here for 3 months each year and we do a national search and picked the cream of the crop to come study here for 3 months. I would like to do that and actually recruit more students each year to be fellows to really learn about the field of cancer genetics, but I would also like to target certain at risk audiences. For example, we know that there are three common Jewish mutations that really increase the risk of breast and ovarian cancer. One in 43 American Jews carry one of those mutations and the majority of those men and women do not know they carry them. I would love to hire more genetic counselors and to target synagogues, Hadassah groups, because there are a lot of Jewish organizations that are very well known and very well run. I would love to go there and offer them as a group educational sessions and even genetic counseling and testing so that these men and women would not die of breast, ovarian, or pancreatic cancers for no reason.
- Barber Now let us talk about when you get into that thing where you know you have got a family history, you have come to genetic counseling, what does that all mean going forward? You have got some tough decisions, what is your process for figuring that out?
- Matloff Each family is so unique. You would be amazed at the different scenarios. Some people say, well I am not talking to this sister and we have not seen this relative in 20 years and I do not want to bother this cousin because he is going through a divorce, there are so many interesting family dynamics going on, but what we basically do is we take a look at the family history, we determine the risk, we determine if testing is appropriate, whom in the family would be best to test first in terms of accuracy, and then we take it from there.
- Barber Spend a little time talking about what your option areas with respect to treatment these days with some of these scientific advances that have taken place.
- Matloff I mentioned a few moments ago that personalized medicine is the wave of the future. This is where it is going, and we now have some treatments, including PARP inhibitors, that look promising for cancers and people who carry a BRCA1 or BRCA2 mutation, these are treatments that are specially geared toward those types of cancers and we certainly have a long way to go in clinical trials to prove this, but it is our first promising BRCA related break through.
- Barber This has been in the news a little bit, the decision to not allow patents on genes, explain that to me.
- Matloff Let me give a little disclaimer here, I am a plaintiff in this lawsuit.
- Barber No kidding.

25:05 into mp3 file <http://www.yalecancercenter.org/podcast/aug2910-cancer-answers-matloff.mp3>

Matloff Yes, I am. Although it is important for me to mention that I represent myself in the lawsuit, and I am not a representative of Yale University, Yale Cancer Center, or Yale School of Medicine. I represent myself and in this lawsuit the American Civil Liberties Union, ACLU brought together a few clinicians like myself who are outraged about the patents and what has happened in the wake of the patents, but also some patient's and patient organizations and some professional organizations, and sued this company Myriad Genetics in Salt Lake City, Utah over patents. We thought these patents should have never been issued and much to our surprise, someone who reviewed the case, a judge who reviewed the case, decided that these patents were unconstitutional. It has been, as we expected, appealed and it will go to the next level. A lot of people are saying that this will go to the Supreme Court so we will have to wait and see what happens.

Barber I would imagine that with science advancing so quickly you get into a lot of these questions of ethics.

Matloff I think so, and one of my biggest beefs with the patent is that the genes could have never been found without hundreds and hundreds of families donating their DNA and their family pedigree's to these scientists saying please help us, and I do not think that they thought a big company was going to come along and stick their flag with a patent, into their genes and say we now own these, and anyone who wants to do this testing has got to pay us thousands and thousands of dollars more than the test cost, to get this test.

Barber As this moves forward, and it is obvious to me at least that these things are being figured out as we go, what are some of the exciting things for you that are on the horizon that you can see, not way out there, but that you can really see?

Matloff One of the big ones has to do with the kind of targeted therapies we have mentioned. Right now, if I learn that someone carries the mutation, let's say in one of our colon cancer genes that we test for, Lynch syndrome, one of my recommendations to women who are age 35 or older is this mutation, in addition to causing colon cancer, causes ovarian and uterine cancer, so you need to have a total hysterectomy. Have you ever tried to say that to a 35-year-old woman? That is a tough conversation to have, and one of the things I am excited about is that I think within the next 5 to 10 years I may have something better to offer them.

Barber That is terrific, and as we as close out, what would you say to someone who has not reached the point where they may have been told they need cancer genetic counseling, or they have not even had a diagnosis, but may be aware of it in their family? What are the types of things you can do to prepare yourself in case you have that conversation at some point with a physician?

Matloff I learned a lot from a patient I saw recently. She is 38 years old and she had known for a long time

28:42 into mp3 file <http://www.yalecancercenter.org/podcast/aug2910-cancer-answers-matloff.mp3>

that there was breast cancer on her dad's side of the family, her grandmother and aunt had it. And she is busy, she is married, she has got little kids, and a job. It was scary for her. She wanted to ignore it and she did ignore it until she was diagnosed with an advanced breast cancer, and that is where she is now, going through a bilateral mastectomy and chemotherapy. She is sick. She has had to talk to her children about this. There is a possibility that she could die and the thing that she said to me was, "I was knew this was looming in the background and I chose to keep my head stuck in the sand, and if only I could back." So for the person listening who is thinking this is scary, I do not want to open Pandora's Box, it is not as scary as getting a diagnosis of cancer, prevention is the way to go.

Barber Ellen Matloff is Director of Cancer Genetic Counseling at Yale Cancer Center.

If you have questions or would like to share your comments, visit yalecancercenter.org where you can also subscribe to our podcast and find written transcripts of past programs. I am Bruce Barber and you are listening to the WNPR Health Forum on the Connecticut Public Broadcasting Network.