Cancer Genetic Counseling

Guest Expert:
Ellen Matloff, MS
Director of the Yale Cancer Center Cancer Genetic Counseling Program

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I am Bruce Barber and this is Yale Cancer Center Answers with Drs. Ed Chu and Ken Miller. Dr. Chu is Deputy Director and Chief of Medical Oncology at Yale Cancer Center and an internationally known expert on colorectal cancer. Dr. Miller is a Medical Oncologist and the Director of the Connecticut Challenge Survivorship Clinic. Ken specializes in pain and palliative care. If you have a question that you would like to submit about cancer, please e-mail us at canceranswers@yale.edu or you can call 1-888-234-4YCC. If you are interested in listening to past editions of Yale Cancer Center Answers, each segment is posted on the Yale Cancer Center website at yalecancercenter.org. This evening, Ken Miller talks to Ellen Matloff about genetic testing and counseling.

Miller Ellen let's begin this evening by having you explain to the audience what genetic counseling is.

Matloff Genetic counseling is really a communication process where we see families who are at increased risk for hereditary cancer. We take their entire family history, draw out a pedigree and determine the chance that the genetic mutation may run through the family and cause them to have a higher rate of cancers. Then we can offer them genetic testing if appropriate and use the results to manage their medical care.

Miller Unfortunately, the majority of families have had a relative with cancer; someone with breast cancer or colon cancer. Of that large group of people, what number has a genetic cause?

Matloff That is a great question. Most of us unfortunately have, or have had, a family member with cancer, but it is the minority of people, only 10%, that have a hereditary cancer. So the majority, 90%, are not caused by an inherited mutation.

Miller So in general, if someone has a relative with cancer, odds are it is not something that runs in their family.

Matloff That does not usually run in the family, but it is really important to know the risk factors that would indicate that the cancer may run in the family.

Miller So there are a group of people walking around who do have a family genetic risk for developing cancer. Do you have any sense about what percentage of that group knows about it?

Matloff It is a tough question. I would say probably less than half of people who actually carry a mutation know that they carry the mutation. However, compared to 10 years ago, we have made great strides in genetic testing and counseling. Keep in mind that we did not even have this kind of testing 15 years ago.

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Miller: Along those lines, genetic counseling is a field less than 15 years old, how has it changed?

Matloff: The field of genetic counseling really is about 35 years old, but when genetic counseling began it was mainly for pregnant women and had to do with amniocentesis and CVS. That is still a huge part of genetic counseling, but the field of cancer genetic counseling, my specialty, really took off about 15 years ago.

Miller: What technology in terms of testing coincided with that?

Matloff: Probably the greatest landmark has been the discovery of the breast and ovarian cancer genes called BRCA1 and BRCA2; that happened in the mid to late ‘90s.

Miller: What is BRACA1 and 2?

Matloff: BRACA is the name of the test developed by the company. It is like the difference between tissue and Kleenex. One is the brand name and one is the actual name. It is BRCA1 and 2.

Miller: That is the best explanation and illustration I have heard. This made it a very public thing with breast cancer being so common. Let us talk about some other types of cancer such as colon cancer. What kind of testing has come up in that field?

Matloff: We actually have very good testing for colon cancer. The two main colon cancer syndromes are Lynch syndrome, or hereditary nonpolyposis colorectal cancer which is its long name. Basically what it means is that in those families we tend to see an abundance of colon cancer before the age of 50, and we also see an excess of uterine and ovarian cancers in those families as well as some rare findings called sebaceous adenomas or carcinomas. That is the first colon cancer syndrome. The second colon cancer syndrome we have known about for a long time and it has to do with hundreds or thousands of polyps in the colon, often times occurring in children and certainly occurring in most people by the age of 40.

Miller: This is an important topic. Let us talk more about colon cancer. Can you give me an example of the family history that you have seen with Lynch syndrome?

Matloff: I saw such a family recently. The young woman in the family developed a colon cancer at age 37, and when we looked at her family history her mom had had both colon and uterine cancer before the age of 50. Her aunt had survived ovarian cancer and she had an uncle who had survived colon cancer. I recently sat down 

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with the entire family and took their extended history. It turns out that the
grandfathers’ relatives all had early-onset colon cancer and there was a very clear
pattern in the family. It is not always quite that clear and many families do not
know that much about their family history.

Miller: That is very true. For our audience listening, if someone has Lynch syndrome,
what is something that would make them say, "Perhaps I should be screened."

Matloff: Anyone who had colon cancer before the age of 50, or has several colon cancers
on the same side of the family within the same bloodline should be screened. In
addition to colon cancer, if there is a uterine or ovarian cancer in the family plus a
sebaceous adenoma or carcinoma, that would add to the risk. We also do testing
on the actual colon tumor. In that testing called MSI, or microsatellite instability,
along with another test called IHC, or immunohistochemistry, we can get a good
sense about whether or not that particular tumor is part of a Lynch syndrome
family. That would lead to genetic counseling and testing.

Miller: If there is suspicion that a family may have Lynch syndrome, you are saying that
a tumor itself that was removed can still be tested?

Matloff: That is right.

Miller: And it is tested in the pathology department?

Matloff: A lot of people do not realize that when a tumor is removed, the pathology
department keeps part of that tumor in paraffin embedded blocks for at least seven
years and often times longer. We can go back and test that colon tumor a year, or
five years later. As long as we can get our hands on it, we can do that testing.

Miller: Let me ask you a general question. If you have tumors that are being saved, why
wouldn't everybody who has had colon cancer be tested?

Matloff: You bring up a great point and I have tried to convince the pathology departments
of many hospitals that we should at least be screening using MSI and IHC on
every single colon tumor. Some hospitals do it and others aren't really there yet.

Miller: That may be a tip off for a family that even though they haven't been identified,
they may be at risk.

Matloff: Absolutely.

Miller: We said a couple of words about patients with polyposis. Can you give us an
example of what this might be like?

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Keep in mind that many people have polyps removed that turn out to be hyperplastic polyps that are not thought to lead to colon cancer. Then there are adenomatous polyps which we think may be precancerous. I have learned a lot about polyps in the last decade. There are all sorts of polyps. There are polyps called juvenile polyps, which you would think are polyps seen in young people, but what it means is that it is a specific type of polyp seen in a syndrome called juvenile polyposis. There are also polyps called hematomatous polyps, which are seen in other syndromes. Not all polyps are created equally.

Well said.

When we see a polyp in a very young person, someone before the age of 40, we make sure to get the pathology on that polyp and see if it requires further genetic counseling. In a polyposis syndrome I am talking about people who have dozens, hundreds, or thousands of polyps.

This is very different than the usual situation with maybe one or maybe none.

We had an email question from Suzanne on a different topic; a different type of cancer. She says, "I am 35 years old and both my father and my sister have had melanoma. Am I at risk also?"

That is a great question. With melanoma there are several things that can put people at increased risk. One of them would be shared physical characteristics. For example, people with light skin, light color hair and light color eyes are definitely at increased risks for all skin cancers. With melanoma, there are a couple of genes that are known to cause hereditary melanoma. We don't do a lot of testing for this syndrome but the risk factors would be; people diagnosed with melanoma at young ages; under the age of 50, people who are diagnosed with multiple melanomas; several primary melanomas in different parts of their body, and people with a family history of melanoma. The surprising risk factor is people who have a family history of pancreatic cancer.

How are those related?

The same genetic mutation that puts people at risk for melanoma can also put them at risk for pancreatic cancer. Now this does not mean that everyone with melanoma is at risk for pancreatic cancer. It is a small subset of melanoma patients that carry this mutation who are at increased risk for pancreatic cancer. I would also like to mention that I have been shocked because a lot of people know that skin cancer, in general, can be caused by sun exposure, but believe it or not, they do not know that tanning booths can cause skin cancer as well. I have

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noticed, and I am really horrified by this, that a lot of health clubs and gyms have
tanning booths. If this is a health club promoting good health, why have a tanning
booth? If you are going to have a tanning booth, you might as well hand-out a
pack of Camels. It is just ridiculous. We should not have a tanning booth at any
place that promotes good health.

Miller  Thank you and I hope everybody heard that and listens to it. One of the issues
that I've heard come up in regards to genetic counseling is that people do not want
to be tested and don't want to know. What is your experience with that? Is that
what people are saying?

Matloff  That it is a very common first approach, because people do not want to open
Pandora’s Box. Why would they want to know? The answer is, you want to
know so that you have the knowledge to use to change your risks. If we were able
to say to you, "I have looked into my crystal ball and you are going to be hit by a
car today somewhere on College street," you might do your best to avoid College
street today. We do have things that we can do to reduce cancer risk for people
we know they are at great risk.

Miller  We would like to remind you to email your questions to canceranswers@yale.edu.
We are going to take a short break for a medical minute. Please stay tuned to learn
more information about the cancer genetic counseling with Ellen Matloff from the
Yale Cancer Center.

This year over 170,000 Americans will be diagnosed with lung cancer. More
than 85% of lung cancer diagnoses are related to smoking and quitting even after
decades of use can significantly reduce your risk of developing lung cancer. Each
day patients with lung cancer are surviving thanks to increased access to
advanced therapies and specialized care. New treatment options and surgical
techniques are giving lung cancer survivors more help than they ever had before.
Clinical trials are currently under way at federally designated comprehensive
cancer centers like the one at Yale, to test innovative new treatments for lung
cancer and patients enrolled in these trials are given access to medicines, not yet
approved by the Food and Drug Administration.

This has been a medical minute. More information is available at
yalecancercenter.org.

Miller  We were talking a couple of minutes ago about the process of genetic counseling;
the reasons why patients may want to go or may not want to go. What actually
happens when you see a new patient for an appointment?

Matloff  When the person comes in we sit down and we take an extended family history,
which we call a pedigree. Together we basically draw a picture of four

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generations of their family. People listening may be thinking that they do not know all the information on their relatives, and that is very common, but you will be surprised at how much you actually do know once we start asking the questions. We can figure it out using very basic information, and based on that pedigree we are able to determine the risk that there may be a genetic mutation in the family. We then talk to the patient about all of their options. One option is genetic testing, another option may be increased surveillance or some lifestyle changes. We do not just draw their blood at the first visit, which I think is important for everyone to know. If they do have genetic testing, we navigate the insurance authorization process for the patient to make sure that their insurance company will cover these charges.

Miller Do you offer genetic testing without counseling?

Matloff We do not. We get a lot of phone calls from people who say they do not want the genetic counseling, just the blood test. One thing I would like to make very clear is that people think we only have genetic tests for a few genes. They think that if there is breast cancer in their family, clearly they need that BRCA1 and 2 testing. What they are responding to is propaganda from a major company that sells that test and gets a commission on every test that is sold. In fact, I saw a family and on their little referral slip it said test for BRCA1 and 2. When I went to get the women out of the waiting room she was with a man whom I noticed had these skin-colored, wart-like lesions all over his mouth. I thought to myself, how I am going to tell this patient that her husband has Cowden disease? In fact, it was her brother. It turned out that in their family there is a genetic mutation that causes not only breast cancer, but also thyroid cancer and the skin lesions I saw. If I had ordered BRCA1 and 2 testing in that family, we would have never found the mutation.

Miller When I used to live in Maryland I sent one of my patients to a genetic counselor regarding his history of colon cancer. What they found was that it was not familial, but that he had a neuromuscular disorder that ran in the family. I never noticed these changes in the man’s facial features etc., but the genetic counselor really tuned into it. I give people in your field a lot of credit. What is the training background for genetic counselors?

Matloff I would say that most genetic counselors have a bachelor's degree, and also premed in biology, chemistry and also psychology, perhaps more psychology than the average clinician, and then their graduate degree, which is a master's degree in genetic counseling.

Miller When a patient comes to you, is it usually by referral or can people seek counseling themselves?

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Matloff I would say that most people come by referral, but we also have plenty of self-referrals.

Miller Let’s talk about the psychology part. Where does the psychology come in? What are some of the issues that you deal with?

Matloff There are a lot of issues. These are people coming in, many of whom have dealt with the cancer diagnosis themselves or with a loved one, and here they are drawing out their family history. It is bringing up all of these family issues. Could they have possibly passed the mutation on to their children? Could their siblings be at risk? Did they inherit it from their mother or their father? Would their parents feel guilty that they have somehow passed on a mutation? These are all tough issues. Although it can be very frightening for people at first, my experience is that in the long term people find it empowering. They are taking charge of this and they know what their risks are. Some patients when they are diagnosed with cancer feel that now they are at risk for everything and that they could develop leukemia or melanoma. We are able to say to them that based on their profile, these are the things they are at risk for and these are the things that we should be doing. It is tailored medical management in a way that really makes sense.

Miller So if someone leaves your office knowing that they have BRCA1 or that they may have a gene related to the Lynch syndrome, this is very powerful information, but it is also powerful in terms of what they can do with it.

Matloff What I often say to people is that we are going to use this information to change the legacy in their family. We are now in control. We know where the mutation is. We know what it means and we are going to change their family story in the next generation.

Miller With that in mind let’s talk about breast cancer a little bit. A woman comes to you with a strong family history and you test her and she is positive for a mutation and has been treated for the breast cancer. How would her followup be different than another woman for example?

Matloff It depends on her age, where she is in her life and how she feels about her risks. We would want to begin screening in those women at age 25 rather than 40. Rather than have someone come in for their first mammogram at 40 when perhaps they have an advance breast cancer, we are going to start early. We start at 25, and then six months after their mammogram we offer them a breast MRI which, as you know, is a high-risk surveillance method. It is only offered to a subset of patients who are at greatest risk for breast cancer. That is where we would start. We would also be concerned about their risk of developing ovarian cancer so we...
might mention to them that birth control pills are a good option until they reach an age at which they might be ready to have their ovaries and fallopian tubes removed preventively. We can do some things to manage their risks along the way.

Miller Would that, I mean, I could have picture woman is positive saying that I want to have mastectomy and my ovaries removed now. Is that a common decision that women make, to have a mastectomy or have their ovaries removed?

Matloff It really depends on the person. I have one patient who is very bright, but she is very concerned about her risks. At age 30 after having a couple of mammograms with ambiguous findings that required a biopsy, she decided to have a prophylactic bilateral mastectomy with reconstruction. For her that was the right decision. It’s what allowed her to sleep at night. She has little children and is married and has a happy life, she wants to have a long life that is cancer free. For other women, they feel more comfortable managing their risks. They trust screening intervention and choose to be followed carefully. Perhaps they would choose a mastectomy if they were ever diagnosed with breast cancer. One thing I have learned is that this is really an individual decision.

Miller The counseling part of it sounds fascinating. What is your clinical responsibility as a genetic counselor? Can you tell us a little about some of the research that you are doing and the people in your department that are involved?

Matloff Research is such an important part of this process. You hear about people who take research from the laboratory to the bedside, we actually take our research from the bedside back to the laboratory. For example, I had a patient who called me and said that her sister had recently died of pancreatic cancer. When I took the family history, what had not been taken was that she had a paternal history on her dad's side of breast and ovarian cancer. We found that she carried a BRCA2 mutation.

Miller Is it a myth that you inherit the BRCA gene from the mother’s side?

Matloff It’s a myth. You can inherit all of these changes from your father or your mother and we can pass them on to sons and daughters with equal frequency.

Miller Thank you.

Matloff We were able to determine that in this family there was a BRCA2 mutation that had caused the people who carry it to either have breast, ovarian or pancreatic cancer. This patient mentioned to me that a year before her sisters diagnosis, she had been diagnosed with diabetes, a very severe rapid onset diabetes. We researched that from our end and found that several of our patients had the same
experience. We have written a grant with collaborators at Yale in all different departments and we are hoping to be funded to explore that further.

Miller This is a case where seeing it at the bedside is going to lead to some interesting and important science.

Matloff Absolutely.

Miller Any other research that you want to share with the audience?

Matloff One of the things that we are looking into is trying to find ways to find prostate cancer at an earlier stage in people known to be at high risk. That is another important initiative and we are always looking for new genes and new genetic mutations that cause cancer.

Miller You and I have talked a little bit about thyroid cancer. What kind of family with thyroid cancer should be screened?

Matloff First of all with thyroid cancer it is really important that the patient finds out what kind of thyroid cancer they have; is it follicular, papillary, or medullary. All different types are associated with different genes and different risks. Medullary thyroid cancer is often recognized to be hereditary and those are the patients who usually need genetic testing. It is fair to say that all patients with medullary thyroid cancer need genetic testing. With papillary and follicular cancer, it is less clear.

Miller We have covered a lot of ground. We have talked about colon cancer, melanoma, Lynch syndrome, breast cancer, and thyroid cancer. It has been a wonderful session. Ellen, I want to thank you.

Matloff It was my pleasure.

Miller On behalf of myself, and my co-host Dr. Chu who was traveling today, we would like to send you our wishes for a safe and healthy week from the Yale Cancer Center.

If you have questions, comments or would like to subscribe to our Podcast, go to www.yalecancercenter.org where you will also find past broadcasts in written form. Next week, we will discuss cancer survivorship and life after cancer with Dr. Ruth McCorkle and Tish Knopf.