

Healthline with Yale Cancer Center

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WTIC Newstalk 1080

Cancer Genetic Counseling

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This is Healthline. A joint venture of WTIC NewsTalk 1080 and Yale Cancer Center. Yale Cancer Center is a resource for cancer programs throughout Connecticut, developing new advances in prevention, screening, diagnosis and treatment. On Healthline you will hear from some of the leading doctors in the country. Healthline is not intended to provide medical advice. Yale Cancer Center urges you to consult with a qualified physician in your community for diagnosis and for answers to your medical questions. And now, our co-hosts Oncologists Ken Miller and Ed Chu.

Miller Good morning and welcome to Healthline. My name is Dr. Ken Miller, and I am the Director of the Survivorship Program at the Yale Cancer Center in New Haven. My co-host Dr. Ed Chu, who is the Chief of Medical Oncology at Yale, is traveling today and will be back with us next week. Healthline, with the Yale Cancer Center, is our way of providing you with the most up-to-date information on cancer care every Sunday morning at 8:30 a.m. Healthline features some of the nation's leading cancer specialists who are in the forefront in the battle to fight cancer right here in Connecticut. Our goals with this program are to give you help by sharing the latest information about cancer and also to give you hope, because really there is a lot of hope in the battle against cancer. If you would like to submit a question about cancer to Healthline, you can email us at Healthline@Yale.edu. If you are interested in listening to past editions of Healthline, or if you would like to learn more about a specific kind of cancer, all of our shows are now posted in audio and written format on the Yale Cancer Center website, which is www.yalecancercenter.org. Today we are going to be discussing cancer genetics and cancer genetic counseling. We have with us Ellen Matloff who is the Director of the Cancer Genetic Counseling Program at the Yale Cancer Center. Ellen, thanks for coming to join us today.

Matloff My pleasure. Thank you.

Miller We have a lot to talk about but I want to start out with a very basic question. What are genetics, and for that matter, what are genes?

Matloff Genetics is the study of heredity and genes are the most basic unit of heredity. We are talking about family history and things that run in families when we talk about genetics.

Miller In general, for people who develop cancer, is it inherited?

Matloff Most cancers are not hereditary. If you pick up a women's magazine it makes it sound like if you have breast cancer you have probably inherited it, but that is not true. 90% of cancer is in fact not hereditary.

Miller One question that patients ask me is "Do I have to worry about my children developing cancer?"

Matloff That is a great question, and it does warrant a very careful evaluation of the family history. People often say that they have no family history of cancer, but they may have not asked the right

questions. It does require some investigation. If they investigate the family history and their cancer is the only cancer in the family, then most of the time it is not hereditary. It still requires evaluation, particularly if they were diagnosed at a very young age.

Miller When you say, "Ask the right questions," what do you mean? If I wanted to get a more extensive family history, what family members would I ask and how many generations back should I go?

Matloff Ask parents, aunts and uncles, cousins, and grandparents and do not only ask about the cancer in question. For example, if you have colon cancer, don't ask specifically about colon cancer, but any other cancers that may be in the family. Also, ask at what age people were when they were diagnosed. A lot of people do genealogies and draw family trees. They collect all this information, dates of births and deaths, but they never collect medical information. Not many people know why family members died. A challenge I would like to put out for Connecticut residents listening is to make this a family project. Chart your family history and include the medical information.

Miller I have to say, I have not heard that idea before, but it makes a lot of sense. One of my relatives drew out a very fascinating family tree going back many generations, but we do not have health information.

Matloff From my biased perspective, that is the most important part. With family reunions coming up this summer and with family gatherings at Easter or Passover, this would be a great family project.

Miller I think it is a terrific idea. When did genetic testing for cancer begin?

Matloff Genetic testing for cancer has been around for about 20 years, but many of the most common genes have only been discovered and available for testing in the last decade.

Miller What is genetic testing? How do you test someone's genes?

Matloff It is very simple. We use a small blood sample. One small tube of blood will give us enough DNA to do genetic testing for most conditions.

Miller What should raise a red flag that would tell us there may be a genetic predisposition?

Matloff Right now there are six risk factors that we consider in terms of hereditary risk. This list is constantly evolving because genetics changes so quickly. The first thing to look for would be anyone in the family who had an early age of onset for their cancer; specifically breast cancer before the age of 45 or colon cancer before the age of 50. The second thing we look for is multiple cases of the same cancer in the same bloodline. Patients will come in and say that there are three ovarian cancers in their family, but when you take the history, one is on their mom's side, one is on their dad's side and one is through marriage. That is not what we are looking for. We are

looking for the same cancers within the same bloodline. The third risk factor is the combination of cancers that we know are caused by a single genetic mutation within a family. For example, breast, ovarian, and pancreatic cancer can all be caused by a single mutation; as colon, uterine, and ovarian cancer are all caused by the same mutation. This clustering of cancers is extremely important. The fourth risk factor would be a male breast cancer, because a rare cancer like that puts the family at increased risk for hereditary breast and ovarian cancer. The fifth risk factor is any family member who has had multiple primary cancers. This wouldn't be a family who has had breast cancer that has then metastasized to the bone; we are talking about someone who has had breast cancer, and then maybe a few years later, ovarian cancer. The last risk factor has to do with the fact that we know that certain genetic conditions are more common within different ethnic backgrounds. People of Jewish ancestry are at increased risk for hereditary breast and ovarian cancer. Those are the six main risk factors we look for today.

Miller Before the end of the show, I would like to have you go through those again because it is very, very useful information. Let me share with you an email that we received from Mike who lives in West Hartford, he says,

I am a 48-year-old man of Jewish ancestry. My great uncle had breast cancer, and I have read that it may run in the family.

What kind of information can we share with Mike?

Matloff I would tell Mike that it would be very important to chart out his family history. Ask about other male breast cancers, ask about breast cancer in women as well as ovarian cancer and pancreatic cancer; any other findings in the family that would be worth sitting down with a genetic counselor to discuss.

Miller For someone like Mike, what kind of genetic testing would you do? What are the genes you would look for?

Matloff In Mike's situation, because he is of Jewish ancestry and there is a male breast cancer, if it were a perfect situation we would really like to test Mike's uncle who had the breast cancer. We would be testing him for three common Jewish mutations within two genes; one called BRCA1 and the other called BRCA2. Those two genes stand for breast cancer I and breast cancer II, but the names are misleading. They really should have been called breast and ovarian cancer I and breast and ovarian cancer II, because all women who inherit a mutation in either gene are at high risk for both cancers, but it can also have an impact for men. This is an important point that I would like to make; these mutations do have an impact on men and you can inherit them from your father just as easily as from your mother. We can pass them on with equal chance to sons and daughters. So this kind of rhetoric that we hear that only your mom's family history counts is completely inaccurate.

Miller We are going to take a break now for a medical minute. If you have questions for myself, or for Ellen Matloff, who is the Director of Genetic Counseling here at Yale, please e-mail us at Healthline@Yale.edu. We will be back with you to talk more about cancer genetic counseling.

Medical Minute

This is a Medical Minute brought to you as a public service by the Yale Cancer Center. Cancer patients become cancer survivors the first day they are diagnosed. There are over 10 million cancer survivors in the US and the numbers keep growing. However, there are long-term side effects of cancer including heart problems, osteoporosis, fertility issues, impaired growth, and an increased risk of second cancers. Ending cancer treatment can be both exciting and scary. Most people are relieved to be finished with the demands of treatment but many also feel concerned about whether the cancer will come back and what they can do to prevent a relapse. Cancer survivors require long-term specialized care and support. For more information, log on to www.yalecancercenter.org.

Miller Welcome back to Healthline. This is Dr. Ken Miller. I am here with Ellen Matloff, who is the Director of Genetic Counseling at Yale, discussing some of the genetic links to cancer and issues about genetic counseling. I want to talk some more about breast and ovarian cancer later, but you brought up a very interesting point earlier about colon cancer. Can you tell us a little bit more about what you think when you see someone who has had colon cancer?

Matloff There are two main types of hereditary colon cancer. One is associated with many polyps. When I say many we are usually looking at dozens to thousands of polyps found in the colon, usually during a colonoscopy. That is one type of hereditary colon cancer. The second type is not associated with many polyps. People usually have 10 or fewer polyps, but there is often a strong family history of colon cancer, particularly at young ages, and some of the women in the family may have had uterine or ovarian cancer. There is also a skin finding called a sebaceous adenoma or sebaceous carcinoma that is very strongly linked to the syndrome. That skin finding alone can help us determine who may be at risk for hereditary colon cancer.

Miller And the skin finding goes with the second type not with the first type of colon cancer with all the polyps?

Matloff That is right. It is the type with few polyps.

Miller There is a term I have heard used called the Lynch syndrome. Who was Dr. Lynch and what are Lynch syndromes?

Matloff Dr. Henry Lynch is still alive; he is in his late 80s I believe. He is the person who coined this syndrome. Interestingly enough, when he brought up the fact that he thought some types of cancers were hereditary back in the 70's, people thought he was crazy; now he has turned out to be

correct. The Lynch syndrome is also called HNPCC standing for Hereditary Nonpolyposis Colorectal Cancer. We tend to call it HNPCC these days.

Miller Instead of Lynch syndrome.

Matloff We know that people with a mutation in one of these genes has a high risk of colon cancer as well as ovarian and uterine cancer in women.

Miller If one of our listeners is hearing this program and has colon cancer, uterine cancer and ovarian cancer in their family, and they are tested and found to be positive for this syndrome, can we fix genes? What can we do for these people other than telling them that they have this diagnosis?

Matloff We can't fix genes yet, but we can do a lot of other things that are important. For example, in these families we begin surveillance with colonoscopy at age 25, or 10 years before the first colon diagnosis in the family. Most people are not even offered a colonoscopy, as you know, until 50. By performing this early surveillance we can often find cancers at an earlier, more detectable stage. Also, in women at risk for HNPCC, when they are done having children or when they reach age 40, we can actually remove their uterus and their ovaries preventatively and reduce their risk of ever getting those cancers.

Miller So essentially we can prevent uterine and ovarian cancers, and we can detect colon cancer earlier.

Matloff That is right.

Miller There was interesting information on Celebrex many years ago being used in people. Celebrex is essentially off the market now, but can you tell us a little bit more about that?

Matloff There have been some prevention trials using aspirin-like derivatives, and also using birth control pills, to see if these will reduce polyp formation and also reduce the risk of colon cancer. Those studies are still underway, but it does appear that some aspirin-like derivatives do decrease the risk of colon cancer in these families.

Miller Aside from a very careful surveillance, perhaps we can make dietary changes that would have an impact.

Matloff I would say yes, cautiously, because even in these families for people who are eating a perfect diet, low in fat, full of vegetables and fruits, the risk of cancer is still very high.

Miller It does not replace careful surveillance in good medical care.

Matloff It does not.

- Miller Thank you for sharing that. I want to ask a little bit more about the psychosocial side of things. I care for a lot of women who have breast cancer. When I talk with women about going to see you or seeing members of your team, they have serious concerns. One is that this will in a sense brand them for when they try to get insurance in the future. Is that a myth or is that reality?
- Matloff I think it is a fair concern, and I can tell you that when we started this program 11 years ago, this was one of our concerns as well. What will happen to these people, will this cause more harm than good? The good news is that we now have more than 10 years of data and we have not seen insurance discrimination emerge as a reality for our patients. We do know that life insurance can discriminate against you because of your family history or because you carry a mutation. If people are considering genetic counseling or testing they may want to make sure that their life insurance is in place. With that being said, however, it is very common that people report to their physician that they have a strong family history of breast, ovarian or colon cancer. It's already documented on their chart. We are really not adding anything new to that equation.
- Miller By doing the actual test you haven't changed it very much. I have women I have taken care of who are positive for the BRCA gene and have breast cancer, and they have sisters or daughters who absolutely do not want to be tested. What are some of the reasons people will tell you that they do not want to be tested?
- Matloff There are a lot of different reasons that people do not want to be tested. Probably the most common reason is they are not ready for the information. The thing that concerns me is that even if they are not ready to hear the information, they either carry the mutation or they don't; the test doesn't change the medical reality. It is okay for people to take their time. I worked with a family recently where it took them several months to decide to be tested. By the time they went forward with the testing, they were ready to hear the results. It is definitely not something to rush in to and it is not something you have without having genetic counseling first. I saw a patient the other day that had the test before having the counseling and learned she carries a mutation. She really didn't know much about it and that's a difficult conversation to have with someone on a Friday afternoon at 5 o'clock. They have these results and have no idea what it means.
- Miller I want to learn a little bit more about this because it is a common scenario where people have the test and then try to figure out what it means. We are going to take a break now for a survivor's story. I encourage you to please stay with us and learn more information about genetic counseling with Ellen Matloff from the Yale Cancer Center.

Survivor Story

A few years ago, the diagnosis of cancer was a death sentence for many patients, but today, thanks to advances in clinical research, we are turning the corner in the battle against cancer. There are

over 10 million cancer survivors now living in the US. They are the true heroes in the war against cancer. Here is a story of a hero from Hamden.

Ten years ago, when I was diagnosed with aplastic anemia, there was no cure. After teaching Math for 35 years, I was forced to retire. Dr. Tom Duffy at the Yale Cancer Center told me about the new procedure called a mini stem cell transplant. He encouraged me to put my life in the hands of Dr. Stuart Seropian, one of the few doctors in the country doing this procedure. On January 17, 2004, I had a stem cell transplant at the Yale Cancer Center. At age 70, I feel like a new man. I owe a great debt of gratitude to my anonymous stem cell donor and to the terrific staff at the Yale Cancer Center. They literally saved my life.

This survivor story has been brought to you by Yale Cancer Center.

Miller Welcome back to Healthline. This is Dr. Ken Miller, and I am here in the studio with Ellen Matloff who is the Director of Genetic Counseling at the Yale Cancer Center. We are talking about genetic counseling of people with cancer and people who are concerned about the risk of developing cancer. Ellen, I want to get back to the counseling component of what you do. When a person finds out that they have a gene that predisposes them to cancer, what is that like for them, and where do you go from there?

Matloff It is a different experience for each person; even for sisters and brothers within the same family. People have a range of emotions. Some people are not surprised at all, and they say, "I knew cancer ran in my family, and now I know that I am at risk and I know what I want to do." They feel very clear about it. Other people are surprised by it. The most difficult situation is when it really knocks someone off their feet and they had no idea that they might carry a mutation. It is very normal for people to feel anxious or sad. It brings up a wide range of emotions because most of the time they have a family history and they realize, "Gosh! This is why my son got colon cancer" or "This is why my dad died," of whatever type of cancer. The one thing that I always underline to people is that there is such hope and that genetic testing is in many ways a gift that you give to your entire family to change the next generation. If people in the previous generation have developed and died of cancer, now we know and we can do something to change the future; that is a remarkable gift.

Miller It can be helpful for the individual, but it can do even more for the next generation.

Matloff That is right.

Miller That's very powerful. Do you find that after counseling people walk around with a feeling of dread, or is it more a sense of empowerment?

- Matloff What the research in this area shows is that people who test positive do have an increased rate of anxiety for about 3 to 6 months, but then about a year out that they are almost back down to baseline. It tends to be patients who postpone coming in for genetic counseling or testing who fair the worst psychologically.
- Miller Give me some advice. What might be the best way to present it to someone?
- Matloff I think it is very difficult when someone is newly diagnosed because there are so many things to talk to them about, as you know. There are also so many people that they have to go and see and the last thing they need is one more person. What I would say about genetic counseling is that if we know someone carries a mutation, it can change their surgical decision-making in the management of their cancer. For example, if a patient learns they are carrying a mutation, even if the cancer they already have could be contained and removed by lumpectomy and treated with radiation, it means that that mutation is present in every single breast cell of both breasts. That person is at high risk to develop a new cancer in the future. Some people in this situation would choose a bilateral mastectomy and reconstruction instead of lumpectomy and radiation. Knowing this information early in the process can allow that patient to take some control and to manage their own healthcare in a way that makes most sense for them. There is not one right answer for every patient, everyone chooses something different.
- Miller Listening to some of the work that you and your team do, is there a lot of science and also a lot of psychology involved? What is your background; what kind of training do people have to become a genetic counselor?
- Matloff As undergraduates we all had a science background; mine was in biology. There are some schools where you can go and get a background in molecular biology or genetics, but we all have a background in biology as undergrads. We all have our Masters degree specifically in genetic counseling, which is a blend of the hard science and the psychology.
- Miller Does health insurance cover genetic testing and counseling?
- Matloff Most of the time health insurance does cover testing and counseling, which is another misconception. People think it is not covered, but usually when it is needed, and we can write a letter of medical necessity justifying the use of the testing, it is covered.
- Miller Do patients need a referral from a primary care physician?
- Matloff It depends on their insurance, but usually not.
- Miller How would somebody go about making an appointment? What number do they call and if they are not here in New Haven, where does someone call in general to find a genetic counselor?

- Matloff Go to Google and type National Society of Genetic Counselors. A website will come up that will allow you to find a counselor in your area. If you live in the state of Connecticut, we do have Outreach Clinics all over the state and you can call us at 203-764-8400, or you can put Yale Cancer Genetic Counseling into Google and our website will come up.
- Miller And for people in other states, there are resources available to them as well. Ellen, I would like to find out what other research initiatives you and your department are working on.
- Matloff We have several exciting research initiatives that are ongoing. One of them has to do with pancreatic cancer and the hopes of finding pancreatic cancer at an earlier, more treatable stage in patients at high risk. We are also looking at some early prostate cancer surveillance in populations of men who are at high risk to develop the disease. Those are two of our really exciting studies that we are launching.
- Miller Hopefully in the same way that you have been able to find the BRCA gene, which is useful in finding women who are at high risk for breast cancer, there may be a test for prostate cancer.
- Matloff What we have found with prostate cancers is that there are many different genes involved, but prostate cancer is also associated with BRCA1 and BRCA2 in some of these families, and that's an issue.
- Miller Again, if you have questions for Ellen Matloff or for Healthline, e-mail us at Healthline@Yale.edu. Ellen, I want to thank you again for joining us on Healthline. Can you quickly review the six main reasons people should go for testing?
- Matloff If there are many family members with cancer; if they have been diagnosed at a young age; if there are clusterings of cancer; if people have had multiple cancers; if you are of Jewish ancestry and have a family history of breast, ovarian, or pancreatic cancer then you may want to look in to this a little bit more.
- Miller Terrific, thank you for sharing that. Thank you for joining us on Healthline. We want to remind our listeners to tune in to WTIC NewsTalk 1080 every Sunday morning at 8:30 a.m. for Healthline with the Yale Cancer Center. Our next program will feature a discussion on colorectal cancer in honor of Colon Cancer Awareness Month, which is March. Until then, this is Dr. Ken Miller from the Yale Cancer Center wishing you a safe and healthy week.