

Prevention *matters*

| A FOX CHASE CANCER CENTER RISK ASSESSMENT PROGRAM PUBLICATION | FALL/WINTER 2017 |

Why it is important to see a genetic counselor

A lot of people think that cancer “runs in the family,” but the truth is that fewer than 10 percent of cancers are the direct result of a genetic defect.

Still, patients are often referred for genetic counseling, because the results can help them make treatment decisions for themselves and also help family members to better understand their own risks.

A genetic counselor’s job is to help guide patients as they make difficult choices surrounding genetic testing — a DNA test done on a small sample of blood or saliva.

For Jane, diagnosed with breast cancer, genetic counseling helped her decide on a lumpectomy rather than a bilateral mastectomy. Phyllis, who had run out of treatment options for her ovarian cancer, tested positive for the *BRCA1* gene, which made her eligible for a new round of chemotherapy. Melissa, 28, whose mother had colon cancer at 45, tested negative for colon cancer risk genes, but was told to start colonoscopies at 35 rather than 50.

By going over which tests are right for each person and taking the time to review and talk over the possible results, a genetic counselor helps patients to understand information, answers any questions, and spends time listening to a patient’s hopes and worries related to genetic testing.

What should you expect when working with a genetic counselor?

- A meeting prior to genetic testing to review your medical and family history and decide whether you meet testing criteria for a specific hereditary cancer syndrome.
- An explanation of the pros and cons of the different testing options and what each test result could mean for you and your family.



Genetic counseling team above left clockwise:
Michelle Savage, Sue Montgomery, Andrea Forman
and Kim Rainey

- A review of possible cancer risks and medical management changes that could be in store once the results are in, as well as the importance of these results for other family members.
- A chance to ask questions and decide if you feel comfortable about your decision to have or not have genetic testing.
- Coordination of a blood or saliva sample to be sent to a genetic testing laboratory.
- Help with any health insurance challenges that can go along with testing.
- A wait of 2 to 6 weeks before test results are reported to the genetic counselor, who will then make sure he or she has the most up-to-date and relevant risk and medical information for you.
- A follow-up visit in person to go over the final results and to give you more information, based on those results, for screening and prevention options for you or your family members.
- A written summary of your results and visits for you to share with your doctors and family members.

➤ **Please call the Risk Assessment Program at 877-627-9684 if you have any questions about genetic testing.**

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SCREENING REMINDERS FOR BUSY WOMEN

For many women, juggling work, home life, and family leaves little time for them to take care of themselves. But taking the time to know what cancer screening tests are right for you could help save your life. The American Cancer Society® encourages you to talk with your doctor about screening tests for breast cancer, cervical cancer, colon cancer, and lung cancer.



BREAST SCREENING recommendations for women at AVERAGE risk

- Women between 40 and 44 have the option to start screening with a mammogram every year.
- Women between 45 and 54 should get mammograms every year.
- Women 55 and older can switch to a mammogram every other year, or they can choose to continue yearly mammograms. Screening should continue as long as a woman is in good health and is expected to live at least 10 more years.
- All women should be familiar with how their breasts normally look and feel and report any changes to a doctor right away.

BREAST SCREENING recommendations for women at HIGHER risk

- Women with Lobular carcinoma in situ (LCIS) OR Atypical hyperplasia or personal history of breast cancer OR dense breast tissue should get a mammogram every year and talk to the health care provider about MRI screening.
- Women who have a *BRCA1/2* gene mutation OR a first-degree relative with a *BRCA1/2* gene mutation but have not been tested for mutations themselves OR had radiation treatment to the chest between ages 10-30 OR have Li-Fraumeni syndrome, Cowden syndrome, or Bannayan-Riley-Ruvalcaba syndrome in themselves or in first-degree relatives OR have a higher risk of breast cancer based on family history should get a mammogram and MRI every year starting at age 30 or age recommended by doctor.

CERVIX SCREENING

- Women should start being tested for cervical cancer at age 21.
- Women between the ages of 21 and 29 should have a **Pap test** every 3 years.
- Women between 30 and 65 should have a Pap test plus an **HPV test** every 5 years (or a Pap test by itself every 3 years).
- Women over age 65 who have had regular cervical cancer testing and have had normal results can stop getting Pap tests.

The Pap test is a procedure used to collect cells from the cervix so that they can be looked at under the microscope to find cancer and pre-cancer.

If a woman has a history of cervical pre-cancer, she should continue to be tested for at least 20 years after that diagnosis, even if testing continues past age 65.

HPV is short for human papilloma virus. HPVs are a group of more than 150 related viruses. HPVs are called papilloma viruses because some of the HPV types cause warts or papillomas, which are non-cancerous tumors. But some types of HPV are known for causing cancer, especially of the cervix (the base of the womb at the top of the vagina).

COLON SCREENING

This cancer can develop in men and women. Testing can help prevent some colon cancers by finding and removing pre-cancerous polyps to stop cancer before it starts. Beginning at age 50, women should start having regular colorectal cancer screening tests. Since several types of tests are available, women should ask their doctor which tests are right for them.

If a woman is at an INCREASED or HIGH risk of colorectal cancer, she might need to start colorectal cancer screening before age 50 and/or be screened more often. The following conditions make risk higher than average:

- A personal history of colorectal cancer or adenomatous polyps
- A personal history of inflammatory bowel disease (ulcerative colitis or Crohn's disease)
- A strong family history of colorectal cancer or polyps
- A known family history of a hereditary colorectal cancer syndrome such as familial adenomatous polyposis (FAP) or Lynch syndrome (hereditary non-polyposis colon cancer or HNPCC)

➤ **CONTINUED ON NEXT PAGE**



New mutations that increase risk for breast and ovarian cancer found analyzing a panel of 25 genes in women

A new study shows the level of risk associated with a given gene mutation which will help doctors and patients make decisions about medical care.

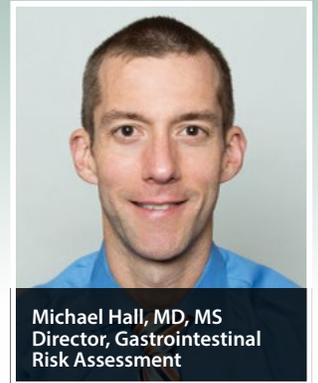
Samples from 95,561 women with and without cancer were analyzed using a panel test of 25 cancer related genes. One or more mutations associated with higher risk of breast and ovarian cancer were detected in 7% of women tested. Eight genes were associated with higher breast cancer risk: ATM, BARD1, BRCA1, BRCA2, CHEK2, PALB2, PTEN, and TP53. The highest risk was associated with BRCA1 and the lowest risk with ATM.

The study did not show a higher risk of breast cancer among people with Lynch syndrome genes MSH6 and PMS2 as was recently reported in another smaller study.

Eleven genes were associated with higher ovarian cancer risk: ATM, BRCA1, BRCA2, BRIP1, MLH1, MSH6, MSH2, NBN, RAD51C, RAD51D, and STK11. The highest risk was with STK11 and the lowest risk with ATM. This is the first report of higher ovarian cancer risk associated with an ATM mutation. It is estimated that as many as 1% of

breast cancer patients and 0.5% of the general population may carry an ATM mutation. Many of these patients have no family history of ovarian cancer; therefore, defining their risk of developing ovarian cancer is an important research priority.

The results from this study may help doctors, nurses and genetic counselors to guide patients at a higher risk for cancer make screening and prevention decisions. "As more patients with cancer and at risk of cancer get access to genetic testing, we will gain a more comprehensive view of which genes have an impact on cancer risk and how large those risks are. Some genetic mutations will necessitate increased screening, but others may be low enough that we don't need to do more than standard prevention and early detection," said Dr. Hall, one of the investigators and co-author of the article.



Michael Hall, MD, MS
Director, Gastrointestinal
Risk Assessment

[Read the full article at
www.ascopubs.org/doi/pdf/10.1200/PO.16.00066](http://www.ascopubs.org/doi/pdf/10.1200/PO.16.00066)

September is National Ovarian Cancer Awareness Month

October is National Breast Cancer Awareness Month

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LUNG SCREENING

Screening for lung cancer is recommended for those who are at a high risk for lung cancer due to cigarette smoking. Talk to your doctor about screening if you meet the following:

- Are between 55 and 74 years old
- Are in good health, with no signs of lung cancer
- Have at least a 30 pack-year smoking history AND are still smoking or quit within the past 15 years. (A pack-year is the number of cigarette packs smoked each day multiplied by the number of years a person has smoked. Someone who smoked a pack of cigarettes per day for 30 years has a 30 pack-year smoking history, as does someone who smoked 2 packs a day for 15 years.)

Learn about lung cancer screenings at Fox Chase Cancer Center

www.foxchase.org/clinical-care/conditions/lung-cancer/lung-cancer-screening



November is Pancreatic Cancer Awareness Month

PurpleStride: Pancreatic Cancer Network Action Walk/Run

Saturday, November 4, 2017 6:00am-12:00pm / Memorial Hall in Fairmount Park, Philadelphia, PA

To join the Fox Chase Cancer Center team, please register

http://support.pancan.org/site/TR/PurpleStride/PurpleStride/1090898754?pg=team&fr_id=1310&team_id=7467

FINDING HARMFUL HEREDITARY MUTATIONS IN PANCREATIC CANCER PATIENTS WITH NO FAMILY HISTORY OF PANCREATIC CANCER

Inherited mutations in genes such as *BRCA2*, *ATM*, *PALB2*, *CDKN2A*, *PRSS1*, *STK11*, *MLH1*, and *MSH2* are known to increase pancreatic cancer risk in patients with a family history of the disease. However, little is known if these harmful gene mutations contribute to the pancreatic cancer risk in patients with no family history of pancreatic cancer.

In a recent study, researchers from Johns Hopkins Hospital analyzed 32 genes in 854 patients with pancreatic adenocarcinoma and 288 patients with other pancreatic and periampullary tumors.

Harmful hereditary mutations were found in 33 (3.9%) of 854 patients with pancreatic cancer. 31 of 33 patients had a mutation in well-known pancreatic cancer genes (*BRCA2*, *ATM*, *BRCA1*, *PALB2*, *MLH1*, *CDKN2A* and *TP53*). Two patients had a mutation in genes that may increase the risk of pancreatic cancer (*BUB1B* and *BUB3*). Patients with these mutations were younger than those without. The majority of patients did not report a family history of cancer that would suggest a hereditary cancer syndrome. Five (1.7%) of 288 patients with other periampullary tumors also had a harmful hereditary mutation.

For common cancers such as breast, ovarian and colorectal cancers that have well-established genetic counseling and testing guidelines, family history is used to identify family members at risk of cancer who will benefit from genetic testing. The results of the study show the limitation of this approach for pancreatic cancer patients. Since most mutations were found in patients who do not have a family history of the disease and do not meet criteria for genetic testing, these mutations would have been missed if testing had just been based on family history.

It would be valuable to examine ways to incorporate risk assessment and genetic testing into routine pancreatic cancer practice. Genetic testing can have an impact on patients' treatment, as patients with specific mutations can be treated with a personalized therapy, immunotherapy, or radiotherapy to control local disease. It also helps family members who are at risk make decisions about their cancer screening.



Read the full article at
www.ascopubs.org/doi/full/10.1200/JCO.2017.72.3502

FERTILITY PRESERVATION IN CANCER PATIENTS

Sharon Schwartz, Certified Registered Nurse Practitioner, provides survivorship care to gynecologic oncology patients. She also cares for women with a genetic predisposition and/or family history of cancer. Sharon also serves as the oncofertility coordinator for Fox Chase Cancer Center



What is fertility preservation in cancer patients?

Fertility preservation is the effort to help cancer patients protect their ability to biologically conceive in the future, or ability to have children.

Cancer treatment can have a big impact on fertility. Certain types of chemotherapy can cause early menopause in women and/or infertility. Surgical procedures, such as having the ovaries or uterus removed, can prevent patients from carrying biological children. Radiation treatments also have an impact, especially for patients who have radiation to their pelvic area.

What is the role of the oncofertility coordinator?

I serve as the point person for the oncofertility referral service at Fox Chase to help patients with fertility issues caused by cancer treatment. At the treating physician's request, I contact patients to discuss resources and information. I can also coordinate referrals and appointments so people are able to have all options on the table before moving forward with their cancer treatment.

What are the options for women diagnosed with cancer to preserve the ability to have children?

Depending on the patient's gender, age, partner status, type of cancer, available time, and future reproductive goals, different options for fertility preservation are available. A consultation with a reproductive endocrinologist should be offered to all cancer patients who have not completed childbearing. Some options recommended for women are: freezing embryos (fertilized eggs), freezing unfertilized eggs or surgery and radiation therapy given in specific ways to avoid harming a woman's reproductive organs. Women may have their ovaries moved to a different part of the abdomen before pelvic radiation. For men, the most common and successful option is sperm banking.

How long does the fertility preservation process take?

Fertility preservation methods can take as little as 24 hours for men and for women take 2 to 4 weeks. Timely referral to a reproductive specialist is very important.

What are the barriers for patients to proceed with fertility preservation?

The largest barrier is typically cost. I work with people to identify financial assistance that may help bring down the costs of freezing eggs. Additionally, Fox Chase has relationships with fertility clinics in the Philadelphia area that offer a discount for Fox Chase patients.

Is this an option for a woman without cancer but who has a genetic predisposition to cancer, such as a BRCA mutation?

Yes. Women with *BRCA* mutations are advised to have preventative surgery to remove their ovaries and fallopian tubes (bilateral salpingo-oophorectomy, or BSO) before they would naturally reach menopause, some as early as ages 35-40. By this age, many women have not completed childbearing. Women with a *BRCA* mutation could consider freezing eggs or embryos before having BSO. There are some potential grants to help women with *BRCA* mutations who want to preserve fertility.



For men and women with a *BRCA* mutation, with or without cancer, there is an option to have pre-genetic diagnosis (PGD) for their future children. This is something that is done before pregnancy. The process involves participating in In-vitro fertilization (IVF). The female partner takes medication to make many eggs in one cycle. The eggs are not ovulated naturally but taken out via an egg retrieval process and then combined with the male partner's sperm.

Once fertilized and developed, the embryo will reach a certain number of cells, one of which will be able to be sent to the lab for testing. The lab can then test that cell for the *BRCA* gene mutation in the family. The couple can then decide if they only want to use the embryos without the *BRCA* mutation. Patients interested in this option should meet with a fertility specialist prior to getting pregnant to discuss the procedure and costs involved. This is not typically covered by insurance.



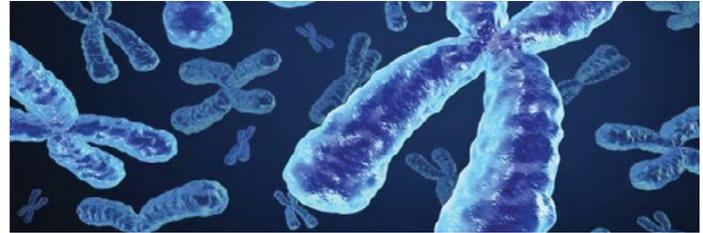
MAN 2 MAN



GENETIC EVALUATION OF MEN (GEM) STUDY UPDATE

The GEM study includes men at high risk for prostate cancer (based on family history) and men that have been diagnosed with prostate cancer. The study takes place at Fox Chase Cancer Center and Thomas Jefferson University Hospital. Participants at Fox Chase watch a short video narrated by a genetic counselor which explains what genetic testing is and what can be expected. Men are able to ask questions and sign consent. A blood sample is collected and sent to be evaluated. A multigene panel test is performed to look for genetic mutations or alterations known to be associated with a risk for cancer.

We are very excited that our study, Genetic Evaluation of Men (GEM), has had its first publication titled, "Inherited Mutations in Men Undergoing Multigene Panel Testing for Prostate Cancer: Emerging Implications for Personalized Prostate Cancer Genetic Evaluation". The publication focused on our first 200 men and the results of their panel testing. The majority of mutations were found in genes involved in DNA repair (*BRCA1*, *BRCA2*, *ATM*, *BRIP1* and *MSH6*). DNA repair is a collection of processes by which a cell identifies and corrects damage to the DNA molecules that encode its genome.



This is important for men with prostate cancer because recent data suggests that men with metastatic disease can be treated with therapies based on their mutation status. These findings are also important for men at high risk for prostate cancer because guidelines at this point only address prostate cancer screening for men with *BRCA* mutations. Our study identifies other cancer gene mutations in unaffected high risk men. The genetic testing results show additional cancer risks for the men and their families, such as colon, pancreatic, melanoma and male breast cancer based on their mutation.

We want to thank all the men that participated in our study!

Read the full article at www.ascopubs.org/doi/full/10.1200/PO.16.00039#



PHYSICIAN'S CORNER



The FDA (Food and Drug Administration) approved the immunotherapy drug Keytruda for patients whose cancer has a certain genetic change (biomarker). This is the first time the FDA has approved a treatment based on a biomarker rather than a cancer location in the body.

Keytruda (pembrolizumab) can now be used for adults and children with tumors that cannot be removed through surgery or metastatic solid tumors with specific genetic change that have high levels of microsatellite instability (MSI-H) or mismatch repair deficiency (dMMR). Keytruda will be available only to patients whose cancer progressed after previous therapy and who have no

treatment alternatives. MSI-H and MMR tumors have abnormalities that affect the proper repair of DNA in a cell. These types of tumors are found mostly in colorectal, endometrial and gastrointestinal cancers. Keytruda works by helping the body's immune system recognize cancer cells and attack them. It has already been approved for treating people with some types of lung, head and neck, bladder, skin cancers, as well as Hodgkin's lymphoma.

A total of 149 patients with 15 cancer types, with MSI-H or dMMR tumors, were enrolled across five clinical trials. The most common cancers were colorectal, endometrial, and other

gastrointestinal cancers. The primary measures of Keytruda's effectiveness were complete or partial shrinkage of their tumor (overall response rate) and the length of their response. 39.6% of patients had a complete or partial response. For 78% of those patients, the response lasted for six months or more. Common side effects of Keytruda include fatigue, itchy skin, diarrhea, decreased appetite, rash, fever, cough, difficulty breathing, musculoskeletal pain, constipation and nausea.

Keytruda was approved using accelerated approval, a special process the FDA uses to speed up the availability of drugs to treat serious diseases. Keytruda is manufactured by Merck & Co.

Read the full article at www.fda.gov/newsevents/newsroom/pressannouncements/ucm560167.htm

HEALTHY RECIPE

Sweet Potato and Spinach Turkey Burgers

The secret to these flavorful turkey burgers is the aromatic combination of fresh herbs, onion, and garlic mixed with mashed sweet potato and spinach. Filled with extra veggies, this burger makes following AICR's recommendations to eat more plant foods and less red meat easy to follow. Plus the fresh herbs are loaded with an abundance of phytochemicals that promote good health and may help prevent inflammation.

Ingredients:

- 1 medium sweet potato, cut into 3/4-inch chunks (about 2 cups)
- 1 lb. lean ground turkey
- 2 cups medium packed fresh spinach, chopped small
- 1 small onion, finely chopped
- 2 large cloves garlic, minced
- 2 tsp. finely chopped fresh rosemary
- 2 tsp. finely chopped fresh sage
- 2 tsp. finely chopped fresh thyme
- 3/4 tsp. salt
- 1/2 tsp. black pepper
- 1 Tbsp. extra virgin olive oil
- 6 whole-grain buns or 6 large lettuce leaves
- Cooking spray

Microwave sweet potato 4-6 minutes or steam 15 minutes until tender.

While sweet potato is cooking, prepare grill and set heat on medium-high. If broiling, set top rack on second rung (at least 6 inches from broiler) and set heat on broil. Prepare large broiler pan with cooking spray.

In large bowl, mash sweet potato. Add turkey, spinach, onion, garlic, herbs, salt, pepper and oil. Mix together and form 6 patties about 1/2-inch thick.

Grill patties 4-8 minutes on each side or until center is 165 degrees F. If broiling, arrange patties on broiler pan and broil 4-8 minutes on each side or until center is 165 degrees F.

Makes 8 Servings

Per serving: 134 calories, 7 g total fat (1.5 g saturated fat), 7 g carbohydrate, 12 g protein, 1 g dietary fiber, 351 mg sodium.

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PROTECT YOUR BRAIN

A new study at Temple University shows extra-virgin olive oil may protect the brain against Alzheimer's disease and memory loss. In the study researchers show that eating extra-virgin olive oil protects memory and learning ability and reduces formation of a certain type of plaque and neurofibrillary tangles (a protein known as a marker in Alzheimer's disease) in the brain.

Researchers found that olive oil reduces brain inflammation and activates a process known as autophagy. Autophagy is the process by which cells break down and clear out debris and toxins.

Read the full article at <https://medicine.temple.edu/news/extra-virgin-olive-oil-preserves-memory-and-protects-brain-against-alzheimers-disease-new>

Prevention *matters*

The Department of Clinical Genetics offers one of the most comprehensive risk assessment programs in the Philadelphia region. It encompasses all of Fox Chase Cancer Center's clinical services for people at risk for cancer, as well as innovative research in the areas of cancer prevention and genetics.

CONTACT THE RISK ASSESSMENT PROGRAM:

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** We will continue to mail a paper copy for those who do not use email*



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TEMPLE HEALTH



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