



The MSKCC Pancreatic Tumor Registry

Newsletter #3: Summer 2012

Happy Summer from the Pancreatic Tumor Registry Staff!

In our third issue of the MSKCC Pancreatic Tumor Registry newsletter, we focus on two members of the **Clinical Genetic Service (CGS)** at MSKCC, Dr. Zsofia Stadler and Ms. Erin Salo-Mullen. We know that some pancreatic cancer diagnoses could be related to a family history of cancer. For some families with pancreatic cancer, we can identify genetic mutations that are responsible for the cancers in the family. For people in these families, meeting with a genetic counselor and a physician can be very helpful in finding out their individual risk and what steps can be taken to reduce cancer risk or find it at an early stage. We sat down with Dr. Stadler for a brief question and answer session about her work and the genetic counseling and testing process.



MEET THE INVESTIGATORS



Zsofia K. Stadler, MD

Dr. Zsofia Stadler is a medical oncologist with a dual appointment in the Clinical Genetics and the Gastrointestinal Medical Oncology Services at MSKCC. She completed her medical training at Weill Cornell Medical College followed by an internal medicine residency and medical oncology fellowship at Harvard University.

Her clinical focus at MSKCC is the diagnosis, treatment, and management of patients and families with an inherited genetic predisposition to gastrointestinal cancers. Her research interests include the identification of new cancer susceptibility genes through the emerging field of cancer genomics. Dr. Stadler's research accomplishments have been recognized by the American Society of Clinical Oncology through a Career Development Award and she was recently selected for a Clinical Investigator Award by the Damon Runyon Cancer Research Foundation.



Erin E. Salo-Mullen, MS, CGC

Erin Salo-Mullen is a genetic counselor specializing in gastrointestinal hereditary cancer predisposition syndromes at MSKCC. Erin trained at the University of Texas Health Science Center and M. D. Anderson Cancer Center. She holds certification through the American Board of Genetic Counseling (ABGC).

One of Ms. Salo-Mullen's primary roles is to work directly with the three familial gastrointestinal cancer registries at MSKCC: the Pancreatic Tumor Registry, the Hereditary Colorectal Cancer Family Registry, and the Early-Onset and Familial Gastric Cancer Registry. In addition to clinical care and registry collaborations, Erin has worked on several clinical cancer genetics research projects.

Are all pancreatic cancers genetic?

While all cancers are caused by genetic changes in tumor cells, most cancers (and most pancreatic cancers) are not hereditary. In other words, they are not due to genetic changes that are passed down in one's family. Most pancreatic cancers occur by chance, although we do know that certain risk factors such as advancing age, cigarette smoking, and a history of chronic pancreatitis (inflammation of the pancreas) or diabetes may increase an individual's chance of developing pancreatic cancer. Recent studies have indicated that obesity may also increase risk.

Are there any signs that the pancreatic cancer present in a family is hereditary?

Yes. Some signs we look for are cancers at young ages (i.e., before age 50), more than one cancer in an individual (i.e., a woman having breast cancer and pancreatic cancer), or several family members in different generations with the same types of cancer.

Who should consider genetic counseling and testing for hereditary pancreatic cancer?

Genetic testing is most informative when it is first done on the person in the family who has cancer. People with pancreatic cancer should consider genetic testing if:

- they were diagnosed at a young age;
- they have close family members who also had pancreatic cancer;
- they have a personal or family history of:
 - early-onset breast cancer (breast cancer diagnosed before age 50),
 - ovarian cancer,
 - early-onset colorectal cancer (colorectal cancer diagnosed before age 50),
 - early-onset melanoma (melanoma diagnosed before age 50), or
 - early-onset and recurrent pancreatitis.

Although genetic testing can be performed on someone who does not have a personal cancer diagnosis but does have a strong family history of pancreatic cancer, such testing is generally less informative and more costly, and may not be covered by insurance.

How is genetic testing performed?

The first step to genetic testing is meeting with a genetic counselor. Erin Salo-Mullen, MS, CGC, a genetic counselor on the Clinical Genetics Service, works directly with the Pancreatic Tumor Family Registry and sees most of the patients with familial pancreatic cancer. During the genetic counseling consultation, she reviews medical and family history information and uses this to determine which, if any, type of genetic testing is recommended. Most genetic tests are performed on blood samples (taken at the time of the genetic counseling consultation), but some are performed on cancer tissue (taken during a previous surgery or biopsy). When test results are available, another meeting is held to review the results and next steps. Each patient also has the chance to meet with one of the physicians on the Clinical Genetics Service to discuss cancer risks and receive personalized recommendations for cancer screening and prevention based on family history and genetic test results.



What are some possible implications of abnormal genetic test results?

For the person who has pancreatic cancer, an abnormal genetic test result might change the type of chemotherapy that is recommended, or it might make a patient eligible for certain new drug trials.

If an abnormal genetic test result is found, we often recommend that a patient's close family members (children, siblings, and parents) undergo their own genetic testing with the help of a genetic counselor. If a family member is also found to have an abnormal genetic test result, medical management recommendations for that person might include:

- having additional cancer screening exams (e.g., breast MRI or colonoscopy),
- considering joining the investigational pancreatic cancer surveillance program through the MSKCC Pancreatic Tumor Registry,
- considering cancer preventing surgeries, and
- changing lifestyle to reduce risk, such as stopping smoking.

The recommendations for cancer screening and prevention are based on the individual's genetic test results and careful evaluation of the family history. They are unique to each patient and his or her family members.

Do insurance companies pay for the genetic counseling consultation and genetic testing?

Most insurance companies pay for the genetic counseling consultation and genetic testing, although some companies have specific eligibility criteria. To see if genetic counseling and genetic testing at MSKCC would be paid for by your insurance, the first thing to do is to give our genetic counselors basic information about your personal and family history of cancer (see page 4 for details on the Family History Questionnaire). Then, the genetic counselor can provide you with the information that your insurance company may need to determine if the tests are covered by your specific insurance plan.

Can an insurance company discriminate against patients based on their genetic test results?

No. A federal law, called **GINA (Genetic Information Non-discrimination Act)**, provides protection against health insurance companies and employers from discriminating against people based on their genetic test results.

What specific research is being done at MSKCC in pancreatic cancer genetics?

Pancreatic cancer genetics is an area of active research at MSKCC with a number of physicians and scientists focused on trying to improve our understanding and current management of hereditary pancreatic cancer.

- Within the Clinical Genetics Service, together with **Dr. Mark Robson**, CGS Clinical Director, and **Dr. Kenneth Offit**, CGS Chief, we have made major contributions to the understanding of familial pancreatic cancer. In the Ashkenazi Jewish population, Dr. Robson studied the occurrence of *BRCA1* and *BRCA2* mutations in patients with pancreatic cancer and then studied how common these mutations are in families that had a history of both pancreatic cancer and breast cancer. We also studied how often mutations occur in ***PALB2***, a gene that may play a role in breast and pancreatic cancer, in patients who had both of these cancers. Our current studies are focusing on the occurrence of *BRCA1* and *BRCA2* mutations in more diverse populations of breast-pancreas cancer families.
- On the clinical side, **Dr. Eileen O'Reilly**, together with **Dr. David Kelsen** and **Dr. Maeve Lowery**, from the Gastrointestinal Medical Oncology Service of MSKCC, is starting clinical trials to study a new class of anti-cancer drugs called **poly(ADP-ribose) polymerase (PARP) inhibitors**, in combination with other chemotherapies, in the treatment of patients with advanced pancreatic cancer who also have *BRCA* or *PALB2* mutations. PARP inhibitors have been shown to be helpful in patients with advanced *BRCA*-associated breast and ovarian cancer, with initial studies also showing promise in pancreatic cancer. These two studies, funded by the National Cancer Institute and the Lustgarten Foundation, are currently enrolling patients and hold the potential for improving the treatment of *BRCA/PALB2*-associated pancreatic cancer.

Other researchers at MSKCC are studying how inherited genetic variation may affect someone's likelihood of getting pancreatic cancer. In contrast to the studies of rare mutations in genes known to strongly increase risk, these studies look at genetic variants that are common in the population but lead to only small increases in risk.

- **Robert Klein, PhD**, a computational biologist at MSKCC, and **Sara Olson, PhD**, an epidemiologist at MSKCC, are conducting research to understand how inherited genetic variation may affect an individual's predisposition to pancreatic cancer and the course of the disease. Dr. Klein has performed genome-wide association studies that screen samples from a control population and from patients with pancreatic cancer to identify genetic changes that are linked with cancer risk or progression. Dr. Olson has joined with other investigators in large studies sponsored by the National Cancer Institute that have identified several genetic regions that are important to pancreatic cancer development. Dr. Olson has also studied genetic pathways, particularly those related to immune function, that influence risk and survival.

If patients are interested in scheduling a genetic counseling consultation, how do they do this?

The first step in scheduling a genetic counseling consultation is to fill out the **CGS Family History Questionnaire (FHQ)**. The FHQ can be requested by calling CGS or can be found online (see CGS contact information below). FHQs should be returned to CGS via postal mail or fax. Once the FHQ is received, one of the CGS staff members will review it and then contact the patient to schedule an appointment. Questions about health insurance coverage can only be addressed after the FHQ has been returned to the CGS office. For general questions, genetic counselor Erin Salo-Mullen can be reached at 646-888-4060.

Contact the MSKCC Clinical Genetics Service

- **CGS Main Line** | 646-888-4050 **Erin Salo-Mullen, MS, CGC** | 646-888-4060
- **Online** | <http://www.mskcc.org/cancer-care/hereditary-genetics/clinical-genetics-service-pre-visit>
- **Fax** | 646-888-4051 / ATTENTION: ERIN SALO-MULLEN
- **Postal Mail** | Erin Salo-Mullen, MS, CGC
1275 York Ave. Box 295
New York, NY 10065

Contact the MSKCC Pancreatic Tumor Registry

- **Robert C. Kurtz, MD, Principal Investigator**
Participants may call Dr. Kurtz's office to schedule appointments at **212-639-7620**.
- **Amethyst Saldia, Research Study Assistant**
Contact Amethyst with questions about the study and eligibility, to update contact information and participant status, and for other pancreatic cancer resources.
Main Line | 646-735-8194
E-mail | FPCRegistry@mskcc.org
Online | www.mskcc.org/mskcc/html/75408.cfm
Postal Mail | Amethyst Saldia
MSKCC Department of Epidemiology
307 East 63rd St, 2nd Floor
New York, NY 10065

You received this newsletter because you are enrolled in the MSKCC Pancreatic Tumor Registry study.