MOUNT SINAI HOSPITAL
Joseph and Wolf Lebovic Health Complex
Zane Cohen Centre for Digestive Diseases

NETWORK

Familial Gastrointestinal Cancer Registry

Spring 2015

A Message from the Director

Dear friend,

I am most pleased to share with you the Spring 2015 edition of our Familial Gastrointestinal Cancer Registry Network.

At the Zane Cohen Centre for Digestive Diseases we are fortunate to have an outstanding team of well over 40 national and international researchers and clinicians whose varied work is reflected in this edition. We have also featured some of our newer team members in the *Meet and Greet* section and have highlighted our Education Night for Families with Lynch Syndrome, coming up on May 27th 2015. We are also planning an Education Night for Families with Familial Polyposis, which will be upcoming in the Fall of 2015.

I do hope that you find the research updates informative, and the patient support material instructive. We very much look forward to your feedback. Please feel free to contact us to learn more about any opportunities for donations to support our work. Our work is centered on you, and for the generations to come.

Dr. Zane Cohen
Director
Zane Cohen Centre for Digestive Diseases

Dr. Zane Cohen (centre, front)
with members of the FGICR and IBD teams.

Education Night Meetings in 2015

May 27 2015: Education Night for Families with Lynch Syndrome
&
NEW in Fall 2015: Meeting for Families with Familial Polyposis

See inside for details (Page 9)

RESEARCH UPDATE

New Gene Causing Hereditary Mixed Polyposis Syndrome in Individuals of Ashkenazi Jewish Descent

Melyssa Aronson, MS (C)CGC, Senior Genetic Counsellor

Hereditary Mixed Polyposis Syndrome (HMPS) is a genetic condition that increases the risk to develop multiple polyps in the GI tract. Polyps are non-cancerous growths and there are many different types of polyps. Some types, like adenomatous polyps, have a higher risk to become cancer, while other polyps like hyperplastic polyps have a much lower cancer risk. Mixed polyposis refers to the fact that individuals with HMPS can get all different types of polyps.

A new gene has been linked to HMPS in families that are of Ashkenazi Jewish descent. Researchers in London England met a 28-year old man who had HMPS. They tracked his family for 40 years and created a very large family tree, over 5 generations with hundreds of relatives. The family originated in Lithuania and was traced back to a marriage in 1897.[1] Relatives from this family live all over the world and have been found to have a strong family history of multiple polyps and early onset colon cancer. Most relatives presented with polyps around age 40, some younger at age 23 and some older at age 65.

This family was studied for many years trying to figure out what gene might be causing the polyps and cancer. In 2012, Dr. Emma Jaeger and her team published an article showing that affected relatives in this family had a mutation in a gene called *GREM1*.[2] The mutation is passed down from parent to child in a dominant way, meaning there is a 50-50 chance of inheriting the syndrome if your parent is affected. At this time, no other cancers outside of colorectal cancer have been associated, but the syndrome is newly described and we may learn more about it in the future. Screening of the GI tract is recommended when we find a *GREM1* mutation.

Genetic testing is now available to individuals of Ashkenazi Jewish descent with mixed polyposis. If you are interested in learning more about this condition, you may contact us at fgicr@mtsinai.on.ca.

1. Whitelaw et al, Gastroenterology, 1997;112 (2):327–334
2. Jaeger et al, Nat Genet. 2012;44(6):699-703

Family Reaches a Diagnosis Through FGICR Research Testing

Ashton Connor, MD (pictured left)

Jordan Lerner-Ellis, PhD, FACMG (pictured right)
Head & Director, Advanced Molecular Diagnostics, Pathology & Laboratory Medicine, Mount Sinai Hospital

An answer has been found for a family with a strong history of gastrointestinal cancers. The family had genetic testing through the Zane Cohen Centre and no syndrome was identified to explain the cancers in the family. The family agreed to be part of a research study where their sample was tested with a new high-powered technology called next generation sequencing (NGS); a technology that gives results in greater detail, in less time and at a lower cost than with previous methods. Using this new technology, research exome sequencing was performed, which is essentially a way of reading their entire genetic code.

In our research, we discovered that the family has a new change in the MSH6 gene. The MSH6 gene is known to cause Lynch syndrome, the most common inherited colorectal cancer predisposition syndrome. This diagnosis has important implications in the care and treatment of affected family members. The family kindly gave us permission to publish this finding in the journal Familial Cancer. This paper also reviews the literature to date on the clinical applications of NGS from other "early adopters" in North America and Europe.

NGS technology has been used in gene discovery research to search for new links between diseases and genes. It was only recently that NGS has moved to the clinical diagnostic laboratory, rather than solely for research. The Laboratory for Advanced Molecular Diagnostics at Mount Sinai Hospital was amongst the first labs in Canada to incorporate the new technology into the diagnostic setting. Today it is being used to diagnose mutations in the *BRCA1/2* breast/ovarian cancer genes, and will soon be used to diagnose mutations in the colorectal cancer genes as well.

Along with the ability to sequence DNA at great speed and detail, there has come an overwhelming amount of information from the genome. Storing and interpreting the meaning of this data is now the most daunting challenge faced by labs using this technology. To face this challenge we participated in establishing a national database to centrally store this information, catalogue the genetic changes found, and foster discussion on how to properly interpret them in the context of disease. This resource, the Canadian Open Genetics Repository (opengenetics.ca), will be crucial to managing the genetic well-being of Canadians now and in the years to come.

Learning from Long-Term Survivors of Colorectal Cancer

Tae Hart, PhD, Psychosocial Research Lead

Colorectal cancer (CRC) is the third most common type of cancer and second most common cause of cancer-related deaths for both men and women. Information about the long-term physical and psychological symptoms, symptom management problems, and their effect on quality of life (QOL) after treatment for CRC is not readily available. The lack of information directly affects survivorship care planning, which must take potential long-term effects (e.g., depression, bowel dysfunction, fatigue) and their treatment as well as symptom management into consideration.

Dr. Tae Hart, along with co-investigators Drs. Steve Gallinger, Michelle Cotterchio, and Nancy Baxter, just completed recruitment for a project funded by the Canadian Institute of Health Research to describe physical and psychological symptom severity, symptom management problems, and QOL in CRC survivors who were diagnosed more than 10 years ago. Dr. Hart's team recruited participants for this study from the Ontario Familial Colorectal Cancer Registry (OFCCR), which is a population-based registry of CRC survivors (diagnosed at least ten years prior) and their age-and gender-matched unaffected controls. To date, 295 CRC survivors and 233 healthy controls completed self-report questionnaires assessing physical symptoms, psychological symptoms, symptom management, and QOL. Dr. Hart's team is analyzing these data and plans to disseminate the findings to both the CRC community as well as the scientific community later this year. The data from this study have the potential to improve both CRC treatment decision-making, as well as long-term survivorship care planning.

Lynch Syndrome Testing Helps to Determine Best Surgery for Young Patients with Colorectal Cancer

Rob Gryfe, MD, PhD, FRCSC, Colorectal Surgeon

Colorectal cancer is unfortunately quite common in western society; however, it is usually diagnosed in people who are over age 50 years. It is quite rare for young adults and teenagers to develop colorectal cancer. When diagnosed in younger people, this can be suspicious for an inherited predisposition to cancer, such as Lynch syndrome (also known as Hereditary Non-Polyposis Colorectal Cancer, HNPCC).

When someone is found to have colorectal cancer, we must consider what type of surgery will be best for them. An "extended resection" involves removing more of their colon than a "standard resection". If someone is known to have a significantly high risk for getting another cancer in their colon, an extended resection may be the better option, since there will be less at-risk colon. However, the decision to remove more of the colon must also be weighed against the possibility of poor bowel function with increased numbers of bowel movements, bowel urgency or diarrhea.

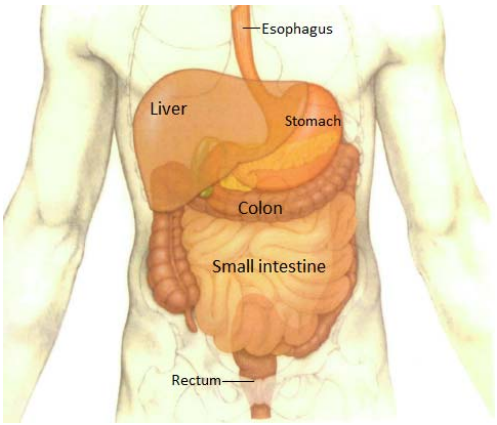
To find out more about young people with colorectal cancer and gain insight on how best to treat their cancer, we looked at data from the Familial Gastrointestinal Cancer Registry (FGICR). We identified 285 people who were diagnosed with colorectal cancer at age 35 years or younger, who did not have polyposis.

Among this group of people with colorectal cancer at age 35 or younger we found:

- **38% had Lynch syndrome**
- 6% likely had Lynch syndrome
- 56% did not have Lynch syndrome

The diagnosis of Lynch syndrome was known before surgery in just 4% of these cases.

The Digestive System



People with Lynch syndrome are known to be at increased risk for multiple cancers. Our study found that in the young people with Lynch syndrome, the risk of needing more surgery for another colorectal cancer was approximately 15% after 10 years and approximately 60% after 20 years from their initial surgery. The risk for a second colorectal cancer was more than 3.5 times higher in the young people with Lynch syndrome compared to the young people without Lynch syndrome. Furthermore, the young people who had a "standard" colon resection had nearly a 5-fold increased risk for developing another colorectal cancer compared to the young people who had an "extended" resection.

Our results, due to be published soon, demonstrate that young people with Lynch syndrome are much more likely to develop future colorectal cancers compared to those without Lynch syndrome; however, this risk can be significantly decreased by undergoing an 'extended' cancer resection at the time of the first cancer diagnosis.

Finding out whether someone has Lynch syndrome before they have surgery would be helpful in deciding what surgery would be best for a young person with colorectal cancer. A screening test can be done on a biopsy, prior to surgery, to see if there are features of Lynch syndrome in the cancer. While this does not confirm Lynch syndrome, it can help determine whether Lynch syndrome is highly likely, or not. We think this tumour testing should be performed for all young people with colorectal cancer before their surgery to help decide whether an extended or standard colorectal cancer resection should be performed.

Universal Screening for Lynch Syndrome in Women with Gynecologic Cancer

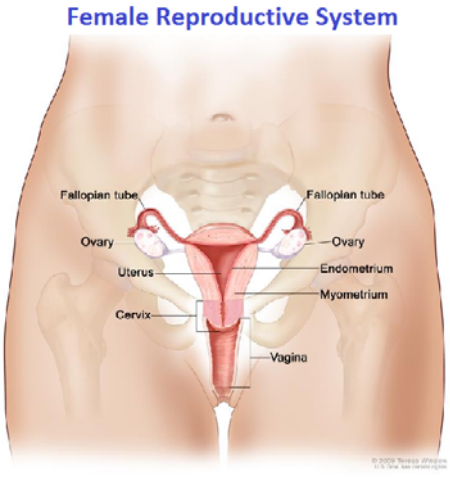
Sarah Ferguson, MD, Gynecologic Oncologist, Princess Margaret Cancer Centre

For Canadian women, endometrial cancer (also called uterine/womb cancer) is the most common, and ovarian cancer is the most aggressive, when considering cancers of the female reproductive system. We know that a small proportion of women with endometrial and certain types of ovarian cancer have Lynch Syndrome (LS), sometimes called Hereditary Non-Polyposis Colorectal Cancer (HNPCC). LS is an inherited condition that increases a person's lifetime risk for several types of cancer, most commonly colorectal and endometrial cancer. In order to prevent cancers, or catch them early when they are most treatable, there are targeted high-risk screening recommendations for people with LS. If LS is discovered to be the cause of a woman's endometrial or ovarian tumour, this information can be helpful for both the woman with cancer and her relatives. The woman herself could pursue screening of other organs, for example colonoscopy for colorectal cancer. For relatives, there is the potential to identify who in the family has also inherited the increased risk for cancer, and would therefore benefit from high-risk LS cancer screening, and who did not inherit LS and therefore can follow general population cancer screening.

One way of finding individuals who may have LS is through testing of tumour samples, after a biopsy or surgery. This screening test looks for specific features suggestive of LS. If an individual's tumour screens positive for features of LS, genetic testing on a blood sample may then be used to clarify whether or not that person has LS.

We recently completed a pilot study where we provided LS tumour screening to 118 women with endometrial cancer at Princess Margaret and Mount Sinai Hospitals in a research setting. Women with a family history of LS-related cancers or women whose tumours screened positive for features of LS were offered genetic counselling and genetic testing. Through this approach we found that 6% of the women had LS, including some who would have been missed using the traditional family history-based criteria alone. **This finding led to tumour testing being offered in a clinical setting for all women under age 70, newly diagnosed with endometrial cancer at hospitals within Mount Sinai Hospitalt and he University Health Network (UHN).** Importantly, only 55% of women who were eligible for genetic testing in our research study completed testing, suggesting that there is significant room for improvement.

We are now beginning an exciting new research study in which we are providing tumour screening for more than 1200 women under age 70 with endometrial and non-serous type ovarian cancer from four cancer hospitals across Canada to identify individuals with LS, representing the largest study of its kind. Recruitment will start in early 2015. We are aiming to find out what proportion of these women with endometrial and ovarian cancer have features of LS in their tumours, and how many of them are confirmed to have LS through genetic testing. For the women with features of LS seen in their tumour but whose genetic testing does not confirm LS, the study will investigate other possible reasons for the tumour test results using next -generation DNA sequencing. We are also aiming to improve the proportion of people eligible for genetic testing who pursue genetic counselling, including not only the women with cancer themselves, but their at-risk family members. By maximizing the identification of LS in patients and their families in this study, we will provide the opportunity to prevent many future cancers in patients and affected relatives. If adopted across Canada this could have a great impact, preventing hundreds of cancers every year.



Update on the Canadian Colorectal Cancer Consortium (C4)

Spring Holter, MS (C)CGC, Genetic Counsellor
Project Manager

The Canadian Colorectal Cancer Consortium (C4) is a Terry Fox Research Institute-funded multi-province project led by Dr. Steven Gallinger. The C4 aims to look at known genetic factors that cause colorectal cancer (CRC), to assess how family members get screened for CRC, and to look for new genes that may give some families a higher chance of developing CRC. The C4 is currently recruiting newly diagnosed patients with CRC under the age of 60. The C4 is screening for a type of hereditary colorectal cancer condition called Lynch Syndrome (LS). Screening for LS in CRC patients can be done relatively easily through a tumour test performed on a patient's CRC surgical or biopsy specimen. To date, 165 individuals have had LS tumour screening and of those 12 (7%) have been confirmed to have LS.

Three years of recruitment will take place at each centre and to date over 40% of estimated participants have been recruited at seven hospitals in five provinces:

- Mount Sinai Hospital, Toronto, ON – Dr. Robert Gryfe
- Sunnybrook Health Sciences Centre, Toronto, ON – Dr. Paul Karanicolas
- St. Michael's Hospital, Toronto, ON – Dr. Nancy Baxter
- Memorial University of Newfoundland, St. John's, NL – Dr. Patrick Parfrey
- McGill University Health Centre, Montreal, QC – Dr. William Foulkes
- Alberta Health Services, Edmonton, AB – Dr. Haili Wang
- St. Paul's Hospital, Vancouver, BC – Dr. Carl Brown

The study is also recruiting close family members (parents, siblings, and adult children) of the individuals with CRC. We will be determining if relatives go for CRC screening, through colonoscopy, and the factors that determine whether or not they participate in screening.

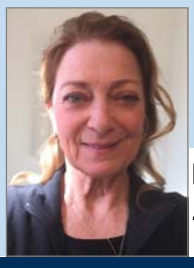
The C4 was reviewed by an external committee in February 2014 and it was decided that funding would continue until September 2017. We look forward to continuing this collaborative study that will help bring centres across Canada together in finding better ways to screen, treat, and prevent colorectal cancer. ■

NOW OPEN: Clinical Drug Trial for Polyposis

A new study has recently opened, focusing on reducing polyp development and how medication might change the quality of life for people with FAP. The Zane Cohen Centre is one of 14 centres participating, with patients taking part across the U.S., Canada, and Europe.

About FAP

Familial Adenomatous Polyposis (FAP) is an inherited disorder characterized by the development of numerous adenomatous polyps in the colon and rectum and typically begins in the teenage years. While these polyps start out benign (noncancerous), they may progress into colorectal cancer if left untreated.



You may be eligible if you have FAP and:

- Have not had your colon removed, but are considering this surgery, *OR*
- Have advanced disease in your upper small intestine (duodenum), *OR*
- Have had your colon removed and have advanced disease in your pouch/rectum

If you meet one of the above criteria, you may be eligible if:

- You have a mutation in the APC gene. If you have not been previously tested, your blood will be checked to see if you have the mutation required for participation in this study. Some, but not all people with FAP have this mutation.
- You are at least 18 years old

For more information please contact: Beverly Schmocker, RN, CCRP at bschmocker@mtsinai.on.ca or 416-586-4800 ext. 8286 # Or you may visit : http://canprevent.com/faptrial

Anticipation of Colonoscopy... Perspectives of Children and Teenagers

Carol Durno, MD, MSc, Pediatric Gastroenterology and Nutrition

Sometimes children and teenagers need to have a colonoscopy. People with conditions such as Familial Adenomatous Polyposis (FAP) and Lynch syndrome are at increased risk of colorectal cancer. A colonoscopy is a procedure that has been shown to lower the risk of colorectal cancer and save lives. It can be scary and anxiety provoking for some children and teenagers when they learn that a colonoscopy is needed. In order to make the experience as positive as possible, it is important to understand concerns faced by kids.

Children and teenagers recently filled out a questionnaire in our outpatient clinic before receiving teaching from our gastroenterology nurse about colonoscopy and what to expect. Below are questions from some of these children and teens.

1. I am afraid I will wake up during the procedure. Does this happen?

During a colonoscopy most young patients will be completely "asleep". Some doctors use medications which are given into an intravenous (IV) line. Once the medicine is given most patients fall asleep in less than 10 seconds. Sometimes a mask is used to give a gas that puts patients to sleep. During the colonoscopy you will be monitored very closely. Your heart rate and rate of breathing help to determine how much medication you need. By monitoring you closely the anesthetist will make sure you don't wake up until after the colonoscopy is over.

2. Will I have to get a needle?

This is a good question to ask the anesthetist when you meet before the procedure (the person who will be giving the medicine). Some patients prefer to have gas sedation and then have the IV inserted once they are asleep. Sometimes no needle is needed and the gas may be the only sedation necessary.

3. Will it hurt?

Most children and teenagers don't find colonoscopy painful. Young people are given medicine to make them sleep during the procedure. Air is pumped into the cleaned out colon to keep it open so that the doctors can get the best pictures. The pressure from the air can sometimes cause some discomfort and cramping after the colonoscopy. The majority of patients don't experience any cramping.

4. Who will do the procedure?

Colonoscopy is almost always done by a doctor, usually a gastroenterologist (a doctor who speciality is the digestive tract) or a surgeon.

5. Will I have privacy?

Colonoscopy is done in a private area. The majority of colonoscopy procedures are done in a procedure room or in the operating room. It is a good idea to ask your doctor about where the procedure will be done.

6. How do I prepare for the test?

It is important for the bowel to be clean before a colonoscopy so the doctor can get the best pictures. You will take some strong laxatives to help clean out the bowel. You will receive instructions from the doctor's office ahead of time. If you don't understand the instructions it is important to call the doctor's office and go over them step by step. Preparing for a colonoscopy makes you go the bathroom a lot. You want to ensure you are close to a bathroom once you take the laxatives.

Tips for Parents to Help Prepare Young Kids for Colonoscopy

- Read appropriate children's books and play "hospital" at home
- Bring a favourite toy or stuffed animal to the procedure
- Make baby-sitting arrangements for other children so you can focus on the child undergoing colonoscopy
- In simple terms explain to your child what to expect

We would like to hear from you about your experiences with colonoscopy. Do you have suggestions on how to make the procedure more positive? Let us know at fgicr@mtsinai.on.ca

Polyposis Registry Update

Kara Semotiuk, MS, (C)CGC

As many of you already know, our former Polyposis registry coordinator, Terri Berk, has retired. Her families with polyposis are now being assigned to one of the four genetic counsellors at the FGICR; Spring, Melyssa, Kara, and Laura (pictured to the right). If you would like to know which counsellor is the new point of contact for your family, feel free to contact us.

We would like to highlight some aspects of how the registry works, and what we can offer to you and your families.

Stay On Top Of Your Screening

It is important to keep track of your screening appointments. When it is time for your next screening, we will be happy to refer you to the appropriate doctors for your appointments. The appointments will be booked by the doctors' offices directly. Appointments will not be booked automatically; please contact us before your next screening appointment is due and we will gladly refer you wherever needed.

Is Your Genetic Testing Up To Date?

Genetics is constantly evolving. Some of you may be eligible for further genetic testing that was not available at the time you originally met with us. If you already have a known gene mutation in your family this would likely not apply to you, however; if you had genetic testing in the past that did not find any gene mutations or if you have multiple polyps and haven't had any genetic testing, you might be eligible for updated genetic testing. Genetic testing can help determine your appropriate screening recommendations, and can also provide useful information for your relatives. Please contact the registry at the main phone number, 1-877-586-5112 to determine if genetic testing for your family is up to date.

Share your story

We have heard some inspiring stories from families who are living with a predisposition to cancer. We are looking for individuals to help others in a way that we can't – by sharing your stories and experiences to help people who may be going through a similar situation.

HOW CAN I HELP?

⇒ **Be a buddy** to someone going through a similar experience. This may involve talking to someone over the phone, by email, or meeting them in person.

⇒ **Share your story** with us to be included on our website.

If you would like to be a buddy and/or share your story, please let us know by contacting your genetic counsellor or the FGICR at 1-877-586-5112, or fgicr@mtsinai.on.ca

Contact us

6th Biennial Lynch Syndrome Education Night

Lynch syndrome (also known as Hereditary Non-Polyposis Colorectal Cancer or HNPCC) is a rare genetic condition that greatly increases the risk to develop cancer, primarily colorectal and endometrial cancer. At one time, it was thought that all colorectal cancer followed a similar pathway to becoming cancerous, but we now know that this is not the case. While most lectures on colorectal cancer refer to the general population, talks for families with Lynch syndrome should be tailored to the research on risk, screening, and treatment of cancers in these families.

It was for that reason that the biennial education night for patients with Lynch syndrome was created. We began hosting this night in 2003 and have invited surgeons, gastroenterologists, gynecologists, pathologists, a family physician, dermatologist and psychotherapist to update our families on the latest research and understanding of this syndrome. We also invited patients with Lynch syndrome to share their stories, which were always greatly appreciated by the audience.

We will be hosting the 6th Biennial Lynch Syndrome Education Night on **May 27, 2015**. If your family has Lynch syndrome, you should receive a flyer in the mail with details on the time, location and speakers for this night. If you do not receive a flyer and are interested in attending, please feel free to contact us at 416-586-4800 ext. 5112 and we can add you to the mailing list designed for this evening.

NEW in 2015: A Meeting for Families with Familial Polyposis

Familial Adenomatous Polyposis (FAP) is a rare genetic condition where the main feature is that individuals develop multiple polyps (mainly adenomas) in the colon and rectum. People may develop hundreds or thousands of these polyps, starting from as young as childhood, and there is a very high risk for the polyps to turn into colorectal cancer. There is also a milder form of FAP called Attenuated FAP, where polyps may grow at later ages and fewer polyps typically develop. In addition to FAP and Attenuated FAP, there is another condition called *MUTYH*-Associated Polyposis (MAP), and it can be difficult to difficult to tell the difference between MAP and (Attenuated) FAP without genetic testing. A main difference between these conditions is in the way they are passed down in families, and therefore risks to relatives will vary.

Team Members at the Zane Cohen Centre for Digestive Diseases

We are planning to host an education night for families with FAP, Attenuated FAP and MAP to address issues specific to these conditions. The meeting is planned for a weeknight in the Fall of 2015 and will be held in Toronto. More information about the meeting will be sent out later this year. To make sure that you are on the mailing list for this meeting, you can contact us at 416-586-4800 ext. 5112.

MEET AND GREET

As a multidisciplinary Registry, we offer many different services to patients and their families. We would like to introduce you to team members who have recently joined us.

Medical Geneticist Joins the FGICR

A message from Dr. Raymond Kim:

I am delighted to join the Familial Gastrointestinal Cancer Registry as the new medical geneticist. In this role, I see patients with the genetic counsellors and complement the care delivered in the genetics clinic. In particular, patients with a hereditary form of gastrointestinal cancer often have other medical manifestations which need a physician's assessment. Also, many patients with hereditary cancers have children affected with the same disorder, and as a medical geneticist I am experienced in both pediatric and adult care facilitating this transition and focusing on family-centred care. I have had a wonderful experience in my short few months since joining the team and hope to meet many more families and continue working with such a rich team.

Dr. Raymond Kim is currently a medical geneticist at the FGICR, Mount Sinai Hospital and University Health Network. He sees adults and children in a variety of cancer clinics, with a particular emphasis on transition of care and multi-disciplinary models. He is an assistant professor in the Department of Medicine at the University of Toronto. He received his MD/PhD from the University of Toronto, and completed his PhD with Dr Tak W Mak at the Princess Margaret Cancer Centre using animal models of tumourigenesis. Through a screen examining genes which interact with the tumour suppressor PTEN, he identified a novel oncogene, DJ-1 which was later characterized as a familial Parkinson's disease locus. He then completed a residency in Internal Medicine, followed by a fellowship in Medical Genetics at the Hospital for Sick Children with Dr Stephen Scherer with a focus in next generation sequencing in rare diseases. His research interests lie in hereditary cancer surveillance, the use of novel molecular genetic analyses in patient care and their application in research studies. This includes whole exome sequencing, and whole genome sequencing in high-risk pedigrees and tumours.

Mount Sinai Hospital's Department of Pathobiology Welcomes Dr. Charames

Dr. George Charames is the Director of the Advanced Molecular Diagnostics Laboratory in Mount Sinai Hospital's Department of Pathology and Lab Medicine, and Assistant Professor in Lab Medicine and Pathobiology at the University of Toronto. Dr. Charames earned his MSc and PhD from the University of Toronto, and went on to complete a Clinical Molecular Genetics fellowship at the Johns Hopkins School of Medicine, before returning to Toronto. Dr. Charames has implemented Next Generation Sequencing technology in the molecular diagnostic lab for the detection of genes associated with Hereditary Breast and Ovarian Cancers and is currently developing this superior technology for inherited colorectal cancers such as Lynch and polyposis syndromes. Dr. Charames' research interests are in the development of genomic and proteomic strategies for improved clinical diagnostics of cancers, including gastrointestinal cancer. His translational research approach aims to use high throughput genomic sequencing, in addition to proteomics and biomarker discovery technologies, to improve individualized medicine.

FGICR Welcomes a New Genetic Counsellor

Laura Winter is a certified genetic counsellor who joined the FGICR team at Mount Sinai Hospital in February 2014. She joins us after working as a Family History Assessor and then as the Auckland Regional Coordinator with the New Zealand Familial Gastrointestinal Cancer Service. Before obtaining her Master of Science degree in Genetic Counselling from the University of Toronto, Laura was a volunteer here at the FGICR. In addition to being involved with the research registry, Laura's main role is as a clinical genetic counsellor, where she and the other counsellors assess family histories of gastrointestinal and related cancers, as well as familial polyposis.

Visiting Researcher at the FGICR

Dr. Kate McNamara is a visiting research fellow from Brooklyn, New York. Dr. McNamara is a surgical resident at the State University of New York Downstate Medical Center in Brooklyn and is taking two years away from clinical training to complete a research fellowship. She has a particular interest in colorectal surgery and is the first recipient of the Colon Cancer Challenge Foundation Research Scholar Award designed to support a surgical resident doing research focusing on hereditary and early-onset colorectal cancer at a specialized centre. With this award, she has been able to travel to Toronto to work under the mentorship of Dr. Zane Cohen and Dr. Steven Gallinger doing exciting research on the genetics of early-onset colorectal cancer using the latest DNA sequencing technologies.

News Flash

Kara Semotiuk, MS, (C)CGC
Genetic Counsellor

Once again, several of our team members attended the 18th Annual Meeting of the Collaborative Group of the Americas on Inherited Colorectal Cancer, held in New Orleans in September 2014. This educational conference is an excellent forum for groups who study hereditary gastrointestinal cancer and polyps to come together to present their research and to learn from each other. Because hereditary cancer syndromes are rare, advances in research depend on collaboration between groups internationally.

Genetic counsellors, physicians and researchers from The Zane Cohen Centre for Digestive Diseases attended the meeting in New Orleans. Several people from our group presented posters, gave talks and ran workshops on a variety of topics, including Biallelic Mismatch Repair Deficiency, gene panels, the Canadian Colorectal Cancer Consortium and genetic counselling issues.

Some interesting topics presented by other groups included information about the upcoming CAPP3 trial looking at the optimal effective dosage of aspirin for reducing polyp growth in people with Lynch syndrome, a review the relatively new GREM1 gene associated with Hereditary Mixed Polyposis Syndrome (HMPS), and updates on universal tumour testing for Lynch syndrome (screening all new colorectal cancers for Lynch syndrome features at individual institutions).

Attending this annual meeting helps to keep our group abreast of the latest research, and also provides the opportunity to present our own research on hereditary gastrointestinal cancer and polyposis. We look forward to what the 2015 meeting in Baltimore has in store.

We Build Knowledge
To Improve Treatment

Our growing successes have been made possible because of the support of patient donors

We invite you to partner with us

To donate on line
www.zanecohencentre.ca/donate

To discuss other options for support, please contact

Patricia Tolkin Eppel PhD
I Advancement Director I

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Glossary

Adenoma: A precancerous polyp.

Anesthetist: Medical specialist who gives medication that lowers sensitivity to pain, often before/during a medical procedure.

Colorectal: Large bowel/intestine (colon) and rectum.

Colorectal Resection: Surgery to remove all or part of the colon/rectum.

Duodenum: First part of the small bowel/intestine.

Endometrium: Lining of the uterus/womb.

Exome: The coding portions of all of a person's genes. About 1-2% of all of their genetic material.

Familial Adenomatous Polyposis: (FAP) Genetic condition primarily affecting the gastrointestinal tract characterized by 100 or more precancerous polyps (adenomas) of the colon. There is also a milder/later onset form called Attenuated FAP (AFAP).

Gynecologic: Relating to the female reproductive system.

Lynch Syndrome: Genetic condition in which people are at increased risk of cancer of the large bowel, endometrium, and other associated cancers. Sometimes called HNPCC.

***MUTYH*-Associated Polyposis (MAP):** Genetic condition of the gastrointestinal tract characterized by 10 or more precancerous polyps in the colon. Inherited in different pattern than (A)FAP.

Mutation: A problem/change in the code of a gene which causes the gene to not work properly.

Next-Generation Sequencing: New high-powered technology for reading through the sequence of large amounts of DNA, which is faster and less expensive than previous methods.

Pilot Study: A smaller version of a larger study that is carried out first to decide whether, and how, to launch a full-scale study.

Polyposis: A condition characterized by numerous internal polyps, often in the colon and rectum.

In an effort to be more **environmentally friendly**, this newsletter has been sent electronically to those who have provided their email addresses. To receive future newsletters and updates electronically, please email us at **fgicr@mtsinai.on.ca**