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NETWORK

Familial Gastrointestinal Cancer Registry

SUMMER 2008

CLINICAL FOCUS

Kara Semotiuk, MSc, (C)CGC

Ontario's Colorectal Cancer Screening Program

Colorectal cancer is the fourth most commonly diagnosed type of cancer in Ontario, and it is the second overall highest cause of cancer death in the province. However, many cases of colorectal cancer are preventable. In the event colorectal cancer does occur, if diagnosed early, the majority of people would survive.

On March 14, the Ministry of Health and Long-Term Care announced a campaign to encourage more Ontarians to get screened for colorectal cancer. You might have seen television commercials advertising this initiative, starring the "see-through" people having a conversation about screening for colorectal cancer. The goals of this campaign are to raise public awareness of colorectal cancer, screening and the benefits of early intervention.

Who is eligible?

This program is aimed at people aged 50 and older with no symptoms and no family history of colorectal cancer. However, by boosting public awareness of risk factors and screening options for colorectal cancer, this program should also help identify people who are at increased risk.

What tests are being offered?

Fecal occult blood test (FOBT)

FOBT is offered to men and women 50 years of age and older at average risk for colorectal cancer (no family history of colorectal cancer and no symptoms). The Ontario Colorectal Cancer Screening program recommends FOBT be done every two years.

FOBT is a noninvasive, self-administered test that requires collecting stool samples at home on three different days. FOBT kits will be readily available through primary health-care providers, at pharmacies, and through Telehealth Ontario at 1-866-828-9213. FOBT checks for invisible traces of blood in the stool that could be signs of colorectal cancer. Approximately 2 per cent of average-risk people who complete FOBT will have a positive result, but this doesn't necessarily mean someone has cancer. People whose FOBT is positive are referred for further investigation with colonoscopy.

Colonoscopy

Colonoscopy is offered to people at increased risk for colorectal cancer (see below). Colonoscopy is an internal exam looking at the lining of the colon (large bowel) and rectum using a long thin tube with a camera on the end. It is performed by a specialist, usually in a hospital or gastrointestinal clinic. There is a small risk to tear the colon with colonoscopy since it is an invasive test; however, it is the most effective screening tool for colorectal cancer. It is not only a screening tool used for early detection, but also a cancer prevention tool in that polyps (small, mushroom-like growths on the lining of the colon) can be removed during the procedure itself.

Who is at increased risk?

The following are some of the known risk factors for colorectal cancer:

- People with a family history of colorectal cancer or other cancers related to hereditary colorectal cancer syndromes, for example Hereditary Non-Polyposis Colorectal Cancer (HNPCC)/Lynch Syndrome, Familial Adenomatous Polyposis (FAP), MYH-Associated Polyposis (MAP) and other hereditary cancer and polyp syndromes

- People with symptoms of colorectal cancer. There are many possible explanations for these symptoms - experiencing them do not necessarily mean that someone has colorectal cancer:
    - Change in bowel movements/habits
    - Blood in stool
    - Abdominal discomfort
    - Unexplained weight loss
    - Fatigue
    - Vomiting
- People with inflammatory bowel disease (Crohn's disease/ ulcerative colitis)
- People with multiple polyps or polyps that are left unchecked

It is important to distinguish between average-risk and those at increased risk for colorectal cancer as the screening recommendations are quite different. Please speak to a health professional about what screening tests are appropriate for you. For more information on the Ontario Colorectal Cancer Screening Program visit **www.coloncancercheck.ca**.

RESEARCH UPDATE

Spring Holter, MSc

A New Hereditary Paediatric Cancer Syndrome

Hereditary Non-Polyposis Colorectal Cancer (HNPCC), also known as Lynch syndrome, is caused by mutations (genetic changes) in one of four different genes: MLH1, MSH2, MSH6 and PMS2. Having a mutation in one copy of any of these genes is sufficient to cause HNPCC. It is well known that people with HNPCC have an increased chance of developing many different cancers, including colorectal cancer. The average age for someone with HNPCC to develop colorectal cancer is 44. In 2005, a study was published by our paediatric gastroenterologist, Dr. Carol Durno, which showed that HNPCC is a rare cause of paediatric colorectal cancer (diagnosed under age 18).

Over the past decade, the genetic cause of a new paediatric cancer syndrome has been identified. This new condition is caused by having a mutation in both copies in any of the HNPCC genes.

When people have mutations in both copies of their genes it is often referred to as having "**biallelic**" mutations. Those who have biallelic HNPCC mutations are at an increased risk of developing brain tumours, leukemia, lymphoma, small bowel cancer, and colorectal cancer. Unfortunately, this new condition does not have any agreed upon name, although it has been referred to as Turcot syndrome, Lynch syndrome III, and MMR-D syndrome.

Having biallelic HNPCC mutations is rare. Current medical literature reports 69 people from 36 families with biallelic HNPCC mutations. These 36 families have mutations representing all four HNPCC genes: 17 families have biallelic PMS2 mutations, eight have biallelic MSH6, and seven have biallelic MLH1 and four have biallelic MSH2.

These 69 people have a total of 68 colorectal cancers, 37 brain tumours, 15 lymphomas, 10 leukemias and four small bowel cancers. People in this category may commonly be diagnosed with a brain tumour, leukemia, or lymphoma as their first cancer and then go on to develop a cancer of the gastrointestinal tract. However, 18 per cent will be diagnosed with colorectal cancer as their first cancer. The average age for colorectal cancer diagnosis is 16, but ranges from 5 to 24 years old. One of the most important and often unrecognized features of this condition is the growth of numerous colorectal polyps. Some people have been found to have ~50 polyps. This is important to be able to give an accurate diagnosis to a family because FAP and MAP can also lead to the development of multiple polyps.

Identifying these people before they develop cancer is difficult. The only physical features that seem to be associated with this condition are dark spots on the skin called café-au-lait (CAL) spots. CAL spots are a common feature of another genetic syndrome known as Neurofibromatosis type 1 (NF1). Children with biallelic HNPCC mutations are commonly evaluated for NF1because of their café-au-lait spots; however, most of them do not meet the medical definition of NF1. Currently, there is a collaborative study underway through Mount Sinai Hospital's Familial Gastrointestinal Cancer Registry and the NF1 Clinic at Sick Kids Hospital in Toronto. The goal is to identify children with biallelic HNPCC mutations who may be evaluated due to their CAL and before a diagnosis of cancer.

A unique point about this condition is that since each individual has mutations in both copies of their HNPCC genes, each of their parents likely has a mutation in one of their HNPCC genes and, therefore, has the typical form of HNPCC. Parents of these children need to have the appropriate surveillance recommended for people with HNPCC, such as a colonoscopy, every one to two years.

As well, they need to be aware of the other cancer risks associated with HNPCC (**Please see Network, Summer 2007**).

As more patients with biallelic HNPCC mutations are identified, it will help us provide better information to families. Learning about this condition will help determine specific cancer risks, the best monitoring and prevention strategies, and potential treatment options.

EDITOR'S MAILBAG

Andrea Clark, BASc, RD Clinical Dietitian

Nutrition plays a vital role for patients with FAP, HNPCC and other polyposis syndromes who have had their colon removed and an ileostomy or pelvic pouch created in its place. The goal of nutrition is to:

1. Decrease the risk of dehydration and **electrolyte** (sodium and potassium) loss.
2. Decrease the amount of stool produced.
3. Promote stool thickness (to help with #1).
4. Decrease the risk of obstruction due to undigested foodstuffs.

The following are questions and answers that are normally discussed with patients before and/or after their surgery. These are designed to give you an idea of the nutrition guidelines with an ileostomy and after the pelvic pouch procedure.

Q Do I have to follow a special diet before surgery?

A No, patients with FAP, HNPCC and other polyposis syndromes do not have to follow a special diet before surgery. There are two diet-related issues you may want to consider a day or two before your surgery:

1. Choose bland foods without a lot of added salt or spice.
2. Drink lots of liquids to stay hydrated, including water, juice or other decaffeinated and alcohol-free beverages.

Following these tips can make the day of surgery more manageable, because it can minimize your thirst the morning of surgery (generally, you cannot eat or drink anything from 12 midnight until your surgery the next day).

Q If I have an ileostomy, are there certain foods I should avoid? For how long?

A During the first six weeks after an ileostomy has been created or re-sited, we recommend that patients avoid nuts, seeds, skins, high-fiber foods and stringy foods, as they are not broken down by the body and can put patients at risk of an obstruction while their stoma is still swollen. Other foods to avoid include popcorn, corn, mushrooms and coconut. You will want to chew all of your foods well and eat slowly — chewing is your first line of defence in preventing an obstruction because that is when the majority of your food is broken down!

Stool consistency is important with an ileostomy. There are certain foods that are more likely to cause watery stools and high ostomy outputs (more than one litre of stool per day), which can lead to dehydration and electrolyte loss. Products that are high in sugar — fruit juices, soft drinks — as well as spicy foods, fried foods, caffeine (in coffee, tea, chocolate, energy drinks) and alcohol can all affect stool consistency.

It is important to limit or avoid these foods if you are having consistent diarrhea. Foods that can help thicken stool include: bananas, applesauce, peanut butter, cheese, oatmeal, oat bran, white bread, potatoes, potato chips, matzo crackers, marshmallow, and Jell-O. Try to include some of these foods on a regular basis to keep your stool thick. If food alone is not helping to thicken your stool, you can try oat bran fiber or Metamucil. For a small number of patients, the most effective way to thicken stool is medication, which can be prescribed by your surgeon or family doctor.

Q How much should I be drinking?

A The role of the colon in nutrition is to absorb fluids and electrolytes to maintain hydration. When the colon is removed, the remainder of your bowel will slowly adapt to take on the role of the colon, but it will never be as efficient. This means that drinking enough fluids after surgery is very important. You will want to drink at least two litres of fluid daily, (more if your ostomy outputs are greater than one litre per day) and at least one litre of this should be water.

To ensure adequate sodium intake, add salt freely to your foods and include foods or drinks higher in sodium, such as vegetable juices, soups, salted crackers and processed foods. Potassium is found in orange juice, tomato products and potatoes, to name a few.

Q If I become dehydrated, will a sports drink like Gatorade or Powerade help?

A Most people believe that if they are dehydrated, drinking a sports drink, like Gatorade, will help with hydration. This is not true for patients with an ileostomy or pelvic pouch, as the sugar concentration in sports drinks is too high and drinking them can actually increase stoma outputs and worsen dehydration. The best fluids to maintain hydration or replenish your fluids are: water; broth; an oral rehydration solution (e.g. Gastrolyte, pedialyte); a diluted sports drink (two cups water, two cups Gatorade and ¼ tsp. of salt); tomato or vegetable juice.

Q Is there a special diet to follow after the second operation for the pelvic pouch procedure when my ileostomy is closed?

A Again, we recommend following a low-residue diet for the first six weeks after surgery. This allows healing of the pouch and the bowel reattachments. It also reduces the amount of stool and gas and helps with stool thickness, which may reduce the number of trips to the bathroom. Including foods to promote thick stools is even more important with a pelvic pouch than with an ileostomy, since the thicker the stool, the less often you will visit the bathroom! If you are eating many of the foods known to thicken stools and you are not seeing the results that you want or need, you can try oat bran or Metamucil before going to your doctor for further suggestions. Make sure you are not having a lot of foods or drinks high in sugars, caffeine or alcohol; minimize or avoid greasy and spicy foods. Hydration is also still important, which means drinking two litres of fluid per day, with one litre of this being water.

Q What happens after the six weeks? Can I go back to a regular diet?

A For patients with an ileostomy or a pelvic pouch, you can start reintroducing the foods that you have been avoiding, one at a time, after six weeks. We suggest leaving 24 to 48 hours between each new food you introduce to make sure you can tolerate them. Signs or symptoms that you may not tolerate the food are: pain or abdominal cramping; no output from your stoma for an extended period; a large amount of watery stool being put out from the stoma. These symptoms are rare, but if this happens to you, follow up with your surgeon or go to your nearest Emergency Department. These symptoms could be signs of an obstruction.

To decrease their risk of an obstruction, patients with an ileostomy may want to permanently avoid the following foods: nuts, popcorn, corn and coconut. Otherwise, most patients are able to return to a normal diet. Just remember that there are several foods that will always have an effect on your stool consistency.

Q Am I at risk for any vitamin or mineral deficiencies?

A No. The biggest nutritional risks after surgery are dehydration and loss of electrolytes, which can be dealt with easily by ensuring proper fluid intake, and consuming food or drink with sodium and potassium. The remainder of the vitamins and minerals are digested and absorbed in the stomach and small intestine, which is fully intact.

The low-residue diet does restrict whole grains and many fruits and vegetables, so some patients worry about getting enough vitamins and minerals from their diet. Remember, the diet is only for short -term use, when you are at low risk for deficiencies. It is safe and acceptable to take a daily multi-vitamin.

Q Can I drink alcohol with an ileostomy or pelvic pouch?

A If you are on any kind of medication, always check with your doctor or pharmacist to ensure it is safe for you to drink alcohol.

Otherwise, it is safe but there are some special considerations. Any type of alcohol can cause more watery outputs or diarrhea and excess fluid losses through increased urination. This puts you at a higher risk for dehydration. If beer is your drink of choice, be aware that you will likely experience more gas as well.

Remember moderation when including alcohol, and make sure to drink some water in-between drinks and/or after to minimize the risk of dehydration.

Q I am having a lot of gas. What can I do about this?

A To decrease the amount of air you swallow (and therefore the amount of gas put into your body), try to minimize carbonated drinks (including beer); chewing gum; drinking through a straw or talking while you are eating. You can also minimize or avoid gas-producing foods, such as cabbage, Brussels sprouts, broccoli, cauliflower, beans and legumes, onions, spicy foods, melons, milk and milk products such as cheese.

All patients will be seen by a Registered Dietitian after their surgery for the appropriate diet education. At that time, patients are given detailed handouts outlining the foods to choose and avoid, and troubleshooting tips to work through the common challenges faced by patients in the days and weeks after surgery. Contact information is also provided for any questions you may have after discharge from hospital.

MYTH OR REALITY?

Ten years ago, I was diagnosed with FAP at age 14 and was told that I would be cured once my large bowel was removed. I had bowel surgery so I don't need to have any more check-ups.

Myth

The word "cure" can be confusing. Removing the colon will treat the immediate problem of adenomas (precancerous polyps) to prevent the development of cancer.

Adenomas can occur wherever there is lining in the gastrointestinal tract. If you had your large bowel removed and the small bowel attached to your rectum, the remaining bowel must be scoped. If you had a pelvic or Kock pouch created out of your small intestine, here too, adenomas can develop in the lining. If you had your rectum removed and have an ileostomy, polyps may occur either on the opening or within the ileum (the last part of the small intestine). The good news is that, since the areas to be checked are smaller, the scopes are shorter and there is less bowel preparation required. You are also able to have these tests done with sedation so that you are comfortable. Each time you have a scope, your specialist can do a biopsy if any polyps are found. This will guide the specialist in determining when you should come back.

People with FAP cannot get polyps outside the large bowel.

Myth

People with FAP produce more bile, especially after the colon is removed. Bile appears to promote the growth of adenomas in the duodenum (the first part of the small intestine. Generally, these adenomas tend to be smaller in number and grow more slowly than in the colon. There is a blind spot where the stomach opens to the duodenum that can be very difficult to see unless a particular scope is used, called a side-viewing endoscope. Much like the mirror on the passenger side of a car, the side-viewing endoscope allows the specialist to see the blind spot clearly and this is important because adenomas often grow here. There are many different ways to treat these polyps through the scope. There are no symptoms for early adenomas, which is why regular scopes are suggested from age 25. How often they are repeated depends on whether there are any polyps, how big they are, and what the biopsy shows when polyps are checked under the microscope. (**Please see Network, Spring 2006**).

My brother and I were diagnosed with multiple adenomas in our late 40s. Neither of our parents, now in their 80s, is affected. APC testing did not identify a genetic change. My brother says there isn't another genetic test but I told him he is wrong.

Reality

There is a new gene called MYH, which is responsible for an inherited form of polyposis called MAP, or MYH-associated polyposis. MAP appears to be passed on in a different way than FAP.

We all have two copies of every gene, one from each parent. In MAP, both copies of the disease gene must be mutated, or changed, for the disease to appear. In other words, if your mother has a mutated copy of the gene and so does your father, you and your brother had a 1 in 4 (25 per cent) chance of inheriting both MYH mutations and developing MAP. In Ontario, the Ministry of Health and Long-Term Care has now approved genetic testing for MYH, which is offered through our Registry's molecular laboratory. Genetic counseling is available across the country through provincial genetic clinics and health units. (**Please see Network, Spring 2004**).

My mother had Peutz-Jeghers Syndrome but she died of breast cancer. I also have Peutz-Jeghers and I think her breast cancer had something to do with this disease.

Reality

There is a higher risk for breast cancer in women with Peutz-Jeghers syndrome (PJS) than in the general population. For this reason, monitoring is recommended from age 25. Women with PJS are also at risk for benign (non-cancerous) tumours and cancers of the cervix and ovaries. It is strongly suggested that you have an annual examination with your gynecologist. Monitoring guidelines are available from the Registry.

My son was diagnosed with juvenile polyposis at age 19 and recently had genetic testing. He told me there is a connection between the type of gene found and his juvenile polyps.

Reality

There are three different genes associated with juvenile polyposis (JPS): SMAD4, BMPR1A, and ENG. Those who have a SMAD4 mutation tend to have large numbers of stomach polyps and tend to have a family history of stomach polyps. The stomach and small intestine should be scoped from the age of 15. This examination should be repeated every three years if no polyps are found, or more frequently if indicated. This scope is recommended in addition to colonoscopy.

SMAD4 mutations are also associated with vascular malformations called hereditary hemorrhagic telangectasia, or HHT. These malformations may occur in the brain, lungs, gastrointestinal tract, liver, or spine because minute blood vessels between the arteries and veins are absent.

Sometimes, people may be diagnosed with HHT and may be unaware of the inherited association with juvenile polyposis.

REGISTRY NEWS

Melyssa Aronson, MSc (C)CGC

Education Night for Hereditary Colorectal Cancer Families

On May 14, the Familial Gastrointestinal Cancer Registry hosted the third Biennial Education Night for patients with Hereditary Non-Polyposis Colorectal Cancer Syndrome (HNPCC), or Lynch syndrome. Patients are referred to the Registry from all over Ontario because of their personal and family histories of cancer. The Registry is the largest colorectal cancer family study centre in Canada and known throughout the international community.

We began hosting an Education Night in 2003 for patients with this rare condition for updates on the changing genetic information and education on cancer monitoring as well as offering a forum where patients can share their stories with each other.

This year, three experts from the Registry, Drs. Robert Gryfe, Aaron Pollett and Mary Jane Esplen, spoke to a group of more than 65 Lynch syndrome families and friends. Discussions included updates on HNPCC/Lynch syndrome; surgical options; the pathologist's role in diagnosis; and the psychosocial aspects of having this inherited condition. Two wonderful patient speakers, Monique and Steve, shared their stories about being diagnosed with cancer and Lynch syndrome. Dr. Zane Cohen, co-director of the Registry, moderated this very successful evening.

"It was very informative…it is good to know we have very qualified doctors and staff at Mount Sinai. Thank you for organizing such an event," said Barbara M., a patient and attendee.

NETWORK
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