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NETWORK

Familial Gastrointestinal Cancer Registry

SUMMER 2007

RESEARCH UPDATE

Interviews and Focus Groups with Teenagers with FAP

Robert Gryfe MD, Department of Surgery

A few years ago, Terri Berk, Co-ordinator of the Familial GI Cancer Registry helped us organize a number of interviews and focus groups with teenagers from age 12 to 18. These teens spoke to the psychosocial research team headed by Dr. Mary Jane Esplen about what it is like to have Familial Adenomatous Polyposis (FAP).

We learned a lot about how the challenges of living with a rare, life long condition like FAP at a young age can build an inner strength and create a new perspective on friends, family as well as life in general.We would like to share some of the positive and some of the more challenging aspects of FAP for teens in this issue. The comments and topics made by the teens fit into five main categories, including:

Body Image and Bowel Functioning

There were a number of comments made about changes in body and self-image and concerning feelings of attractiveness (not necessarily in a negative way, just different). Some teens expressed feeling self-conscious about the surgery scar(s) and others feared the loss of control of bowel function or sounds that might arise in public.

Relationships with Others

There were many comments about relationships with others, including relations with family and friends. Some worried about being accepted. Some talked about how their relationships were strengthened because of knowing who your friends really are. Other themes related to being able help or protect younger family members and to provide mentorship and role modelling.

Other Strong Feelings

Some teens talked about feeling a little more vulnerable, but also feeling stronger. At times teens felt the need to put on a front if they weren't feeling well. Some felt angry about their situation and others talked about concerns of passing on the condition to their children.

Misunderstanding

A lack of privacy and feeling over-protected by one's family was a major concern for some teens. Feeling misunderstood because of the ignorance or lack of knowledge about FAP by friends and schoolmates was common. For example, some teachers misinterpreted a student's diagnosis of FAP as cancer.

Coping

Some teenagers expressed issues concerning self-esteem and being able to cope with the changes caused by FAP. One coping mechanism for some teens included being able to joke and make fun of potentially embarrassing situations. Some teens were worried about intimacy and sexuality and expressed a need to come up with a way to explain their condition to future boyfriends and girlfriends.

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CLINICAL FOCUS

Kids...What to Expect with Early-Onset Familial Adenomatous Polyposis (FAP)

Carol A. Durno MD, MSc,
Registry Pediatric Gastroenterologist

People with classic FAP often have some polyps by age 10 to 12. It is important to have a first sigmoidoscopy between age 10 and 12 in order to determine if polyps are present and, if so, how many. The recommendations (for Canada and the United States) are to have annual colonoscopies after this first sigmoidoscopy. This allows children and adolescents to take the best care of their bodies by having the doctor keep an eye on the number and size of the polyps. There is a small group of patients with FAP who develop many polyps at an earlier age and have a more severe form of FAP. These patients require earlier intervention usually including surgery (colectomy) at a much younger age than most people with FAP.

Most of the studies and articles in pediatric FAP focus on teens with FAP. Little has been written on younger children with FAP. In particular, no articles have focussed on the very young children with FAP and how they manage. A colonoscopy should be performed every year. Some children and teens with FAP do not have an annual colonoscopy. We are interested to find out why some children and teens do not have the annual scope. Are there things that health-care workers can do to ensure that all children have the annual colonoscopy? As part of this project, we will look at barriers to surveillance in kids and teens. We are interested in looking at the outcome of children with FAP who had a colectomy at age 12 or younger. We want to find out how a patient's bowel function is after surgery (and later in teenage and adult life). Do patients who have a colectomy as young children have the same results as teens or adults who have a colectomy? What is their quality of life? The second issue we want to study is two-fold: how children cope with the operation; and how the whole family has coped with having a very young child who needs early intervention.

We depend on patients and their families for feedback on what issues are relevant. Please let us know if you have specific suggestions or thoughts to include in this study that is currently being designed. It would be very helpful to hear from children, teens, and/or young adults who could tell us "first-hand" about their experiences with FAP. For instance, families report frustration in having to travel large distances for annual consultations and then have to return at another time for the colonoscopy. This can be costly, requiring additional time off school and away from work. Many children and teens find two visits emotionally upsetting and prefer to have a single annual appointment, including the procedure. Many families prefer to have all of the children scoped on the same day, as some parents and kids find a single visit less disruptive to the family. Your input helps us to look after patients and their families in a manner that is most sensitive to people needs.

There are many quality of life issues for patients with FAP and for their families. In a questionnaire sent to our adult Registry patients, the following problems were highlighted: concern about the lack of knowledge among health professionals; changes in awareness of appearance; feelings of loss and uncertainty; concern for family; and difficulties in coping. **Please see Volume V, Issue 1 of "Network" for the full article, either online or on request to the Registry.**

STUDY ALERT

We are looking for teen volunteers with FAP to help us complete the questionnaire study by Dr. Esplen. We will also be contacting patients with FAP from the Registry who had a colectomy at age12 or younger about participating in Dr. Durno's study. If you are interested in being part of either study or have any suggestions, please contact Terri Berk at tberk@mtsinai.on.ca or 416-586-4800 ext. 8334.

EDITOR'S MAILBAG

Spring Holter Msc, Genetic Counsellor

Q I have a hereditary colorectal cancer syndrome and am at high risk of developing colorectal cancer. Why should I have screening if I am going to develop colorectal cancer anyway?

A People with hereditary colorectal cancer conditions HNPCC (Lynch Syndrome), Familial Adenomatous Polyposis (FAP), Attenuated FAP (AFAP), MYH-Associated Polyposis (MAP), Peutz-Jeghers Syndrome (PJS) and Juvenile Polyposis (JP) have a significantly increased chance of developing colorectal cancer. However, having a mutation in one of the genes that cause these conditions does not automatically mean that you are going to develop colorectal cancer. Colorectal cancer is one of the most preventable and treatable cancers if detected early. By following the colorectal cancer screening guidelines recommended by your genetic counsellor and doctor, your chance of developing colorectal cancer with any of these conditions is greatly reduced.

Q I was recently diagnosed with HNPCC (Lynch Syndrome) and I am concerned about my chance of developing other cancers. Is there anything I can do to prevent these cancers?

A Hereditary Non-Polyposis Colorectal Cancer (HNPCC) or Lynch Syndrome is not just a hereditary colorectal cancer condition. For many families, there are several other types of cancer that can be associated with HNPCC. These include endometrial, ovarian, small bowel, stomach, upper urinary tract, primary hepatobiliary, primary brain, and a rare skin cancer called sebaceous carcinoma. Although the chance of developing these non-colorectal cancers is less than the risk for colorectal cancer, screening may be an option for some families. The second most common HNPCC cancer is endometrial (uterine) cancer for women. There are early signs and symptoms associated with endometrial cancer such as heavy or irregular vaginal bleeding. Watching out for these symptoms and having them investigated promptly by a specialist is one way to catch endometrial cancer in its early stages. Gynecological cancer screening has not been proven to be effective in the prevention of the gynecological cancers associated with HNPCC. However, annualtrans-vaginal ultrasound, with endometrial biopsy, if symptomatic, and a CA-125 blood test are available for women who want to be more proactive about gynecological cancer screening.

Based on the family history, some families may be offered additional screening for the non-colorectal cancers; however, the effectiveness of these screening techniques is limited.

Colonoscopy Recommendations for Hereditary GI Cancer*

	HNPCC	FAP	AFAP	MAP	PJS	JPS
Frequency (years)	1-2	1	2-3	2-3	2-3	1-3
Age	20-25	10	15	20-25	15	15

*These are general recommendations and may vary depending on personal and family history

Upper GI Screening Recommendations*

	HNPCC	FAP	AFAP	MAP	PJS	JPS
Technique		EGD	EGD	EGD	EGD OR UPPER GI SERIES	EGD
Frequency (years)		1-5	1-5	1-5	2-3	2-3
Age		25	25	25	10	15

*These are general recommendations and may vary depending on personal and family history

It is essential that your family doctor knows about the diagnosis of HNPCC and understands the implications so that changes in your general health can be properly evaluated.

Q Why are the colonoscopy recommendations so frequent and made at such young ages?

A One of the most common features of hereditary cancer conditions are young ages of cancer diagnosis. The average age of colorectal cancer diagnosis in the general population is in the mid-60s. For the hereditary colorectal cancer conditions, the average age of diagnosis are less than age 50, and the development of polyps is even younger. Some adolescents, young adults, and even older adults may not want to follow the recommended screening guidelines because they don't feel sick or may not be fully aware of the implications of having a hereditary colorectal cancer condition. It is important to understand that beginning colonoscopy at a young age is meant to prevent the development of cancer. Research has also shown that for some of the hereditary colorectal cancer conditions, the time it takes for a polyp to develop into cancer is faster than for sporadic colorectal cancers. Even if your last colonoscopy didn't identify any polyps, you shouldn't delay your next colonoscopy longer than what is recommended because polyps, and possibly a cancer, could develop in the interim. A genetic counsellor is available to discuss any concerns you or your family members may have about the frequency and age of colonoscopy recommendations.

Q My family doctor previously recommended that I have colon cancer screening using a Fecal Occult Blood Test. Should I be doing this?

A A Fecal Occult Blood Test (FOBT) is a colorectal cancer screening test that is available to the general population because it is an easy and cost-effective way of offering colorectal screening to individuals over the age of 50. FOBT is not appropriate for individuals who have hereditary colorectal cancer conditions. Colonoscopy is currently the "gold standard" of screening for individuals at high risk of developing colorectal cancer. If you are from a family that has a known gene mutation for one of the hereditary colorectal cancer conditions and genetic testing confirmed that you did NOT inherit the family mutation, then FOBT may be an option for you. If you do not carry the family mutation, you would not be considered at high risk for colorectal cancer, but you do still have the background risk that everyone has which is approximately 6 per cent. You should discuss colorectal cancer screening with your family doctor when you turn age 50.

Given the multiple cancer risks associated with each of these hereditary cancer conditions, it is important to adhere to the screening guidelines recommended by your specialists and genetic counsellors. Following these guidelines can prevent you from being diagnosed with a cancer and could even save your life.

REGISTRY UPDATE

HNPCC Education Night

Melyssa Aronson MSc (C) CGC

We are planning our 3rd Bienniel HNPCC Education Night. This is an evening that we organize for our families with the inherited cancer syndrome, Hereditary Non-Polyposis Colorectal Cancer (HNPCC). HNPCC may be called different names, such as Lynch Syndrome, and in some families, Muir-Torre Syndrome, or Turcot's syndrome. Our first evening took place in October, 2003 and we were inspired to organize this night after one of our HNPCC patients developed signs of endometrial cancer that were largely overlooked because no one connected a risk of endometrial cancer with her diagnosis of HNPCC. She was eventually diagnosed with endometrial cancer, and she later found a letter from our Registry outlining the symptoms to watch for, including post-menopausal bleeding which she had been experiencing for over a year before her diagnosis. We felt that her story should be told. Afterward, she was invited, along with other patients, to share their experiences. Expert physicians in HNPCC were available to speak to our families about cancer risk, surveillance, and updates in HNPCC.

At our first evening in 2003, Dr. Steven Gallinger, head of our Familial Gastrointestinal Cancer Registry, spoke of the recent advances in HNPCC. Dr. Joan Murphy discussed gynecological issues in women with HNPCC, and Dr. June Carroll spoke about communication with your family doctor about symptoms and screening for HNPCC. We also had two patients share their story, which certainly connected with most people who attended that night.

Our 2nd biennial HNPCC education night was held on May 25, 2005 and was another over whelming success. Back by popular demand, Dr. Gallinger updated us on advances in HNPCC research. We invited Dr. Mark Silverberg, gastroenterologist at Mount Sinai Hospital, to give an overview on colorectal screening. Our audience was able to see how a colonoscope works, along with other screening tools for colorectal cancer screening. We also wanted to show our patients with HNPCC the types of skin lesions that are sometimes seen in these families. Dr. Jim Shaw from University Health Network gave an excellent talk on sebaceous adenomas and keratoacanthomas. As always, our patient speakers shared their stories, and so many of our attendees related to their stories of cancer and survival.

We are pleased to announce that our 3rd HNPCC education night will be moderated by Dr. Zane Cohen. Dr. Gallinger will present an update on HNPCC. Dr. Rob Gryfe, a colorectal cancer surgeon at Mount Sinai Hospital will discuss surgery and screening in HNPCC.

We have also invited Dr. Mary Jane Esplen, psychologist at Toronto General Hospital, and leader of an HNPCC support group, to discuss the impact of HNPCC on a family. Our evening will include patients telling their stories and a question-and-answer period for our audience. If you have HNPCC (Lynch syndrome), or are suspected of having HNPCC, please look for a flyer with details of this night. We close by sharing some audience comments from our last education night:

"Getting updates that impact my family are important."

"Very interested in these education nights."

"Well worth attending. We came over 100 km to be here."

"Excellent — Very good question and answer period."

"Thank you for your stories and advice — live strong!"

We are pleased to announce the opening of the Dr. Zane Cohen Digestive Diseases Clinical Research Centre the new home of the Familial GI Cancer Registry. Dr. Cohen, colorectal surgeon and Co-Director of the Registry, has brought us together with health-care professionals from the Inflammatory Bowel Disease program to promote improved treatment and research for patients and their families.

NETWORK

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Dr. Zane Cohen Digestive Diseases
Clinical Research Centre

Editor: Terri Berk, Clinical Co-ordinator

If you would like to know more about inherited bowel diseases, please write or call us.

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GLOSSARY

Colectomy: The surgical removal of all or part of the colon.

Esophagogastroduodenoscopy (EGD): A procedure in which the esophagus, stomach, and duodenum (first portion of the small intestine) can be examined or viewed on a monitor, using a flexible tube (scope).

Familial Adenomatous Polyposis (FAP): An inherited disease in which multiple polyps (hundreds or thousands) develop in the colon and rectum, which can become cancerous. Attenuated FAP (AFAP) is a milder, later-onset form of FAP, characterized by the development of less than 100 polyps in the colon.

Gastroenterologist: A doctor specializing in diagnosing and treating people with diseases of the digestive system.

Hereditary Non-Polyposis Colorectal Cancer (HNPCC): A hereditary condition which poses a high risk of developing colorectal cancer, as well as an increased risk for endometrial and other gastrointestinal cancers.

Juvenile Polyposis Syndrome (JPS): A condition characterized by the development of a specific type of polyp (hamartomas) in the gastrointestinal tract, usually within the first 20 years of life. It can develop sporadically or it can be inherited.

Keratoacanthoma: A benign, rapidly growing skin tumor, usually forming on exposed areas of skin, typically developing as one or more localized lesions, and can heal spontaneously.

Muir-Torre Syndrome: A subtype of HNPCC, characterized by a combination of characteristic skin tumours and colorectal cancer.

MYH-Associated Polyposis (MAP): A recently identified hereditary condition characterized by multiple polyps and colorectal cancer. MAP is caused by genetic changes, called mutations, in the MYH gene.

Peutz-Jegher's Syndrome (PJS): A hereditary syndrome causing the development of dark freckles on the skin, characteristic polyps in the small intestine, small bowel, stomach, colon, and some times in the nose or bladder, and an increased risk of some cancers.

Sebaceous Adenoma: A benign tumor developing on the skin, usually appearing as a small, yellow bump.

Sigmoidoscopy: An internal examination of the lower large bowel (colon), using a small camera attached to a flexible tube, which is inserted into the rectum and colon.

Turcot's Syndrome: A hereditary syndrome characterized by brain tumors and multiple colorectal polyps.