



# COOPERATIVE FAMILY REGISTRY FOR COLORECTAL CANCER STUDIES

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University of Southern California Consortium — Dartmouth Medical School — Cleveland Clinic Foundation  
University of Arizona — University of Colorado — University of Minnesota — University of North Carolina

## Greetings!

Thanks to families like yours, the Colon CFR is striving to find ways to prevent colorectal cancer. Enclosed with this newsletter is NCI's most recent Colon CFR newsletter, as well as a handy colorectal cancer screening card to help you and your doctor review your screening recommendations and keep track of test dates.

## Researchers in the News

The International Society for Gastrointestinal Hereditary Tumours, the First Conference of InSiGHT, met in Newcastle UK on June 13-17, 2005.

Dr. Steve Gallinger from Cancer Care Ontario and Dr. Jeremy Jass from Australia represented the Colon CFR with paper presentations.

The USC consortium was represented with papers presented by Dr. John Baron from Dartmouth, Dr. James Church and Ms. Susan Fay from the Cleveland Clinic.

Ms. Ellen McGannon, Project Coordinator at the Cleveland Clinic co-chaired a session on psychosocial and genetic counseling issues in which Ms. Fay, a family coordinator and data manager at the Cleveland Clinic, presented two papers on the benefits of counseling in HNPCC patients and use of a wallet-sized card to record HNPCC surveillance and follow up examinations (see insert).

## Research Helps Families Around the World

### Registry Expands to 11,000 Families

Robert Haile, DR.PH  
Principal Investigator, USC Consortium

As Dr. Daniela Seminara describes in her update in the international newsletter (see insert), we have a lot to celebrate in terms of accomplishments, thanks to families such as yours.

Internationally, we now have over 11,000 families and 30,000 individuals in the Colon CFR (Cancer Family Registry). Even this early in the Colon CFR, we're beginning to generate important research results.

We've recently had two articles published in the prestigious *Journal of*

*the American Medical Association (JAMA)*. One article describes a better, more thorough method for detecting mutations that result in colorectal and other cancers in families with a familial syndrome called Hereditary Nonpolyposis

*"We've a lot to celebrate in terms of accomplishments — thanks to families such as yours."*

Colorectal Cancer (HNPCC).

In the other JAMA article, we describe the risk of colorectal cancer and other cancers in HNPCC families. It turns out that the syndrome HNPCC may reflect at least two different types of syndromes. In one group, defects in a set of genes called the mismatch repair genes (MMR genes) appear to play an important role. In the other group, these MMR genes do not appear to play an important role.

In HNPCC families with evidence of a defect in an MMR gene, family members are at increased risk for a number of

cancers, including colorectal, endometrial, uterine and other cancers. In contrast, HNPCC families with no evidence of a defect in an MMR gene appear to have an increased risk only of colorectal cancer, but not the other cancers. If this result is confirmed by other studies, it will change the clinical management of HNPCC families.

In addition, colleagues in Canada, who are also part of the Colon CFR,

## New CFR website

The National Cancer Institute now has a public website featuring the Colon CFR. The website is:

<http://epi.grants.cancer.gov/CFR/>

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## All in the Family

### How genetic testing helped a family come to terms with an inherited cancer syndrome

When Antoinette Marini was diagnosed with colorectal cancer in 1960 at age 42, everyone agreed it was terrible. She was young to get cancer, and with five children to take care of, cancer was a challenge she didn't need.

In 1977, Antoinette's daughter, Vincenza (Vincy), was diagnosed with colorectal cancer at age 34 and had surgery to remove part of her colon. At age 47, she developed uterine cancer. Vincy's brothers also developed colorectal cancer: Gabriel at 48, Mike at 54 and Gino at 63. With treatment, all have survived.

#### A Possible Link

Until Gabriel's bout with cancer, this Ohio family was unaware of a possible genetic link. When Gabriel sought medical treatment at the Cleveland Clinic, physicians took a

family history. They immediately contacted Janet Shenal, a family coordinator in the Collaborative Family Registry, to discuss the benefits of participating in the Registry and of genetic counseling.

"Once the registry and genetic counseling are explained, families are more likely to get involved," says Shenal.

Thirteen members of the family (including three

generations — the largest group they'd ever counseled at one time) met with a medical geneticist and a genetic counselor to learn about the role of genes and DNA in colon cancer and genetic testing.

*"Knowing about an inherited disease ahead of time can help you determine what screening exams should be done to reduce cancer risk."*



#### Genetic Testing

Genetic testing is used to identify a gene mutation that causes cancer in a family. Knowing about an inherited disease ahead of time can help you determine what screening exams should be done to reduce cancer risk.

Vincy was tested first. Her results were positive for a gene that causes

hereditary non-polyposis colorectal cancer (HNPCC).

Eight other members of the Marini family also underwent

genetic testing. Although medically valuable, genetic testing raises challenging emotional and family dynamic issues.

Gino's five adult children were genetically tested. "I was so afraid it was going to be four out of five, or even all of them," his wife, Jackie, recalls. Of their five children, a daughter and son inherited the mutated gene. Once a year, they have a colonoscopy.

Vincy's sister, Mary Ragone, has a history of polyps. Mary's two daughters were tested and neither inherited the mutation. Vincy's son decided not to be tested because when he was just 32, a routine colonoscopy detected several pre-cancerous polyps.

#### Spreading the Word

Vincy preaches all the time to anyone who will listen about the value of colonoscopy. "I thank God for the Cleveland Clinic and genetic testing." †

*Excerpted with permission from Family Matters and Cancer Perspectives, publications of the Cleveland Clinic Foundation.*

## Family Health Promotion Project

*Meghan Hobein, MPH, Research Assistant,  
Family Health Promotion Project, University of Colorado*

The University of Colorado Cancer Center is conducting a new study called the **Family Health Promotion Project (FHPP)** utilizing information from the Colon CFR.

The purpose of FHPP is to evaluate alternative approaches for improving cancer screening behaviors in families affected by colorectal cancer. Many family members of those who have been diagnosed with colorectal cancer have been contacted for participation. The project began in 2003 and will end in 2008.

Participants in this study will benefit in a number of ways:

- They'll receive information on how to reduce their chances of getting colon cancer.
- They'll help develop future programs that may assist families like yours, and the chance to contribute to the research resulting from the Cancer Family Registries!

For more information, visit their website: <http://hedwig.mgh.harvard.edu/fhpp>

## African Americans at Higher Risk for Colorectal Cancer

The American College of Gastroenterology (ACG) has recommended that African Americans begin screening for colorectal cancer at age 45 rather than age 50.

This recommendation was included in a special report in the March 2005 *American Journal of Gastroenterology*, along with other recommendations aimed at reducing the incidence of colorectal cancer among African Americans. The report was based on an extensive review of research findings on colorectal cancer screening and issues related to health screening in African Americans.

As a group, African Americans have the highest incidence of colorectal cancer of any racial or ethnic group in the United States. They tend to be younger at the time of diagnosis, and have more advanced disease compared with Caucasians. Survival rates of African Americans are also lower than that of Caucasians.

This new information makes our effort to enroll minority families in the CFR all the more important.

**The ACG Panel urges African Americans to begin screening for colorectal cancer at age 45**

### More Info on Genetic Counseling

To find out more about genetic counseling and how it might benefit your family, check with your local study coordinator to find out what resources are available. Costs vary around the country.

"It's important to spread the word about cancer and how it affects your family."

## A Survivor's Story

*Terri Goode, Research Associate, University of North Carolina Family Registry for Colorectal Cancer Studies*

When North Carolina participant Vinnie Price was diagnosed with colorectal cancer at age 47, he was already taking steps to be healthier.

He had made a conscious effort to cut back on meats, eat more fruits and vegetables, take vitamins, drink more water and exercise regularly. So when the doctor diagnosed him with colon cancer, he asked himself, "What did I do wrong?"



Vinnie Price

Vinnie knew something was wrong when he began experiencing stomach pains and cramps that became more and more severe. He also saw blood in his stool.

"When the pain became unbearable, I went to see my doctor. The lab work came back fine, and the hemocult test didn't even show any blood in the stool."

But when his symptoms persisted, his doctor referred him for a colonoscopy. "The colonoscopy revealed that I had colon cancer."

Since his diagnosis, Vinnie tries not to let things stress him out. His

advice to those who've just been diagnosed with colon cancer is: "Maintain a positive attitude and pray a lot!" He also encourages people to get involved with helping others. "It's important to spread the word about cancer and how it affects your family."

Vinnie joined the Colorectal Cancer Family Registry in 2003. "I was happy to learn that someone out there was concerned about the effects of the disease on the African American community."

*"I was happy to learn that someone out there was concerned about the effects of the disease on the African American community."*

*Vinnie Price, Cancer Survivor*

He has also taken an active role in promoting awareness by talking not just to his family but to friends and co-workers as well. "I was embarrassed and ashamed, but now I'm spreading the word about colon cancer and the Registry. I even Xeroxed copies of the study pamphlets and passed them out!"

When asked how he feels today, Vinnie says, "I'm just glad to be alive. I thank God for that." †

## More Cancer Resources. . .

### Cancer Care —

A nonprofit social service agency helping patients and family members cope with the emotional and financial consequences of cancer. Responds to phone calls and letters from across the United States. Provides information and referrals whenever possible.

Website: [www.cancercare.org](http://www.cancercare.org)  
Telephone: 1-800-813-HOPE (4673)

### Cancervive —

Provides emotional support, education and advocacy to assist survivors as they cope with the aftermath of the cancer experience. Offers one-on-one and telephone counseling nationally.

Website: [www.cancervive.org](http://www.cancervive.org)  
Telephone: 1-800-4-TO-CURE (1-800-486-2873) or 1-310-203-9232

### National Cancer Institute Cancer Information Service —

The Cancer Information Service (CIS) provides telephone information on the latest state-of-the-art treatment for certain cancers and where clinical trials are taking place. Offers information about cancer detection, prevention, diagnosis and support groups. Callers can request free publications. Funded primarily by the National Cancer Institute.

Website: <http://cis.nci.nih.gov> or [www.cancer.gov](http://www.cancer.gov)  
Telephone: 1-800-4CANCER (800-422-6237),  
CancerFax: 301-402-5874

### National Coalition for Cancer Survivorship —

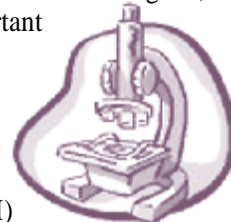
Network of independent groups and individuals offering information, resources and support to cancer survivors, family members and friends.

Website: [www.canceradvocacy.org](http://www.canceradvocacy.org)  
Telephone: 1-877-NCCS-YES (877-622-7937)

## RESEARCH CONTINUED FROM PAGE 1

published evidence suggesting that another gene, called MYH, may be an important cause of colorectal cancer.

The Colon CFR has also been extremely successful in obtaining grants from the National Cancer Institute (NCI) to conduct a number of important studies. For example, we've been funded to study families in which at least two siblings have colorectal cancer in order to identify other genes that may be involved in causing colorectal cancer. We'll actually study genetic markers that scan the human genome. By looking at markers in each chromosome, we hope to find out if there's evidence of a gene being passed down from generation to generation that may increase the risk of colorectal cancer.



In addition, due mostly to the Human Genome Project, science has identified thousands of specific genes. As we learn more about what role each gene plays in humans, some become good targets to study for their possible involvement in colorectal cancer. We call these studies "candidate gene" studies because we have an interest in studying a specific set of genes.

The Colon CFR has just been funded to conduct three candidate gene studies with the following areas of emphasis:

- genes related to folate, vitamin D and calcium
- genes related to obesity
- genes related to aspirin and other non-steroidal anti-inflammatory drugs.

With these studies we hope to better understand the basic biological mechanisms that underlie the development of colorectal cancer and then to develop more effective prevention strategies.

Finally, we're also conducting behavioral research. Our basic science findings won't affect public health unless we can better understand how people make decisions about being screened for colorectal cancer. We also need to learn how people make decisions about changing their behaviors (*e.g. stop smoking, moderate alcohol consumption, enhanced physical activity*) to reduce their risk of colorectal cancer. †

## Confidentiality

Researchers are studying your information and samples in order to learn better ways to prevent, diagnose and treat cancer. Any information obtained from you that can identify you remains confidential as required by law.

Strict measures have been taken to protect your confidentiality at all stages of your participation in this study.

In addition, the Registry has obtained a "Certificate of Confidentiality" from the National Institutes of Health. With this Certificate, our researchers cannot be forced to disclose information that may identify you, even when faced with a court subpoena, in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings.

