

# Genetic Screening Model Identifies Colon Cancer Risk

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WEDNESDAY, June 28 (HealthDay News) -- Scottish researchers say they've developed a method that can better identify people at risk of certain types of hereditary colon cancer, as well as help guide treatment choices for those already diagnosed with colon cancer.

In a study in the June 29 issue of the *New England Journal of Medicine*, the University of Edinburgh researchers detailed the two-stage screening model they developed to genetically identify those with hereditary cancers and the results of the study used to validate their model.

"The method [we developed] allows rapid assessment of predicted likelihood that any given patient newly diagnosed with bowel cancer has a mutation," said one of the study's authors, Dr. Malcolm G. Dunlop, a professor of coloproctology and head of the Colon Cancer Genetics Group at Western General Hospital at the University of Edinburgh.

More than 100,000 new cases of colorectal cancer are diagnosed each year in the United States, according to the American Cancer Society (ACS). Colon cancer is the third most common cancer, and more than 55,000 Americans die each year from the disease.

Finding the genes that cause colon cancer isn't always easy, said Dr. Jay Brooks, chairman of hematology/oncology for the Ochsner Health System in Baton Rouge, La. For some cancers, such as breast cancer, genetic screening is more clear-cut than it is for colon cancer.

"In colon cancer, there are more complicated molecular changes that occur," Brooks said. And, those changes, he added, aren't always easy to detect.

Several models to assess risk have been developed in the past -- the so-called Amsterdam criteria and the Bethesda criteria. These models are commonly used to decide if someone should undergo more detailed and more expensive genetic testing.

However, since some hereditary cancers are missed even when these tests are used, the Scottish researchers hoped to improve upon the existing screening techniques.

The first part of the newly developed model includes information about a person's age, gender, family history of cancer, age of affected family members when diagnosed with cancer and the patients' tumor location. This information is fed into a computer model, which then assesses risk.

Those at high-risk for familial colon cancer then move into the second stage, which involves lab analysis of tumor biopsies.

To develop the current model, the researchers recruited 870 people newly diagnosed with colon cancer. All were under age 55 at the time of diagnosis, which meant they were at a higher-than-average risk of hereditary colon cancer. According to the ACS, more than 90 percent of colon cancers are diagnosed in people over age 50.

Then, without knowing family history, age, and other personal details, the researchers examined blood and tumor samples to look for mutations in the study volunteers' DNA that would increase the risk of colon cancer. They found that 4 percent of those in the study had such mutations.

Using that information, the researchers were able to develop the two-stage model. Then, using a sample of 155 people, the researchers found 62 percent of the known genetic mutations responsible for hereditary cancers.

"The performance of the model was robust among a wide range of cutoff probabilities and was superior to that of the Bethesda and Amsterdam criteria for hereditary nonpolyposis colorectal cancer," the authors wrote.

Knowing who has the genetic mutations common in familial colon cancer can help physicians to guide treatment. However, in this study, survival rates didn't differ between groups.

But, Brooks pointed out, these tests weren't used until after people had been diagnosed with colon cancer. The real benefit from this model would likely come from its ability to assess an increased risk in people who have not yet been diagnosed.

"Survival rates would change if people were diagnosed earlier," said Brooks. He added that it's extremely important for anyone over age 50 to get a colonoscopy, but anyone with a family history of early colon cancer should start colonoscopy screenings earlier.

"Colonoscopy should begin 10 to 15 years before the age of the first known colon cancer in the family. So, if your father had it at age 50, you should start getting screened at 35," Brooks said.

### **More information**

To learn more about detecting colon cancer early, visit the [American Cancer Society](#).