

Colon cancer risk in US traced to common ancestor

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Discovery of founder mutation involves 2 large families in Utah, New York

A married couple who sailed from England to America around 1630 may be the ancestors of hundreds of people alive today who are at risk for a hereditary form of colon cancer.

Researchers from Huntsman Cancer Institute (HCI) at The University of Utah have discovered a founder mutation—a mutation that has been traced from many individuals in the present-day population back to a common ancestor—which may contribute to a significant percentage of colon cancer cases in the United States.

An article reporting the finding was published today in *Clinical Gastroenterology and Hepatology*.

The researchers studied two large families, one in Utah and one in New York, that both carry a specific genetic mutation responsible for increased risk of colorectal cancer. They discovered that the two families share common ancestors—a couple who came to America from England in the 1630s, about the time of the Pilgrims.

“The fact that this mutation can be traced so far back in time suggests that it could be carried by many more families in the United States than is currently known,” says Deborah Neklason, Ph.D., a University of Utah research assistant professor and leader of the study. “In fact, this founder mutation might be related to many colon cancer cases in the United

States.”

The mutation causes a condition called attenuated familial adenomatous polyposis (AFAP). Without proper clinical care, people with the AFAP mutation have a greater than 2 in 3 risk of colon cancer by age 80, compared to about 1 in 24 for the general population. Yet the cancer can be prevented with proper screening and care.

“Knowing one has the condition can be life-saving,” Neklason says. “Not only are affected individuals at greater risk than the general population as they grow older, but precancerous polyps are often found in mutation carriers in their late teens and colon cancer has been diagnosed in individuals in their 20s.”

However, she explains, clinical recognition of AFAP can be difficult because colon cancer develops on average in a person’s 50s and the majority of sporadic, or non-hereditary, colon cancers occur after the age of 50. About a third of people with AFAP also have just a few polyps—again similar to that for sporadic colon cancer—and they may have a limited family history.

“People need to talk with their family, learn their family cancer history, and share this information with their doctors. Doctors need to be aware of AFAP, recognize people at risk, and know the screening and treatment protocols that can prevent colon cancer from developing,” Neklason says.

The Utah family in this study has more than 7,000 descendants spanning nine generations recorded in the Utah Population Database (UPDB), a shared resource for genetics research housed at HCI. Researchers use UPDB to identify and study families that have higher than normal incidence of cancer or other disease, to analyze patterns of genetic inheritance, and to identify specific genetic mutations.

Known individuals in this one family account for 0.15 percent of all colorectal cancers reported in Utah from 1966 to 1995. Based on that percentage, researchers expected to see eight cases of colon cancer from this family among the over 5,000 reported between 1996 and 2003.

But after previous research identified this family as affected by AFAP, aggressive education and clinical intervention resulted in only one mutation carrier in the family being diagnosed with colon cancer during those years.

“Preventing seven cancers may not sound like much,” says Neklason. “But that’s seven colon cancers that didn’t devastate this family. And consider that \$50,000 is a conservative estimate for the cost of colorectal cancer treatment. That amounts to at least \$350,000, and that means a lot for any family.”

Source: University of Utah Health Sciences

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