

Scientists discover major genetic cause of colorectal cancer

August 14 2008

About one-third of colorectal cancers are inherited, but the genetic cause of most of these cancers is unknown. The genes linked to colorectal cancer account for less than 5 percent of all cases.

Scientists at Northwestern University's Feinberg School of Medicine and colleagues have discovered a genetic trait that is present in 10 to 20 percent of patients with colorectal cancer. The findings strongly suggest that the trait is a major contributor to colorectal cancer risk and likely the most common cause of colorectal cancer to date.

If a person inherits this trait -- which is dominant and clusters in families -- the study found the lifetime risk of developing colorectal cancer is 50 percent, compared to 6 percent for the general population. The study will be published August 14 in an advanced on-line report in the journal *Science*.

"This probably accounts for more colorectal cancers than all other gene mutations discovered thus far," said Boris Pasche, M.D., a lead author of the paper and director of the Cancer Genetics Program at the Feinberg School and the Robert H. Lurie Comprehensive Cancer Center of Northwestern University. Pasche also is a physician at Northwestern Memorial Hospital.

"The reasonable expectation is this finding will save some lives," Pasche said. "We will be able to identify a larger number of individuals that are at risk of colorectal cancer and, in the long term, maybe decrease the

cases of colorectal cancer and of people dying from it by being able to screen them more frequently."

Colorectal cancer is the second leading cause of cancer death in the U.S.

The trait, which has been named TGFBR1 ASE, results in decreased production of a key receptor for TGF-beta, the most potent inhibitor of cell growth. With less of this vital protective substance to inhibit cell growth, colon cancer can more easily develop.

In 1998, Pasche and colleagues discovered the first mutation of this gene and in 1999 they showed that it was linked to a higher risk of colorectal cancer.

The results presented in this new study are the first to show that decreased production of this receptor for TGF-beta was present in 10 to 20 percent of patients with colorectal cancer. Decreased production of the same receptor was present in only 1 to 3 percent in healthy control groups.

The findings, which are based on a Caucasian population, need to be confirmed in other studies and may show strong variation between ethnic groups, Pasche said.

Pasche expects that a clinical test will soon be developed that could be offered to families with a history of colorectal cancer and other individuals to determine whether they carry this mutation.

Source: Northwestern University

23 April 2024 from

<https://medicalxpress.com/news/2008-08-scientists-major-genetic-colorectal-cancer.html>

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