MedicalXpress

Researchers demonstrate link between 'jumping gene' and colon cancer

May 10 2016

For more than 50 years, scientists have known of the existence of "jumping genes," strands of DNA material that can move from one location in the genome to another.

Now, for the first time, researchers at the University of Maryland School of Medicine (UM SOM) have demonstrated conclusively that one of these jumping genes appears to play a key role in the generation of cancer. The study was published today in the journal *Genome Research*. It is the first study to ever elucidate this process.

"This is really a new way to understand how tumors grow," said the study's senior author, Scott E. Devine, PhD, associate professor of medicine at UM SOM. "We think it could explain a lot about the mutation process that underlies at least some cancers."

Jumping genes are also known as transposable elements. Dr. Devine and his colleagues focused on one of these elements known as LINE-1, or L1. Until about 25 years ago, researchers thought that L1 had no effect on the genome. But since then, several studies have found that it is active in both the brain and in the body. One study, for example, has found that it plays a role in some cases of hemophilia.

In recent years, scientists have found associations between L1 and cancer. But no study had found a clear link. Devine and his colleagues suspected that L1 might trigger cancer by causing mutations in other genes that suppress cancer. They focused on a tumor suppressor gene

known as APC. This gene is mutated in about 85 percent of colon cancer cases.

The researchers screened tumors from 10 patients, looking for L1 insertions at the APC gene. In one patient, they found evidence of this insertion. They further investigated samples from this patient, and found more evidence that L1 had played a role. In all, they found 27 L1 insertions in the tumor - insertions that were not found in surrounding healthy tissue. They also found evidence that L1 was responsible for inactivating the APC gene. Such silencing may allow tumors to grow unimpeded, Devine says.

Devine notes that the patient whose tumor showed evidence of L1 involvement had a strong family history of cancer. It may be that certain groups or families are more prone to L1-related cancers, he says.

Provided by University of Maryland School of Medicine