Researchers discover gene variant associated with esophageal cancer

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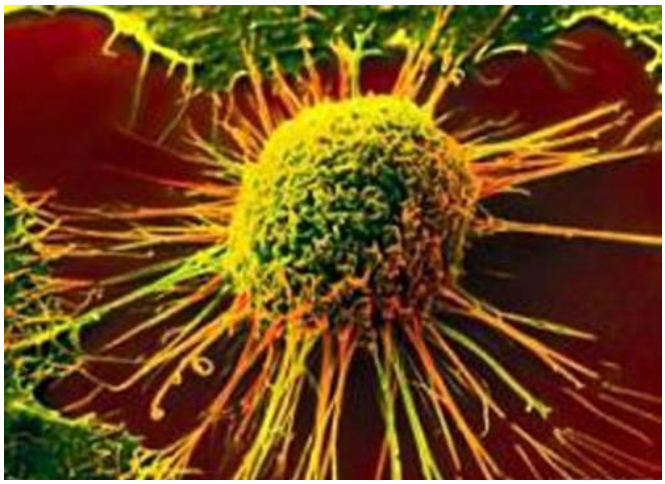

Researchers at University Hospitals Case Medical Center have discovered that a rare genetic mutation is associated with susceptibility to familial Barrett esophagus (FBE) and esophageal cancer, according to a new study published in the July issue of *JAMA Oncology*.

Amitabh Chak, MD, of University Hospitals Case Medical Center's Seidman Cancer Center and Case Western Reserve University School of Medicine, and colleagues set out to identify novel disease susceptibility variants in FBE in affected individuals from a large multigenerational family.

The team, led by Dr. Chak along with collaborating senior author Kishore Guda, DVM, PhD, of the Case Comprehensive Cancer Center, used targeted next generation gene sequencing to find a rare mutation (S631G) in FBE in the uncharacterized gene VSIG10L that segregated with disease in affected family members. Functional studies revealed that this mutation disrupts maturation of the normal esophageal lining.

"Instances of esophageal cancer are on the rise, and the disease has a poor five-year survival rate of less than 15 percent," said Dr. Chak. "However early detection through screening can prevent the development of esophageal cancer. Further research assessing this gene variant may reveal pathways important for the pathogenesis of BE and esophageal adenocarcinoma, leading to earlier detection and better treatment options."

Affecting up to 6.8 percent of the population, BE is a leading predictor of esophageal cancer. Compared with the general population, patients with BE have an 11-fold higher risk of developing adenocarcinoma of the esophagus. But despite a dramatic increase in the disease over the past four decades, there have been few advances in understanding and improving treatment options.

Discovery of this variant, which is the first susceptibility variant discovered in FBE, reveals novel biology in disease pathogenesis, and indicates early screening and close clinical monitoring for individuals harboring this germline variant.

"This is a step forward in combating this deadly disease as we discovered a new way to categorize those at risk for esophageal adenocarcinoma," says Dr. Chak.

Provided by University Hospitals Case Medical Center