Study reveals distinct genomic landscape for young adults with appendiceal cancer

December 9 2020

Credit: CC0 Public Domain

The first study to compare molecular landscapes of early-onset and late-onset appendiceal cancer has revealed distinct non-silent mutations in the tumors of younger patients, setting the stage for the development of

potential therapeutic advances for this rare disease.

The findings, published Dec. 9 in *JAMA Network Open*, were derived from an analysis of tumor genomic profiles of patients under age 50 compared to those 50 and older at diagnosis from the American Association for Cancer Research (AACR) Genomics Evidence Neoplasia Information Exchange (GENIE) registry.

"Appendiceal cancer among young patients harbors a distinct biology, which has potential clinical actionability," said the study's lead author, Andreana Holowatyj, Ph.D., MSCI, assistant professor of Medicine and Cancer Biology at Vanderbilt University Medical Center and an epidemiologist with Vanderbilt-Ingram Cancer Center.

Patients with early-onset appendiceal cancer harbored unique mutation patterns in the genes PIK3CA, GNAS, SMAD3 and TSC2. A targeted therapy for the PIK3CA mutation when it occurs in advanced breast cancer, alpelisib, has already been approved by the U.S. Food and Drug Administration.

"There are currently no targeted therapies specifically for appendiceal cancer patients," Holowatyj said. "After surgery, National Comprehensive Cancer Network (NCCN) guidelines recommend to treat it as a right-sided colon tumor for that reason. But the fact that one of every eight young patients diagnosed with appendiceal cancer has a PIK3CA mutation, a much higher mutation rate compared with late-onset cases, really lends for the potential and the promise to evaluate repurposing existing drugs for early-onset patients diagnosed with this rare malignancy."

Although appendiceal cancer is rare, its incidence is on the rise. Cancer of the appendix is typically found during surgery for acute appendicitis, and even though the rate of appendectomies has been stable over the last

two decades, diagnoses have increased 232% in the United States.

Holowatyj and colleagues used data from 12 of the AACR Project GENIE participating institutions contained within the registry in their analysis. The international registry offers real-world data that would otherwise be difficult to compile. The researchers were able to study the genomic profiles of appendiceal cancer in 385 patients.

"As a rare disease, appendiceal cancer is understudied, and it is challenging to garner sufficient resources and funding to study this malignancy," Holowatyj said. "But given its increasing incidence, especially among young people, it merits further investigation. In contrast to colorectal cancer, where one out of every 10 adults are diagnosed before age 50, this study also shows that one out of every three adults with appendiceal cancer is diagnosed before age 50."

Provided by Vanderbilt University Medical Center

Citation: Study reveals distinct genomic landscape for young adults with appendiceal cancer (2020, December 9) retrieved 7 February 2023 from https://medicalxpress.com/news/2020-12-reveals-distinct-genomic-landscape-young.html