

Cancer of the appendix: Very rare, but genes may play role

November 14 2022, by Cara Murez



While appendix cancer is rare, for a small percentage of patients the

disease may be linked to a particular genetic variant, a new study suggests.

Researchers built on earlier research with this study, finding that 1 in 10 people with cancer of the appendix carries a genetic variant associated with [cancer predisposition](#).

"Based on these data, we are able to recommend genetic counseling and multi-gene panel testing of cancer susceptibility genes for all appendix cancer patients, regardless of age or family history of cancer," said researcher Andreana Holowatyj. She's an assistant professor of medicine and [cancer biology](#) at Vanderbilt University Medical Center in Nashville, Tenn.

"While there is still much to learn from our discovery, we have found the tip of an iceberg—potentially a really big iceberg," Holowatyj said in a center news release.

Historically, experts have not thought this cancer, which affects about one or two people in 1 million annually, was hereditary. The fact that it's rare has made it even harder to understand and study.

For this research, Holowatyj and her colleagues analyzed multi-gene panel testing data from a nationwide clinical testing laboratory in the United States. The data included 131 patients with appendiceal cancer.

Researchers found that 11.5% of the patients had at least one inherited genetic variant in a cancer susceptibility gene.

In the patients whose appendix cancer was the first and only [primary tumor](#), researchers noted a similar prevalence, about 10.8%. This further links the disease to a familial component.

All patients with appendix cancer should consider genetic evaluation, the researchers said. They should also consider testing and [genetic counseling](#) of at-risk family members.

More studies are needed to identify new genetic factors and to use this evidence to tailor which genes are selected in [genetic testing](#) for appendiceal cancer, Holowatyj said.

The findings were published Nov. 11 in the journal *JAMA Oncology* and presented simultaneously at the 2022 Collaborative Group of the Americas Inherited Gastrointestinal Cancer (CGA-IGC) annual meeting, in Nashville.

More information: Andreana N. Holowatyj et al, Inherited Cancer Susceptibility Gene Sequence Variations Among Patients With Appendix Cancer, *JAMA Oncology* (2022). [DOI: 10.1001/jamaoncol.2022.5425](#)

The U.S. National Cancer Institute has more on [appendiceal cancer](#).

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