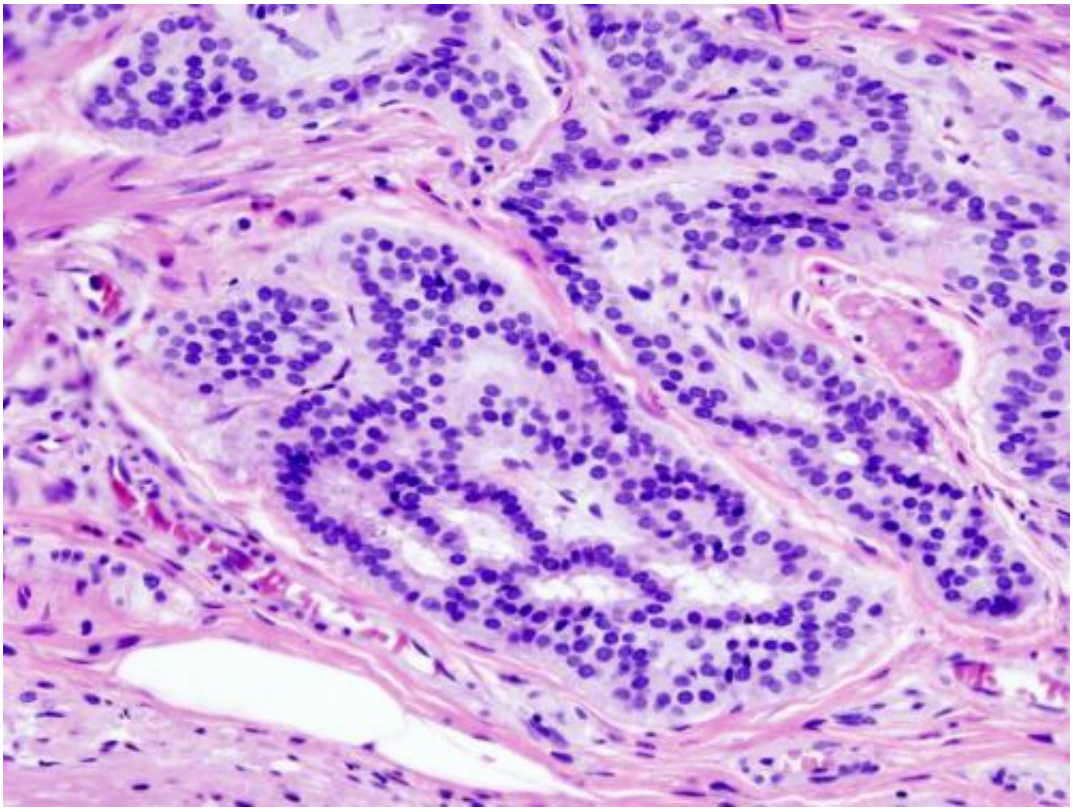
Germline TP53 mutations in patients with early-onset colorectal cancer

March 12 2015



Cancer—Histopathologic image of colonic carcinoid. Credit: Wikipedia/CC BY-SA 3.0

In a group of patients diagnosed with colorectal cancer at 40 or younger, 1.3 percent of the patients carried germline TP53 gene mutations, although none of the patients met the clinical criteria for an inherited cancer syndrome associated with higher lifetime risks of multiple

cancers, according to a study published online by *JAMA Oncology*.

Li-Fraumeni syndrome is an inherited cancer syndrome usually characterized by germline TP53 mutations in which patients can develop early-onset cancers and have an increased risk for a wide array of other cancers including colorectal. The gene's contribution to hereditary and early-onset colorectal cancer is needed for clinicians to counsel patients undergoing TP53 testing as part of a multigene risk assessment, according to the study background.

Sapna Syngal, M.D., M.P.H., of the Dana-Farber Cancer Institute, Boston, and coauthors estimated the proportion of patients with early-onset colorectal cancer who carry germline TP53 mutations. Participants were recruited from the Colon Cancer Family Registry from 1998 through 2007 and were those individuals who were diagnosed with colorectal cancer at 40 or younger and lacked a known hereditary cancer syndrome.

Among 457 eligible patients, six (1.3 percent) of them carried germline missense TP53 alterations and none of the patients met the clinical criteria for Li-Fraumeni syndrome, according to the results. The authors note the fraction of patients found to carry germline TP53 mutations was comparable with the proportion of inherited colorectal cancer thought to be attributable to APC gene mutations.

"With modern techniques for comprehensively genotyping cancer patients, interpreting such germline results will undoubtedly be a prominent challenge in the counseling and management of at-risk individuals," the study concludes.

More information: *JAMA Oncol*. Published online March 12, 2015. DOI: 10.1001/jamaoncol.2015.0197