Patient study suggests broader genetic testing for colorectal cancer risk

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A new study among more than 1000 colorectal cancer patients at Dana-Farber Cancer Institute has revealed that a surprising number of patients, about 10% in total, show mutations in genes thought to increase the susceptibility to cancers. The research suggests an expanded role for genetic testing of inherited risk, both to help in treating people with the disease and in preventing their at-risk family members from developing it.

Physicians have long known that about 3% of colorectal patients, who have a genetic syndrome known as Lynch syndrome, are at high risk of the disease. "But data from our study says we should be at a much lower threshold for genetic testing in colorectal cancer patients, because we're missing another 7% of people with hereditary risk," says Matthew Yurgelun, MD, a specialist in Dana-Farber's Gastrointestinal Cancer Treatment Center and in the Center for Cancer Genetics and Prevention, the lead author of the study published today in the *Journal of Clinical Oncology*.

"What surprised us was that the likelihood of finding an inherited gene mutation linked to cancer was much higher than what has been shown before," says Sapna Syngal, MD, MPH, corresponding author on the paper, who leads the Gastrointestinal Cancer Genetics and Prevention program at Dana-Farber/Brigham and Women's Cancer Center. "This suggests that every patient with colorectal cancer should raise the idea of genetic testing with their physician, and every physician should raise that idea with their patients, because it has implications not only for patients

but for their family members."

Colorectal cancer is the second most common cause of cancer death in the United States. Colonoscopy screening, however, typically can detect these cancers very early or actually prevent them by removing pre-cancerous polyps.

"We've known for a long time that inherited factors often play into somebody's risk of developing colorectal cancer," says Yurgelun. "We've typically relied on factors like having a strong family history of the disease, or being diagnosed with the disease at a young age, to guide who gets genetic testing to look for well-described hereditary syndromes, such as Lynch syndrome."

The study performed a much wider scope of genetic investigation, using a commercial test panel from Myriad Genetic Laboratories to look for mutations in genes associated with inherited cancer risk. (The company provided partial support for the study, with most funding coming from the National Institutes of Health.) Analyses were performed on blood samples from 1058 people with colorectal cancer who received routine clinical care in recent years and agreed to participate in research. The samples were not pre-selected for situations such as a family history of the disease or diagnosis at a young age.

"We ultimately found that the prevalence of these hereditary cancer susceptibility mutations within this cohort was about one-tenth, quite a bit higher than the traditional thinking," says Yurgelun.

To date, no therapies have been approved for colorectal cancer that target the cancer susceptibility genes among the test panel. However, the Dana-Farber scientists speculate that drugs that target these genes for other types of cancer, such as breast cancer driven by mutations in BRCA genes, eventually may also prove effective against colorectal

cancer.More immediately, broader genetic testing could improve follow-up care for patients after therapy is completed. "If you have a genetic mutation that puts you at risk not only for colorectal but for pancreas or breast cancer, then you need to be followed for those cancers," Syngal says. Similarly, family members with an inherited mutation could be regularly screened not just for colorectal cancer but other forms of cancer where the mutation raises risks.

The researchers will follow up on their findings by further examining the role these cancer susceptibility mutations play in colorectal cancer, including analyses of colorectal tumors to see when the mutations are driving progression of the disease.

"It's time for colorectal patients to have the same awareness about genetic testing that exists for other cancers, such as breast and ovarian cancer," says Syngal. "More than a million people in this country have Lynch syndrome, but the vast majority of them are unaware that they carry it," she says.

The path to broader genetic testing is becoming easier because costs of these tests have dropped so dramatically, adds Yurgelun. "If we can find a larger fraction of the population where we can identify a risk ahead of time, for them and even more importantly for their healthy family members, and then intervene to prevent cancer or reduce cancer risk, this testing may easily become cost-effective because the potential payoffs are so high," he says.

Provided by Dana-Farber Cancer Institute

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