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Study: New process that can save at-risk cancer patients is effective and significantly less costly

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People at risk for a certain form of colon and other types of cancer may soon have a better chance at surviving or even avoiding the diseases, thanks to a new study done by the Intermountain Clinical Genetics Institute at LDS Hospital.

The Intermountain Heathcare group of scientists used sophisticated computer modeling to determine a reliable and cost-effective way to identify patients who may have Lynch syndrome, an inherited cancer syndrome that occurs in people who carry a genetic mutation in one of the DNA mismatch repair genes.

The mismatch repair (MMR) genes usually help to repair DNA damage that happens to all of us as a part of daily life. But patients who have genetic mutations in these genes have a substantially increased risk of developing colon, uterine, pancreatic and urologic cancers. For some patients, the lifetime risk approaches 80 percent.

"Being able to identify people who carry a gene change is profoundly important because earlier and more frequent screening — not just for colon cancer, but also for other cancers — could save their lives. It could also save the lives of relatives who have no idea that they may share the increased risk for cancer," says Marc S. Williams, MD, director of the Clinical Genetics Institute at LDS Hospital, and a member of the team that conducted the study, which is published in the August edition of the

American *Journal of Managed Care*.

A national report on colon cancer a few years ago recommended screening all patients with colon cancer for Lynch syndrome, but stopped short of outlining the best way to do it.

"There are many tests available that can be used in different combinations to diagnose Lynch syndrome, but there's little clarity about what's the most effective and efficient approach," says James Gudgeon, an analyst with the Clinical Genetics Institute who led the study.

"Doing full genome sequencing on all colorectal cancer patients would uncover virtually all MMR mutations, but the majority of these patients don't have them. Sequencing costs $4,000 to $6,000 per person, so it would be incredibly costly and inefficient to test everyone."

The Intermountain team set out to determine if they could develop a system for screening colon cancer patients with existing tests that could keep costs down, but also ensure accurate results. The team gathered information from a variety of sources, including Intermountain patient data, published literature and outside groups to define the best approach to screening. They came up with a plan that relies on two relatively inexpensive tests to eliminate possible Lynch patients before doing full genome sequencing.

So far, 272 colon cancer patients have been screened according to the group's system, with 261 individuals ruled out as carriers of the abnormal genes.

"That left only 11 patients who we would recommend going forward with the full genome sequencing test," says Dr. Williams. "That represents the wisest use of the expensive resource of full sequencing."

The benefits extend not only to the colon cancer patients, but also to members of their extended families, who may also have the MMR mutation.

"Confirming the Lynch diagnosis changes the way we treat the disease. This form of colon cancer has a generally better prognosis than sporadic colon cancer, but it doesn't respond as well to certain kinds of chemotherapy," says Dr. Williams.

It can also make patients more alert to other forms of cancer, triggering earlier and more frequent screenings. Some women who are diagnosed with Lynch may choose to have surgery to remove the uterus and ovaries to prevent ovarian or uterine cancer. One unusual aspect of the project was the methodology used to carry it out, says Dr. Williams.

"The team developed customized computer models to examine an assortment of questions that Intermountain decision-makers were interested in. You don't usually see healthcare systems doing this kind of work. We think application of this type of modeling can help healthcare systems make better decisions about how to best treat patients. It can help improve patient outcomes and increase efficiency, which are two of the benefits Intermountain Healthcare is well known for delivering."

Provided by Intermountain Medical Center