New Mayo Clinic test targets Lynch syndrome, a risk factor for colon cancer

May 18 2011

Mayo Clinic has developed a screening procedure that could dramatically increase testing for Lynch syndrome, a hereditary genetic disorder that raises cancer risk, particularly for colorectal cancer. An estimated 3 percent of colon cancers can be attributed to Lynch syndrome. At least 80 percent of people with Lynch syndrome develop colorectal cancer, many of them before age 50.

In the past, as few as 50 percent of patients who fit the profile for possible Lynch syndrome were tested before or after surgery. Now, a group of Mayo Clinic researchers has developed and tested a protocol that could raise the level of testing to nearly 90 percent, helping doctors make important decisions on the timing and delivery of care for patients with the disease. Their findings were presented at The American Society of Colon and Rectal Surgeons Annual Scientific Meeting (http://www.fascrs.org/), May 14-18, 2011 in Vancouver, British Columbia, Canada.

Mayo Clinic researchers in 2003 started testing all newly diagnosed colorectal cancers in patients under 50. Biopsies of the cancers were sent to pathologists, who conducted Microsatellite Instability (MSI) testing on them. MSI testing looks for certain mutations in genes that repair DNA by testing 10 different DNA markers for irregularities. Patients categorized in the "high" group for microsatellite instability were offered additional testing for Lynch syndrome and genetic counseling.

Over the five-year study, 210 of 258 newly diagnosed patients under age

50 who underwent colorectal cancer surgery at MCR had the MSI testing. Of those, 13 percent had MSI-H tumors. Overall, 88 percent of the high-risk group had tests, and the protocol caught 11 percent of MSI-H tumors that would have otherwise been missed.

"Probably the most significant result of this research is that it has stimulated our multidisciplinary team of geneticists, pathologists, gastroenterologists and surgeons to develop new clinical pathways that will direct patients at risk to providers experienced with management of Lynch Syndrome," says Eric Dozois, M.D. who has organized the multidisciplinary Young Onset Working Group and is the lead researcher on this project. This ensures appropriate evaluation and genetic and surgical counseling before critical treatment decisions are made, thus allowing patients and referring physicians to be fully informed regarding options for treatment, especially risks and benefits.

"The benefit of this testing to the patient and their family is huge," says research fellow Rajesh Pendlimari, M.B.B.S. "If they have Lynch syndrome and will, therefore, be more prone to getting cancer, they can get screened more regularly. Plus, with it being a hereditary condition, family members can also get tested." As a result of this testing, cancer may be caught earlier and physicians may be more proactive in treatment. The testing should be done before surgery, because a diagnosis may change the course of treatment. Testing after surgery also is beneficial; the knowledge gleaned can affect future care for patients and their families.

More information: http://www.mayoclinic.com/health/lynch-syndrome/DS00669

Provided by Mayo Clinic