Researchers develop clinical screening program for no.1 genetic cause of colon cancer

February 14 2013

(Medical Xpress)—Cleveland Clinic researchers have found that colorectal cancer outcomes could be improved with regular genetic screening for Lynch syndrome, the most common hereditary, adult-onset cause of colorectal cancer, as published in the online version of the *Journal of Clinical Oncology*.

Lynch syndrome is the most common genetic cause of colon cancer in adults. The study found that a universal screening program of all colorectal cancers surgically removed at Cleveland Clinic resulted in increased identification of Lynch syndrome patients by successfully referring individuals whose tumors screen positive to genetics professionals. Identification of Lynch syndrome will lead to significantly improved outcomes for colorectal cancer patients and their family members.

Most colorectal cancers are sporadic, and are not due to inherited genetic causes. More than 140 million Americans are diagnosed with colorectal cancer each year. With one out of 35 colorectal cancer cases resulting from Lynch syndrome, a hereditary condition that predisposes to a wide variety of cancers at earlier ages, a clinical recommendation was established in 2009 to screen all colorectal patients for Lynch syndrome. With four million Americans having Lynch syndrome each year, and most going unrecognized, a systematic manner of screening for this condition presented a challenge. Furthermore, another challenge is

ensuring that all individuals whose tumors screen positive for potential Lynch syndrome are referred to genetics professionals.

By identifying Lynch syndrome patients, measures can be taken to thwart the development of additional cancers in the patient, as well as increase surveillance in the patient. Once Lynch syndrome has been identified in a patient, his/her family members can be referred to genetics professionals to determine if they too have inherited the same condition. As patients who carry the Lynch gene undergo proactive colon and other cancer screening for the earliest diagnoses, lives are saved.

The study recommends that a successful program should eliminate "silos" and instead include collaborative representation from colorectal surgery, gastroenterology, gynecology, pathology, genetics, as well as bioethics and oncology. The program must also designate who is to report results to patients and facilitate genetic counseling/testing referral. With clearly assigned roles for patient communication and education, outcomes resulting from Lynch syndrome will be significantly improved.

Led by Charis Eng, M.D., Ph.D., Hardis Chair and Founding Director of the Cleveland Clinic Lerner Research Institute's Genomic Medicine Institute, the researchers studied three approaches, moving certain responsibilities (determining which colorectal cancer patients should receive genetic counseling/testing for Lynch syndrome and contacting the patients with results and recommendations) from the surgeon to genetic counselors. Genetic counselors who received genetic pathology reports on colorectal tumor sections, determined patients who fit criteria consistent with Lynch syndrome, and reached out to patients for further testing saw an increase in referrals and number of patients tested for and diagnosed with Lynch syndrome, the study found.

The Cleveland Clinic researchers have addressed one of the two genomic

medicine agenda items of the CDC's Healthy People 2020.

"One of the goals of Healthy People 2020 is to identify as many people who have inherited colon cancer as possible, so they may be referred to genetics professionals for genetic counseling, and management recommendations," said Dr. Eng. "This paper shows successful implementation of a rapid, cost-effective system-wide screening to detect potential Lynch syndrome. Individuals with Lynch syndrome can get colon cancer at a very young age, are susceptible to multiple cases of the disease, and are also predisposed to other cancers. Instituting high risk surveillance early and routinely saves lives and is an example of value-based delivery of healthcare."

The eight-year, collaborative study also highlighted a need to improve patient education regarding genetic counseling/testing. Perceived lack of benefit and underestimated risk of Lynch syndrome and associated cancers caused some patients not to pursue testing.

Provided by Cleveland Clinic