epigenomics

Press release

New Clinical Data Support the Utility of Epi proColon® Blood Test for Colorectal Cancer (CRC) Screening in Patients with Lynch Syndrome (LS), the Most Common Hereditary CRC Syndrome

- *Minimally-invasive blood test could complement colonoscopy in high-risk LS patients who are non-compliant with colonoscopy screening recommendations or may develop precancerous lesions in the interval between colonoscopies*

Berlin (Germany), June 17, 2019 – Epigenomics AG (FSE: ECX, OTCQX: EPGNY; the "Company"), a molecular diagnostics company focused on blood-based detection of cancers using its proprietary DNA methylation biomarker technology, today reported new study results suggesting that Epi proColon®, a colorectal cancer screening test approved for patients who are unwilling or unable to be screened by recommended methods may also be an important complement to colonoscopy for CRC screening in patients with LS, a hereditary disease associated with a 10-80% increase in the risk of developing CRC. The study results appear in the current issue of BMJ Open Gastroenterology.[1]

There is currently no cure for LS, therefore, routine screening that enables early detection and treatment of precancerous lesions has the potential to reduce the risk of CRC and to save lives. Approximately 95 percent of individuals with LS are unaware of their disease status, and nearly a third of patients who know their LS status fail to comply with the recommendation to undergo colonoscopy screening every one to two years. This study evaluated a new approach that could potentially enhance CRC screening for LS patients, which is crucial to reducing the morbidity and mortality associated with the disease.

The study was conducted as a retrospective analysis of preserved tissue and frozen plasma samples from patients with a confirmed diagnosis of LS who underwent either surgical resection for a diagnosis of CRC or removal of a polyp during colonoscopy between March 2006 and February 2019. Study objectives included a comparison of SEPTIN9 gene methylation status (the marker measured by Epi proColon) between patients with LS and patients with non-hereditary forms of CRC and exploratory analyses of the sensitivity and specificity of Epi proColon in LS patients. Key findings from the study include:

- In tissue samples from LS patients, differential SEPTIN9 methylation was found in 97.3% of primary CRC and 90.0% of advanced adenomas, demonstrating that LS-related neoplasia frequently produce the SEPTIN9 biomarker.

[1] Hitchins MP, Vogelaar IP, Brennan K, Haraldsdottir S, Zhou N, Martin B, et al. Methylated SEPTIN9 plasma test for colorectal cancer detection may be applicable to Lynch syndrome. BMJ Open Gastro. 2019;6:e000299. doi:10.1136/bmjgast-2019-000299.

- Of 20 plasma samples collected between 138 and 1 days prior to surgical resection of a primary CRC tumor, all had valid tests and 14 were SEPTIN9 positive, for a sensitivity of 70.0%.
- Of 18 plasma samples collected within one year (328 days to 20 days) prior to a colonoscopy-based diagnosis of CRC, 17 had valid tests, and three of the 17 were SEPTIN9 positive. These three were among five patients with a colonoscopy-based stage I-III CRC diagnosis, for a sensitivity to detect CRC approximately two months prior to diagnosis of 60%.
- Of 13 plasma samples collected after surgical resection of a primary CRC tumor, all had valid tests and 12 were SEPTIN9 positive, for a sensitivity to detect metastatic CRC of 92.3%.
- Of 34 plasma samples from a cancer-free control group, 31 had valid tests and all were SEPTIN9 negative, for a specificity of 100%.

The study authors conclude that Epi proColon may have similar diagnostic performance characteristics in LS patients as in the average-risk population and suggest that a larger, prospective study to confirm these preliminary findings is warranted.

”The sensitivity and specificity that the Epi proColon blood test demonstrated in this first study in LS patients are very promising,” said Megan Hitchins, PhD, Associate Professor, Center for Bioinformatics and Functional Genomics at Cedars-Sinai and lead author on the study publication. “Although preliminary, the study results suggest that Epi proColon may have potential as a screening tool for colorectal cancer in LS and for post-surgical detection of metastatic disease as well. Epi proColon may provide an alternative CRC screening method for the LS patients who are non-compliant with colonoscopy screening recommendations. Confirmation of our findings in larger, prospective studies would be an important advance in the clinical management of LS. Dr. Henry Lynch, the first to identify LS cancers as hereditary and the senior author on this, passed away last week. I can think of no better way to honor his already substantial legacy than to continue advancing the care and outcomes for LS patients.”

“Although colonoscopy is the gold standard for CRC screening, diagnosis and polyp removal, compliance with colonoscopy screening recommendations is suboptimal, even among patients with an LS diagnosis who know that they are at increased risk of CRC and could benefit from CRC screening every one to two years,” said Greg Hamilton, Chief Executive Offer of Epigenomics AG. “Pre-cancerous lesions can progress to invasive CRC very rapidly in LS patients, so the 10-year colonoscopy screening interval recommended for average-risk individuals does not adequately protect patients with undiagnosed LS from a late-stage CRC diagnosis, even if they are compliant. Alternative screening methods that could be used in the interval between colonoscopies, such as Epi proColon, might increase early detection of CRC for undiagnosed LS patients and other individuals with increased, but undiagnosed CRC risk.”

About colorectal cancer (CRC)

Colorectal cancer remains a leading cause of cancer death in the United States. Although screening and early detection of colorectal cancer can save lives, about 35% of eligible U.S. patients are not being screened regularly. The unscreened population disproportionately results in 43% of new colorectal cancer cases and about 76% of colorectal cancer deaths and costs. Approximately $18 billion is spent annually on this preventable disease. Over $13 billion is spent on cases from unscreened individuals.

By increasing screening and detecting more cancers early, the costs and deaths from this disease both can be addressed.

About Lynch Syndrome

Lynch Syndrome (LS) is a hereditary disorder caused by a mutation in one of at least five genes (MLH1, MSH2, MSH6, MPS2 or EPCAM) that are involved in the repair of mistakes that occur when DNA is copied in preparation for cell division. The accumulation of errors in DNA can result in cells that grow uncontrolled and may become cancerous. Individuals with LS have an increased risk of developing colorectal cancer, endometrial cancer, and various other types of aggressive cancers, often at a young age. Lynch tumors are extremely aggressive and may metastasize more rapidly (less than two year) than sporadic cancer (often more than five years). Consequently, it is recommended that LS patients undergo CRC screening more frequently than the individuals with average CRC risk (every 1-2 years for LS compared with every 10 years in the average risk population) and beginning at an earlier age. A key challenge in achieving these screening recommendations is that only 5% of affected individuals are aware of their LS status.

About Epi proColon®

Epi proColon® is indicated for colorectal cancer screening in average-risk patients who are unwilling or unable to perform colorectal cancer screening by colonoscopy and stool-based methods. It is a qualitative, in vitro diagnostic blood test for CRC that uses real-time PCR to detect methylation of a target DNA sequence within the Septin 9 gene promoter; methylation of this DNA sequence is associated with the occurrence of CRC and can be detected in cell-free DNA that circulates in the plasma.

For patients, the test only requires a simple blood sample draw as part of routine healthcare provider visits. There are no dietary restrictions or alterations in medication required for the test. The sample will be analyzed at a national or regional diagnostic laboratory.

Epi proColon is recipient of the 2019 Excellence in Molecular Diagnostics by Corporate LiveWire's Innovation and Excellence Awards.

For more information on Epi proColon, visit www.epiprocolon.com.

About Epigenomics

Epigenomics is a molecular diagnostics company focused on blood-based detection of cancers using its proprietary DNA methylation biomarker technology. The company develops and commercializes diagnostic products across multiple cancer indications with high medical need. Epigenomics' lead product, Epi proColon, is a blood-based screening test for the detection of colorectal cancer. Epi proColon has received approval from the U.S. Food and Drug Administration (FDA) and is currently marketed in the United States, Europe, and China and selected other countries. Epi proLung® and HCCBloodTest, blood-based tests for lung and liver cancer detection, has received CE mark in Europe.

For more information, visit www.epigenomics.com.

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