New research may lead to earlier diagnosis and treatment of primary biliary cirrhosis in families

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Mayo Clinic researchers have found that first-degree relatives (i.e., parents, siblings, children) of patients with primary biliary cirrhosis (PBC) are more likely to have the biomarker of the disease in their blood. Armed with this new information, physicians could screen and assess first-degree relatives of PBC patients with a simple blood test, enabling them to diagnose and treat more patients before the disease causes irreversible liver damage. These findings were published in this month's issue of *Hepatology*.

PBC is a chronic liver disease that affects nearly 50,000 people (primarily women) in North America. In individuals who have PBC, the bile ducts are slowly destroyed, causing harmful substances to build up in the liver and sometimes resulting in irreversible scarring of liver tissue and liver failure. About half of PBC patients have no symptoms and are diagnosed following abnormal results of routine liver tests.

Anti-mitochondrial antibodies are the biomarker, or the substance that correlates with the risk or presence of a disease, for PBC. This study, the largest of its kind, tested for anti-mitochondrial antibodies in 306 first-degree relatives of adult PBC patients and 196 healthy adults. The prevalence in first-degree relatives was 13.1 percent, compared to 1 percent in the control group of healthy adults. Even greater prevalence was found in female relatives, with 20.7 percent of sisters, 15.1 percent of mothers and 9.8 percent of daughters having anti-mitochondrial antibodies in their blood. While testing positive for anti-mitochondrial antibodies does not always lead to a diagnosis of PBC, the presence of these antibodies indicates a predisposition to develop the illness, particularly in the context of family history of the disease.

"Most PBC patients have no symptoms, but early detection is important because timely treatment can slow the progression of the disease before liver failure occurs," says Konstantinos Lazaridis, M.D., the study's lead author and a hepatologist at Mayo Clinic. "Because collectively one in five sisters of a PBC patient has anti-mitochondrial antibodies in their blood, we think it is worthwhile to screen first-degree relatives, particularly those older than 40 years, for this biomarker. It is a simple, inexpensive blood test that could lead to earlier diagnosis and treatment -- and ultimately, better outcomes for PBC patients."

According to Dr. Lazaridis, the study's findings regarding anti-mitochondrial antibodies in PBC relatives could also be important to better understanding the known genetic predisposition to PBC. His research team plans to continue screening and monitoring first-degree relatives of PBC patients over many years to further examine these findings and to shed light on the cause of this disease.

Source: Mayo Clinic