

Frequently Asked Questions

Familial Barrett Esophagus

The Genomic Medicine Institute of the Cleveland Clinic Foundation is conducting a research study to look for gene(s) that may be associated with Barrett esophagus (BE) and/or esophageal adenocarcinoma (EAC). We are trying to understand how BE develops and how EAC develops from BE, We are particularly interested in families that have 2 or more members affected with BE and/or EAC. Participants should be over the age of 18 years.

What is BE and EAC?

BE is a non-cancerous change of the type of cells that line the esophagus. Although many individuals with BE never develop cancer, the risk of developing EAC increases 30-125 times the risk in the general population to ~1%. EAC is a specific type of esophageal cancer.

How does a genetic research study work?

Identifying genes associated with BE or EAC requires looking at the DNA of people with a condition and comparing it to their family members who do not have the condition. Researchers look for differences in DNA between family members with BE or EAC and their healthy relatives. These differences will help the researchers identify the gene or genes associated with these conditions, which may explain why some people develop BE or EAC and others do not.

Why should I and/or my family participate?

The success of studies of most genetic disorders is dependent on the participation of families in which multiple members have the condition. The more families that participate, the better our chances of making a gene discovery. Identifying a gene often leads to a clinical test, which could be made available for all individuals at risk of having inherited a genetic predisposition to BE and/or EAC. With such information, then such individuals might be able to consider screening or preventive measures as early as possible. Because this is a research study, there is no guarantee that it will be successful. In addition, our research laboratory is not certified by the government to give out the results of genetic testing to individual participants, although the results of the overall study will be available to all participants.

What does participation involve?

Participation in this study involves donating a blood sample (which can be drawn by a local physician and sent to us at our expense). In addition, we will ask for your consent to review your medical records to confirm the diagnosis of BE or EAC and to obtain tissue blocks from previous biopsies or surgeries. A genetic counselor will talk to you by phone or in person to discuss your family history in detail, and you may be asked to contact other family members to see if they would be willing to participate in research as well. You will also be asked to complete a questionnaire regarding your medical history, directed at symptoms of gastroesophageal reflux disease and BE.

What about insurance/confidentiality issues?

In addition to federal and state (Ohio) legislation protecting individuals who have genetic testing from insurance discrimination, we further protect our research subjects by using policies that maximize confidentiality. DNA samples are worked on in the laboratory anonymously (by code number), and all family/medical information is kept in locked files in the Genomic Medicine Institute. Because of these safeguards, we feel the risk of employment or insurance discrimination is very small.

I and/or my family is interested in knowing more. Whom can I contact?

You may contact the familial BE research coordinator of this study, Brandie Heald Leach, MS at: (216) 444-8114 or leachb@ccf.org.