

## Biobank by the numbers



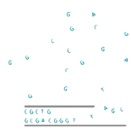
**120,000**  
participants

**300**  
studies



**36,000**  
genotyped  
samples

**11,000**  
sequenced  
samples



**1,100,000**  
stored  
samples

### COVID symptom study

We are inviting ALL Biobank participants to join another Mass General Brigham study called the **COVID-19 Symptom Study**. Participants download and use an app to report symptoms and how they feel. The data from the COVID-19 Symptom Study will be used to better understand the symptoms and potentially any risk factors associated with COVID-19. Join this study by going to:

[covid.joinzoe.com/us-2](https://covid.joinzoe.com/us-2)

## A year unlike others in research

The Mass General Brigham Biobank (formerly called Partners HealthCare Biobank) ceased all standard operations in mid-March, when the unprecedented COVID-19 pandemic started. Like all non-essential workers in Massachusetts, our team went into lock-down.

The lull was short-lived as the research enterprise at Mass General Brigham rapidly shifted into high gear to support multiple research studies focused on understanding, preventing, diagnosing, and treating COVID-19 infection. The Biobank rapidly set up collaborations with studies recruiting patients who had been diagnosed with COVID-19. In parallel, we worked with colleagues across Mass General Brigham to roll out two large studies on the rate of infection of healthcare workers, providing antibody testing for this entire cohort.

We have now built a sizable repository of samples collected at multiple time points from both patients who were admitted to the hospital with COVID-19 and patients recovering from COVID-19. The Biobank has executed more than 50 distributions of samples and data collected from these patients, and we continue to meet the high demand for samples from people who are or were infected with COVID-19.

The intensity of research activity over the past six months has been remarkable. Everyone on the Biobank team is grateful for our ability to support and catalyze this essential research, and we are grateful for your participation, which makes it all possible.

In this newsletter, we provide an update on a campaign we're about to launch to re-consent every participant in the Mass General Brigham Biobank. We plan to vastly expand the scope of research results that are returned to Biobank participants.

## Gearing up for the next decade of research

Starting in late 2020, the Mass General Brigham Biobank will launch a campaign to contact each of its 120,000 participants in order to renew their consent to participate in the study. This means that, as a participant, you may receive a letter, email, and/or phone call from us in the next year asking you to review and sign the Biobank's new consent form.

The purpose of the new consent form is to widen the scope of research results that may be returned to participants. For the past five years, the Biobank has returned research results on actionable genetic variants, which are DNA changes that can increase a person's risk of developing certain health conditions, such as some cancers and types of heart disease. The Biobank's return of research results program will be expanded to potentially include:

- Results on individual responses to different medications that are known to have FDA-approved pharmacogenetic tests. Genetic makeup can impact how our bodies react to certain drugs, and whether certain drugs could cause side effects.
- Hereditary information, such as whether a person is at an increased risk of passing specific health conditions on to their children, even if the parents don't have those conditions.
- We are currently researching, and may eventually be returning, polygenic risk scores, which are scores based on many genetic variations that estimate a person's risk of developing a disease or health condition.

These changes in the Biobank's return of results program will allow us to keep pace with genetic knowledge and contribute even more to personalized medicine research with the goal of improving clinical care. The Biobank's extensive return of research results program expands the potential to offer individual clinical benefits from participating in the Biobank.

You can read the new consent form that covers this expanded return of research results program by logging into Patient Gateway ([www.patientgateway.org](http://www.patientgateway.org)) and clicking 'Biobank' under the Research Menu. You can also email us or call us, and we will help you renew your consent so that you may participate in this expanded return of results program.

# The future of the Biobank: returning polygenic risk scores

Over the last several years, the Mass General Brigham Biobank has been returning actionable genetic variants, a type of genetic result, to some of its participants. These results are qualified as medically important by the American College of Medical Genetics. As the field of genetics rapidly evolves, it has become clear that genetic risk is much more complex than single gene correlations with disease. Many common diseases are actually polygenic, meaning they are influenced by multiple genes.

Calculating polygenic risk scores is a newer way to quantify risk for common diseases like coronary artery disease and type II diabetes. The scores involve taking diverse population data into account. "We want to maximize the predictive value of genetic risk across multiple ancestries so that this information can be beneficial to all participants," said Elizabeth Karlson, MD, one of the Biobank's co-investigators.

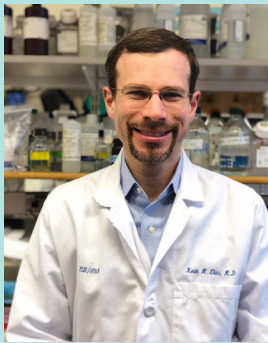
The Biobank's informed consent form was updated to allow for the return of risk categories for common diseases. Ideally, these results will produce actionable medical outcomes, especially in those that fall under high-risk categories. Preventative medical screenings and lifestyle changes are just a few of the outcomes that have been clinically proven to reduce risk of disease in the most high-risk patients.

The current American College of Medical Genetics list of medically actionable genes is only relevant to a small fraction of those enrolled in the Biobank. Polygenic risk scores will be applicable to a larger number of participants, since the concept aims to calculate the risk for a particular disease based on a very large number of genes instead of just a few. Incorporating polygenic risk scores into the Biobank's return of research results program will help further the mission to help researchers understand how one's lifestyle, genes, and environment contribute to overall health.

## Recent research studies

### **Detection of lung cancer risks with serum metabolomics, Leo L. Cheng PhD, Department of Pathology at Massachusetts General Hospital**

There is a need for a lung cancer screening test that is both affordable and appropriate in detecting cancer before symptoms develop. Dr. Cheng and his team at Massachusetts General Hospital are working on a blood test that would be able to successfully screen for lung cancer in its early stages by using data and samples from the Mass General Brigham Biobank. With their study of molecules and their interactions in blood serum, they are hoping to alert possible lung cancer patients about the opportunity to take proactive steps by seeking advanced imaging tests.



### **Hallmarks of gynecologic cancer pathogenesis, Kevin Elias MD, Division of Obstetrics and Gynecology at Brigham and Women's Hospital**

Most women with ovarian cancer are diagnosed at a late stage, significantly decreasing their chances of survival. Dr. Elias and his team at Brigham and Women's Hospital are aiming to create a blood test that screens for the risk of developing ovarian cancer. This test would enable us to detect ovarian cancer earlier and more accurately. To help develop this test, Dr. Elias' team is trying to identify a network of microRNAs, short RNA segments that help control gene expression, associated with the risk of ovarian cancer. Dr. Elias and his team are using blood samples of patients who were healthy when they first joined the Biobank, but then later developed ovarian cancer, in order to discover this network of microRNAs. In the future, Dr. Elias hopes to use this blood test to screen for all gynecologic cancers.

### **Healthy Aging Translational Cohort (HATCH), Jill M. Goldstein PhD, Department of Psychiatry and Neurology with the McCance Center for Brain Health at Massachusetts General Hospital**

18% of the U.S. population is more than 60-years-old, with projections of 25-30% by 2050. Thus, maintaining intact memory function and healthy aging are major public health priorities. Most research on Alzheimer's disease (AD) begins with people over age 65. However, vulnerability for AD begins sometimes 15-20 years earlier. We are developing a clinical risk algorithm (a tool to create a risk score) that integrates genetic, clinical, physiological and brain data for the risk for early cognitive decline, identifying people at ages 50-70 years who may already have accumulated amyloid pathology (one of the first asymptomatic signs of AD), but who are not cognitively impaired as yet. We are using the Mass General Brigham Biobank to identify high and low risk individuals to develop and validate this tool that could be used by physicians to identify high risk individuals for sex-dependent therapeutic strategies for more effective AD prevention.

