Mass General Brigham

2022 Newsletter

Mass General Brigham Biobank

140,000 participants

1.2 million stored samples

150,000 distributed samples

250 studies that received samples

220 studies that received genetic data

2022 retrospective for the Mass General Brigham Biobank

Thank you for your continued participation in the Mass General Brigham Biobank. The Biobank is a large research program designed to help researchers at Mass General Brigham understand how people's genes, lifestyle, and other factors affect their health.

Two years ago, COVID-19 changed the research landscape. Many institutional resources were diverted to support efforts to learn about this disease. The Biobank rapidly reorganized its operation to focus on COVID-19. As a result, the Biobank has distributed over 13,000 samples collected from patients with acute cases of COVID to more than 40 studies.

Now, we are gradually resuming standard research activities across Mass General Brigham. Our plan for 2023 includes expanding recruitment to several Mass General Brigham community health centers, collecting blood samples for all Biobank participants, and generating whole genome sequence data using samples from the Biobank. We also anticipate starting a collaboration to recruit participants at the Long-COVID clinics at both Mass General and Brigham and Women's hospitals.

Collecting blood samples for all Biobank participants

A key focus for the Biobank in the upcoming year is to collect a blood sample for every Biobank participant. To date, blood samples have been collected for only 68% of Biobank participants. To bridge this gap, the Biobank has partnered with the teams that perform blood draws for clinical care in the outpatient setting across Mass General Brigham. Next time you have a clinical blood draw at any Mass General Brigham outpatient site, the person doing the blood draw may add 3 research tubes (2 tablespoons of blood) to your clinical draw.

Return of research results

The Biobank collects blood samples, some of which are processed into DNA. The DNA is then further processed to obtain genetic information. This genetic information is used by many research studies that are trying to understand the association of health and disease with genetics. The Biobank has distributed this genomic information to more than 250 Mass General Brigham researchers. In addition, the Biobank analyzes this information to find genetic results that tell doctors that their patients are at higher risk to develop medically actionable health conditions. A result is medically actionable when there are known steps that a patient and their doctor can take to detect a medical problem early, manage a patient's care, or prevent a health condition from developing.

When the Biobank discovers an actionable finding, a genetic counselor contacts the participant to tell them about the research result. So far, the Biobank has contacted 356 people with actionable findings. Some people decide to have their genetic change confirmed by a clinical laboratory. Other people decide they would rather not know more about the result. That decision is entirely up to every individual, with guidance from the genetic counselor.

Continued on back

The findings may allow participants to make more informed decisions about their lifestyle and medical screening and avoid serious health issues. The Biobank has been returning results for six years now to more than 190 participants. In 2021, 84 participants were contacted about an actionable finding. We expect up to 3% of Biobank participants with genetic testing in the Biobank to have an actionable result.

Some examples of real patient stories about their Biobank findings are listed below.

- A Biobank participant was found to have a genetic risk factor for issues with certain types of anesthesia. They were scheduled for surgery in the next few days. Our team was able to inform the participant and their doctors about this risk before going into surgery. The doctors were grateful for this information so they could use safer medicine for this person.
- The Biobank found that a participant had a genetic risk factor that would increase their risk for breast and ovarian cancer. The participant confirmed this result and acted on the information with their doctors. They had their ovaries, uterus, and fallopian tubes removed and have breast cancer screening every six months. These steps lower the risk of cancer and allow doctors to find cancer in the early stages.
- A Biobank participant was told that they are more likely to have colon cancer due to a genetic risk factor. The participant recently had a colonoscopy that found unhealthy tissue growth, called colon polyps, which may have been caused by the gene change. The participant then reached out to family members and urged them to get tested as well. They told the Genetic Counselor: "It was great to find out that this was a possibility. Nobody wants to hear that they are predisposed for cancer, but to be able to get ahead of it is great."

Starting in the spring of 2022, a new study called eMERGE has been investigating returning other types of genomic risk information. The eMERGE team creates personalized health risk reports for participants for common diseases using genetic and other clinical information. The eMERGE team will study how participants and their doctors use this risk report, and will try to understand whether such reports might change people's behavior. The Biobank will contact many participants to ask them to join the eMERGE study as we continue to build tools to help people make informed decisions about their health.

Study highlights

Associations between head and neck cancer, vitamin D status and HPV infection

Literature shows an increased risk of human papilloma virus (HPV) infection associated with Vitamin D deficiency and an increase in head and neck cancers with individuals with HPV. Dr. Herve Sroussi and his team at Brigham and Women's Hospital are using Biobank samples to examine the role Vitamin D status plays in patients with HPV and head and neck cancers.

Sickle cell and kidney disease

Dr. Sagar Nigwekar and his team at Massachusetts General Hospital are using Biobank samples to investigate renal and dialysis outcomes on individuals with sickle cell disease (SCD) and sickle cell trait (SCT). This study will also explore the impact of renal disease and dialysis on the morbidity and mortality of patients with SCD and SCT.

Estimation of prostate cancer risk by identifying genetic and non-genetic determinants: a bioinformatics approach

With over 100 genetic variants linked to prostate cancer, identifying genetic risk has still not been implemented in a clinical setting. Dr. Adam Kibel and his team at Brigham and Women's Hospital are using Biobank data to identify genetic and non-genetic factors that can be implemented in prostate cancer screening protocols. Their hope is that identifying critical biomarkers may optimize therapeutic interventions.