Thank you for your participation in the Partners Biobank.

The mission of the Partners Biobank is to help researchers understand how people’s health is affected by their genes, lifestyle, and environment. Thanks to the participation of more than fifty-thousand patients at Massachusetts General Hospital, Brigham and Women’s Hospital, Spaulding Rehabilitation Network, and McLean Hospital, the Biobank now supports more than 90 researchers studying many different types of diseases and conditions, including heart disease, diabetes, psychiatric disorders, asthma, cancer, and more.

Plans are underway to expand our Community Advisory Panel, roll out electronic consent to Newton-Wellesley Hospital, North Shore Medical Center, and Faulkner Hospital, and set up infrastructure to collect blood samples at all phlebotomy sites affiliated with Partners HealthCare.

Your participation in the Biobank is an important building block in research that can help us better understand, treat, and even prevent the diseases that might affect your health and the health of future generations. Thank you again for your participation.

In this issue:
- A description of the Biobank’s participation in the eMERGE network
- Detail on recent discoveries that could lead to new therapies for rheumatoid arthritis
- Brief summaries of a few studies that are using Biobank samples and data

The eMERGE Network Advances Genetic Research

The Partners Biobank recently obtained grant funding from the National Institute of Health to join the eMERGE (Electronic Medical Records and Genomics) network, a research consortium sponsored by the National Human Genome Research Institute. The goal of the consortium is to combine the power of genetic information from biobanks across the country with data gathered from electronic medical records (EMR). Together, these data can provide invaluable information that can be used to understand how genetic and environmental factors influence people’s health and contribute to disease.

The Partners Biobank has three broad aims as part of its participation in the eMERGE network:

Examine the genetic cause for common diseases — Study the association of changes in a person’s DNA (known as genetic variants) with increased risk for heart, psychiatric, and immune conditions.

Study the effects of genetic variants on health — Look for new genetic variants in a set of genes to see how often these variants are seen in the general population, and learn how a change in a single gene can impact a person’s health in multiple ways.

Understand how returning results to Biobank participants can influence their medical care — Study the changes physicians make to their patients’ clinical care based on genetic result information and how this affects physician referrals, recommended laboratory tests, changes to prescription medications, medical costs, and the number of family members screened and treated based on the genetic information provided.

The hope is that our participation in the eMERGE network will give us the tools we need to improve genetic risk assessment and create new strategies for disease prevention, diagnosis, and treatment. By combining EMR and genomic data, the Partners Biobank is poised to advance both genetic research and the goals of personalized medicine.
Rheumatoid arthritis (RA) is an autoimmune disease that can lead to inflammation in the joints and other organs of the body. According to the Center for Disease Control and Prevention, this potentially debilitating disease is estimated to affect about 1.5 million adults in the U.S. Treatments for RA have evolved greatly in the last decades, but there is still much progress to be made. Current research is looking to understand why certain people are predisposed to develop this disease and to use this information to prescribe more targeted treatments.

Drs. I-Cheng Ho (pictured above) and Hui-Hsin Chang of Brigham and Women’s Hospital recently published a paper in *Arthritis & Rheumatology* which tackles this research area. Using samples from the Partners Biobank, Drs. Ho and Chang investigated the relationship between a variation in a gene and the risk for the development of RA. The variation, or polymorphism, occurred in a gene that codes for the enzyme (PTPN22), which plays a part in the activation of immune cells. The variation in the gene for the PTPN22 enzyme leads to increased activity of immune cells, which can cause the inflammation characteristic of RA.

The samples used for this research project were found using the Partners Biobank. Participants who carried one copy of the gene variant did not have any symptoms of RA. Still, their samples had higher levels of immune response when compared to the samples without the variation. This shows that this polymorphism can be a potential predictive factor for the development of RA. Having the samples in the Biobank was an important part of the success of this research. According to Drs. Ho and Chang, the frequency of this polymorphism is low, which makes finding enough samples to study difficult. The access to patients’ genetic information through the Biobank samples made this study possible.

The polymorphism in PTPN22 is also associated with high risk of development of other autoimmune disorders, including lupus and type 1 diabetes, even though the features of these diseases are quite different. Understanding how this genetic variation plays a role in the development of autoimmune diseases could potentially allow for close monitoring of high risk patients and early intervention.

Current treatments for RA include the drug methotrexate and biologic agents. Unfortunately, it is still not possible to predict which treatments are likely to give the best response to each individual. Patients can present varying outcomes on the same drug. In their future research, Drs. Ho and Chang are hoping to identify biomarkers that could allow for more targeted and personalized treatments, increasing the likelihood of positive outcomes for patients.

Recent Research Studies: We have distributed samples and data to more than 90 studies, including the three below. For more detail, please go to https://biobank.partners.org/research-initiatives.

**Boston Mammography Cohort Study**, Rulla Tamimi, MSc, ScD, Department of Medicine at BWH and Erica Warner, MPH, ScD, Clinical and Translational Epidemiology Unit at MGH - The purpose of the Boston Mammography Cohort is to identify genetic and environmental predictors of breast density and breast cancer risk in an ethnically diverse cohort of women. The cohort consists of women routinely screened for breast cancer at Brigham and Women’s Hospital or Massachusetts General Hospital.

**Health Impact of Genetics on Type 2 Diabetes and Complications**, James Meigs, MD, Division of General Internal Medicine at MGH - Recent genetic studies have identified dozens of common variants associated with type 2 diabetes (T2D). Dr. Meigs is using Biobank samples to test hypotheses that genetic variants known to increase risk for T2D, or associated with measures of blood glycemia, also increase risk for T2D and risk of T2D complications. The study will evaluate translation of genetic discovery into clinical and public health application to improve care and outcomes for type 2 diabetes patients.

**Restoring the Immune System after Radiation Injuries**, James Lederer, PhD, Department of Surgery at BWH - Dr. James Lederer is researching potential therapies for people who would be hurt in a radionuclear event, such as a nuclear blast. There are limited treatment options for protecting the potentially high number of people who might be injured in case of such an event. Dr. Lederer is using samples from the Partners Biobank to investigate ways to restore immune system function after exposure to radiation or radiation combined with traumatic injury (such as a burn or infection).