Thank you for supporting the Partners Biobank. We are grateful.

Thanks to your participation and the participation of tens of thousands of patients at the Brigham and Women’s Hospital, Massachusetts General Hospital, and McLean Hospital, our Biobank continues to thrive and fulfill its research mission. We have now provided samples to more than 60 Partners research groups that are studying diabetes, arrhythmia, immune disorders and other diseases. These studies are now starting to publish discoveries that were made thanks to the Biobank and its thousands of participants. By participating in the Partners Biobank, you are helping us better understand, treat, and even prevent the diseases that might affect your health and the health of future generations.

In this issue:

- Learn about some of the research studies that have received samples and data from the Biobank in the past few months
- Read about a genetic discovery that may reduce heart disease in the future
- Hear from a member of our Community Advisory Panel (CAP)

A Letter from a Community Advisory Panel Member

A world in which all the pain and suffering of disease can be cured may be a pipe dream. Medicine and healthcare have made tremendous progress in mitigating the impacts of diseases and sometimes even in curing them; but there is a long way to go.

How do we close the gap? I believe the answer is in scientific research. Our best and brightest scientists can find these cures, but the cost of doing research remains a stumbling block. Among other impediments, research is subject to spending cutbacks, might be guided by commercial goals, and is apt to focus on outcomes that will benefit larger populations. Oftentimes, those investigating less “popular” diseases may find funding lacking or absent.

That’s one reason the Partners Biobank gives me so much hope. The investigation of human genomics offers so much promise. Within each of us are unique biological traits, genetic markers that research is finding can be predictors of disease. While tremendous progress has been made in understanding genetic markers as predictors of disease, there is so much more to explore.

By developing a genomic databank from which researchers can swiftly and easily select groups of people with the same condition, the Partners Biobank provides a lower-cost alternative for scientists doing research. And all of us can participate in helping the Biobank, and its resulting research studies, accelerate the process towards finding cures. By providing our genetic history through a simple blood draw, Biobank participants are active participants in helping researchers develop cures sooner.

I volunteered to become a member of the CAP for the Partners Biobank, because I believe in and am inspired by its mission. Meeting regularly since 2014, the CAP is a group of volunteers.

We meet with clinicians, scientists, and administrators of the Biobank to discuss its goals, practices, and procedures. By offering the people behind the Biobank our laypersons’ views, we hope to better communicate the benefits it can bring to our children and future generations. Everyone who joins the Biobank is making an investment in a better today and tomorrow for all.

Thank you,
Tom Fryer, CAP member
Dr. Kathiresan (above) and his team of investigators, led by Pradeep Natarajan, MD, and research coordinator Erina Kii in the Center for Human Genetic Research at Massachusetts General Hospital, are looking for a needle in a haystack.

Sekar Kathiresan, MD, uses the Partners Biobank to conduct research that would otherwise be impossible using traditional research methods.

The small but devoted team is trying to locate patients with one variant of a specific gene, APOC3. The abnormality of this gene only affects about 1 in 150 people.

Locating and studying these individuals could have tremendous impact on how heart disease is treated in the future. Through his lab’s research, Dr. Kathiresan found that people with this single dysfunctional APOC3 gene variant were naturally protected against heart disease and hopes to study this further by working with individuals with this variant.

Initial observations when looking at the APOC3 gene showed people with the variant also had a low level of triglycerides—a type of unhealthy fat that circulates in the blood; elevated levels have been shown to increase the risk of heart disease and heart attack. By studying patients who have naturally low triglyceride levels due to the APOC3 defect, the team hopes to develop an inhibitor that will mirror the effect in other patients as well.

This is the first time Drs. Kathiresan and Natarajan will be working with Biobank participants for a research study. When asked whether this research would be possible without the samples from the Partners Biobank, Dr. Natarajan definitively said “no.”

“It is possible to work with collaborators globally and have access to blood and potentially tissue samples. It is, however, the local Partners Biobank that gives our team access to actual patients based on specific genotypes,” said Dr. Natarajan.

“Having access to this local population is key to answering some of the questions about how this gene variation works to block or lower triglycerides in humans,” he said.

Recent Research Studies: We have distributed samples and data to more than 60 studies, including the four below. For more details, please go to https://biobank.partners.org/research-initiatives.

**Testing the Effect of Genetic Variation on the Response to a Mixed Meal Tolerance Test**, Geoffrey A. Walford, MD, and Jose C. Florez, MD, PhD, Diabetes Unit at MGH – Drs. Walford and Florez are examining how commonly occurring genetic risk factors for type 2 diabetes influence the response to a specially prepared breakfast, known as a mixed meal tolerance test.

**The Genetics of Psychiatric Disorders**, Jordan Smoller, MD, ScD, Department of Psychiatry at MGH – As a member of the Psychiatric Genomics Consortium, Dr. Smoller and his colleagues are using 1,000 DNA samples from the Biobank to investigate the genetics of complicated psychiatric disorders such as autism, bipolar disorder, and depression.

**The Role of Viruses and Antiviral Antibodies in Chronic Diseases**, Stephen J. Elledge, PhD, Division of Genetics at BWH – Using a large number of samples from the Biobank will help Dr. Elledge and his team uncover any subsets of patients whose diseases are virus-associated. Identifying the specific virus will provide insight into disease mechanism as well as inform development of diagnostics and therapeutics.

**The Role of Genes in Rheumatoid Arthritis**, I-Cheng Ho, MD, PhD, Division of Rheumatology, Immunology, and Allergy at BWH – Dr. Ho and his study team used Biobank patient samples to study how one gene (the PTPN22 gene) may contribute to the development of rheumatoid arthritis (RA).