

# Mass General Brigham Biobank



**150,000  
participants**



**1.4 million  
stored biosamples**



**250,000  
distributed  
biosamples**



**600 studies  
that received  
biosamples or data**

## 2024 retrospective for the Mass General Brigham Biobank

The Mass General Brigham (MGB) Biobank is celebrating two important milestones in 2024. The first is that the Biobank is now 15 years old! Over the past fifteen years, the Biobank has distributed biosamples and data to 600 research studies. These have resulted in more than 400 peer-reviewed academic articles that describe research findings on a wide array of diseases, from cancer to heart disease, autoimmune diseases, psychiatric disorders, and a lot more.

The second milestone is that there are now 150,000 people participating in the MGB Biobank! This is an important achievement because size and diversity matter in research. Researchers use the MGB Biobank to look for patterns that help understand what makes people sick or healthy. Their research is more reliable when they have a lot of biosamples and data to work with. It is more relevant to all of us when it reflects differences in our backgrounds, lifestyles, and environments.

The Biobank is continuing to grow in an effort to support the research community at Mass General Brigham Hospital. We are deeply thankful for your continued participation.

## Diabetes research that looks at DNA to uncover risks

Diabetes is a disease that causes high blood sugar levels in patients. More than 10% of adults live with some form of diabetes. Dr. Miriam Udler, who directs the MGH Diabetes Genetics clinic, treats patients with genetic forms of diabetes.

There are many forms of diabetes and different forms need different treatments. It can sometimes be challenging to tell what kind of diabetes a person has. Dr. Udler and her team have used the medical record and DNA information in the Biobank to help pinpoint the correct diagnosis for people. They have also created a system that uses medical records from the Biobank to find people with unusual forms of diabetes. Dr. Udler's team then compares Biobank biosamples of people with unusual and typical diabetes. They hope that in the future this will help improve diagnosing and treating patients with different forms of diabetes.

Dr. Udler's team has also done research on how environmental and social factors affect diabetes risk. To do this, they have compared Biobank DNA to information related to living environment in medical records. They found that education and income level can predict how likely someone is to have diabetes in the future.

Dr. Udler and her team have identified different genetic pathways that lead to type 2 diabetes. Type 2 is the most common form of diabetes. They have studied these genetic pathways in people with type 2 diabetes in the Biobank to better understand how they lead to different patient characteristics. They are working on using these pathways to identify new treatments for diabetes.

The kind of work that Dr. Udler does is not specific to diabetes. The same tools can be used to study other diseases. Dr. Udler has been working with other scientists to use her model for diseases like heart disease and high blood pressure. She hopes that this research will give patients more effective treatment and help prevent diseases before they happen.

## A new tool in medicine: Polygenic Risk Scores (PRS)

Dr. Pradeep Natarajan is the Director of Preventive Cardiology at MGH. He and his team use biosamples from the MGB Biobank to study heart disease, which is the leading cause of death worldwide. Many factors can cause heart disease. Some of these factors are environmental, like what you eat and how much you exercise. Other factors are genetic.

Biosamples from the Biobank consist of many cells. These cells contain genetic information, called DNA, which is inherited. DNA provides a code that instructs the cells in our bodies to do specific things. Genes are part of the DNA code that directs cells to perform certain functions. All people have similar DNA, but there are some differences in DNA, called variants, that make each person unique. Some variants can make people more likely to develop a health condition.

Some diseases are caused by changes to a single gene. Many other health conditions are caused by changes to many genes. For example, heart disease has been linked to at least fifty different variants. Dr. Natarajan's team works on tools to study conditions like heart disease that are caused by many variants.

One of these tools is a Polygenic Risk Score (PRS). A PRS is a number that helps doctors understand how likely their patients are to get a disease based on their genes. Not all DNA variants play an equal role in causing disease. One variant may put a person at a 5% risk of developing a condition. A different variant may only increase the risk by 1%. A PRS combines these risks into a single number. PRSs are a new way to evaluate someone's risk of getting a disease. Researchers are working to help bring them into clinical care. Dr. Natarajan's team has begun developing PRSs for heart disease.

Dr. Natarajan analyzes PRSs from many biosamples to identify who is at greatest genetic risk for developing heart disease compared to the rest of the population. It takes DNA from a lot of people to develop a PRS and the more biosamples that are used to make a PRS, the more accurate it is. As such, the Biobank has been a valuable tool for Dr. Natarajan's work. Accurate PRSs will help doctors know who is at the highest genetic risk for heart disease. They can then find patients who might benefit from lifestyle changes, treatments, and clinical trials. This work is important because it could allow doctors to intervene with high-risk patients before they ever feel symptoms of heart disease.

## Study highlights

### Studying the molecular underpinnings of post-operative delirium (POD)

Up to 50% of older patients have confusion after surgery. This confusion is called post-operative delirium. Right now, there are no treatments to lower these effects after surgery. Dr. Shiqian Shen, a physician researcher in Pain Medicine at MGH, looks at small molecule changes to learn how post-operative confusion works in the brain. Research with mice showed that giving a specific chemical substance lowered confused behavior. Dr. Shen uses Biobank blood biosamples to understand this relationship with humans and the use of specific chemicals in post-operative confusion.

### Researching the connection between rheumatoid arthritis and interstitial lung disease

Rheumatoid arthritis (RA) is one of the most common autoimmune disorders. RA causes inflammation in the joints, particularly in the hands and feet, and can affect other parts of the body as well. Up to 20% of patients with RA also develop a lung condition called interstitial lung disease (ILD). It is not well known why some people with RA develop ILD while others do not. Dr. Jeffrey Sparks and his team at BWH want to know if there are markers in the blood of people with RA who later develop ILD. This could help doctors predict who is most at risk of developing ILD. To do this, Dr. Sparks compares biosamples from Biobank participants with RA who developed ILD to biosamples from participants with RA who did not.

### Diagnosing gynecological cancers early

Ovarian cancer is a disease that can be hard to find early, making it tougher for women to survive. Dr. Kevin Elias is a gynecologic oncologist at BWH who is working on a blood test to tell whether someone has a high risk of getting ovarian cancer. He is studying differences in biosamples from women who later got ovarian, breast, or uterine cancer and women who did not get these cancers. Dr. Elias wants to use this test in the future to find women who might be more likely to get gynecological cancers. This could help decrease their risk of getting cancer or even stop it from happening.