ISSUE 1 SPRING/SUMMER 2013

MitoNews

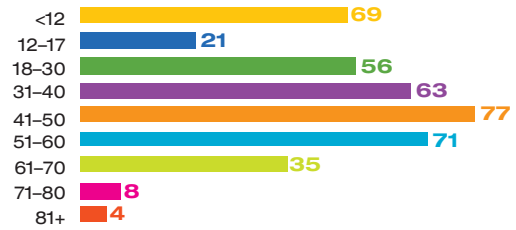
Welcome to the first issue of the Mitochondrial Disease Biobank newsletter. The Mitochondrial Disease Biobank at Mayo Clinic is the first research repository focused solely on mitochondrial disease in the United States. Our mission is to collect blood and tissue samples from patients with known or suspected mitochondrial diseases and their family members to aid the advancement of mitochondrial medical research and medicine. We enrolled our first participant in December 2009, and currently have over 2,800 specimens from 290 individuals. In this newsletter you will find information about our academic and conference activities, including studies that are using materials from the Biobank, and instructions for mitochondrial disease researchers. We hope this information will interest you. Thanks for your commitment to mitochondrial disease research.

Best wishes,
Mitochondrial Disease Biobank team

While Mayo Clinic is located in the Upper Midwest of the United States, our participants are located throughout the world. In particular, the Northeast US has been exceptionally generous to us. We are grateful to everyone who has joined our Biobank. Two-thirds of our participants are female. We would appreciate more male volunteers who would be willing to donate biospecimens.

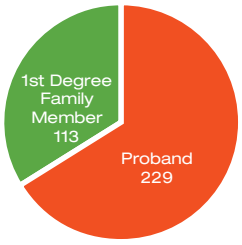
AGE

Our participants span all ages. In the following bar graph, we are pleased to see that our donors range from infants to those over 80 years of age. Researchers interested in using specimens from adults or children will have the opportunity to find what they might need.

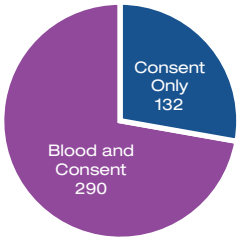


PARTICIPANTS

We are proud to announce that over 200 participants with mitochondrial disease have generously donated their biological specimens for research. In addition, over 140 family members have also demonstrated their commitment to mitochondrial disease research by donating their specimens as controls for their family members.



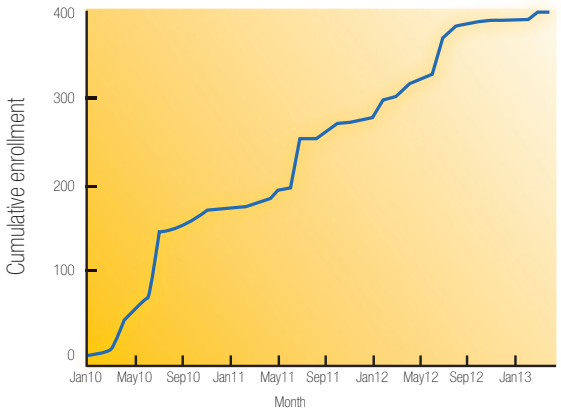
AVAILABLE SAMPLES



There are two steps required to enroll in the Biobank. You must complete and return all the Biobank enrollment paperwork and then provide a blood sample. Currently, over 130 people have not finished the second step, which means we are not able to use their information for research. *See page 2 for more information.*

ENROLLMENT

Enrollment into the Mayo Clinic Mitochondrial Disease Biobank has steadily increased since our inception in 2010. Annual patient advocacy group meetings play an important role in our enrollment activity.



frequently asked **questions**

What is the Mitochondrial Disease Biobank?

The Mitochondrial Disease Biobank is a research program at Mayo Clinic in Rochester, Minnesota, that aims to collect research samples from individuals with mitochondrial disease to improve the state of mitochondrial disease research.

Who can enroll in the Mitochondrial Disease Biobank?

Anyone with a confirmed or suspected diagnosis of mitochondrial disease is invited to participate. Additionally, first-degree relatives (parents, siblings, and children) of individuals with mitochondrial disease who have joined the Biobank may also enroll.

How much does enrollment cost?

Enrollment is free. You will not have to pay for any tests or procedures performed solely for the Mitochondrial Disease Biobank. However, tests ordered by your physician as part of your routine clinical care will not be covered by the Biobank.

Do I have to travel to Mayo Clinic to enroll?

Participants do not have to come to Mayo Clinic to enroll. The Mitochondrial Disease Biobank will send participants kits to have their blood drawn at their local physician's office or at any Quest Diagnostics location. These kits are sent to participants after they have consented to be involved in the Mitochondrial Disease Biobank.

What does the Mitochondrial Disease Biobank do with my information?

Biobank staff will review your medical information. Information about an individual's mitochondrial disease is stored in a secure database and used to determine eligible participants for specific research studies. When specimens are shared with other researchers, we may provide relevant information about a participant's clinical status. It is important to note, that when we send clinical information about a specimen, we never disclose personal identifiers (i.e., name, date of birth, address).

I think I may have enrolled in this study previously. How can I check to make sure I am enrolled?

The easiest way to check if you are enrolled in the Mitochondrial Disease Biobank is to contact us. Biobank staff is available by phone (507-293-1386 or 1-877-594-2149 toll-free) or email (mitochondrialdb@mayo.edu).

Are You Completely Enrolled in the Biobank?

If you are receiving this newsletter in the mail, you expressed interest in the Mitochondrial Disease Biobank. It does not mean you are enrolled. To be enrolled, you must complete and return all the paperwork we sent you and provide a blood sample. You can give the blood sample at Mayo Clinic, or at the annual UMDF Symposium, or we can send you a kit to take to your local physician's office or any location of Quest Diagnostics nationwide. Many people have requested information but have yet to complete the entire enrollment process for inclusion in the Biobank.

If you are unsure whether you have completed enrollment, or you need a new set of enrollment forms, please call us toll-free at 1-877-594-2149.

You can also download forms from our website: http://mayoresearch.mayo.edu/mitochondrial-disease-biobank/enrollment.cfm.

UNITED MITOCHONDRIAL DISEASE FOUNDATION – MITOCHONDRIAL MEDICINE 2012

The United Mitochondrial Disease Foundation (UMDF) is an organization that promotes research and education pertaining to the treatment and diagnosis of mitochondrial disorders. Families, professionals and researchers are encouraged to join UMDF where they can network, share information and learn about ongoing initiatives surrounding mitochondrial disease. UMDF holds an annual symposium with sessions for both families and professionals.

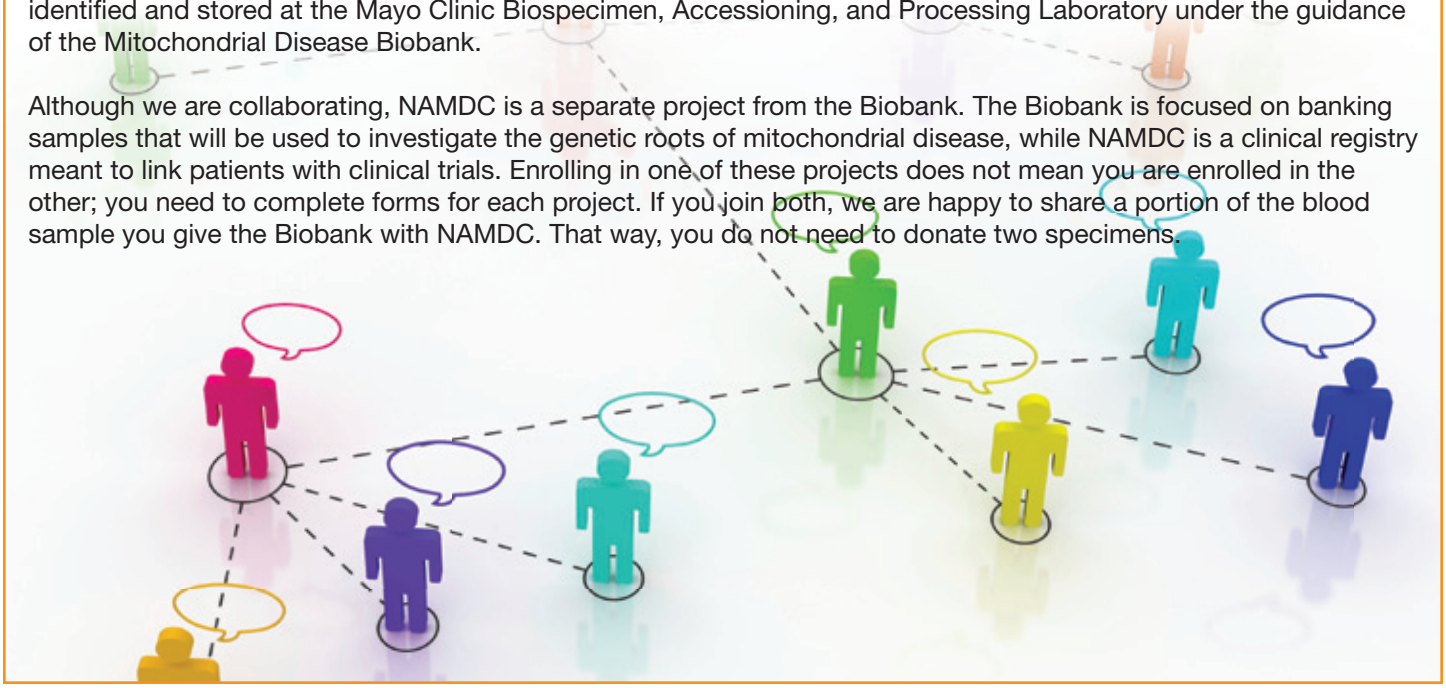
For the third year in a row, the Mayo Clinic Mitochondrial Disease Biobank team attended the UMDF annual symposium, which was held this year in Bethesda, Maryland, from June 13 to 16, 2012. We were also pleased to renew our sponsorship of the symposium this year, as well as the talk by David Thorburn, Ph.D., called "Genetics and Next Generation Sequencing - What does it all mean to Mitochondrial Disease Patients and Families?" We staffed a table in the exhibition area full of information and recruitment materials. Throughout the symposium, numerous families, physicians and researchers stopped by to learn more about the Mitochondrial Disease Biobank. We offered full Biobank enrollment on-site, including a private area where participants could have their blood drawn. We recruited 45 new participants, and two current members gave additional blood samples.

We will have a table and blood draw station at the 2013 UMDF Symposium in Newport Beach, California, June 12–15, 2013. We will be enrolling new members, and if you are already a participant of the Mitochondrial Disease Biobank, please consider giving an additional blood donation!

Collaboration with the North American Mitochondrial Disease Foundation

In 2012, the Mitochondrial Disease Biobank and the North American Mitochondrial Disease Consortium (NAMDC, http://rarediseasesnetwork.epi.usf.edu/namdc/) officially began a collaborative project. With the hope of improving national infrastructure for advancing mitochondrial disease research, the Biobank approved a process to store NAMDC participant specimens at Mayo Clinic. NAMDC is a federally funded mitochondrial disease patient registry. This collaboration takes advantage of the Biobank's experience with biobanking mitochondrial disease-related specimens and opens additional academic sites to the possibility of providing specimens for NAMDC's patient registry program. NAMDC aims to collect new donations from 100 of its participants. These specimens will be de-identified and stored at the Mayo Clinic Biospecimen, Accessioning, and Processing Laboratory under the guidance of the Mitochondrial Disease Biobank.

Although we are collaborating, NAMDC is a separate project from the Biobank. The Biobank is focused on banking samples that will be used to investigate the genetic roots of mitochondrial disease, while NAMDC is a clinical registry meant to link patients with clinical trials. Enrolling in one of these projects does not mean you are enrolled in the other; you need to complete forms for each project. If you join both, we are happy to share a portion of the blood sample you give the Biobank with NAMDC. That way, you do not need to donate two specimens.



Find Us on Facebook

Our Facebook page has been active since April 2011, and in November 2012, we received our 1,000th "Like." In October 2012, we gave a presentation about our use of Facebook at the American Society for Bioethics and Humanities Annual Meeting in Washington, D.C. We have friends in 39 countries so far. Have you joined us on Facebook yet? Here is some of what you will see posted on our page:

- Recent journal articles about mitochondrial disease and biobanking
- Real-time status updates about research using samples in the Mitochondrial Disease Biobank
- Opportunities to support the mitochondrial disease community
- Information about participating in clinical trials for new mitochondrial disease treatments
- Biobank activities beyond collecting samples for research (e.g., publications, presentations, conference participation)

https://www.facebook.com/MitochondrialDiseaseBiobank

MitoAction Walk Sponsorship

For the third year in a row, we sponsored the MitoAction Energy Walk and 5K. This event took place in Boston on Sept. 16, 2012. Over 700 individuals participated, **raising $143,282.69.**

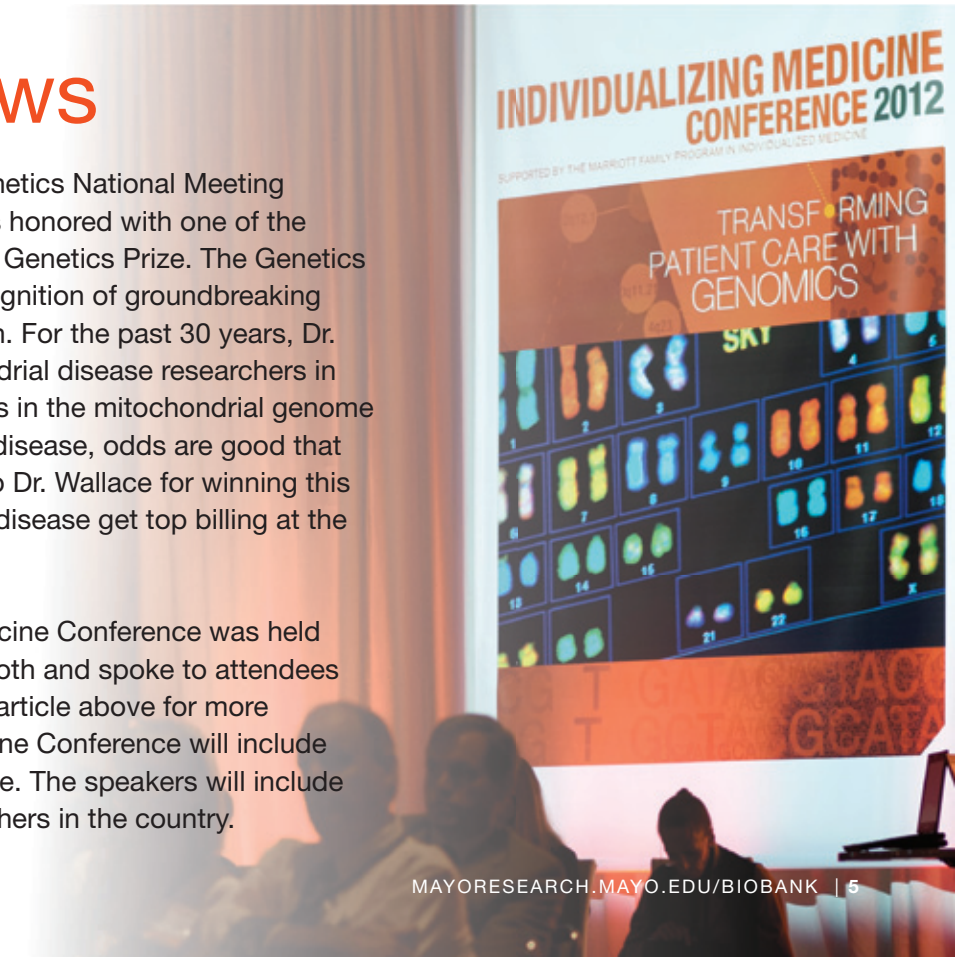
Informed Consent Innovation

Federal law requires that individuals give informed consent before they can enroll in a research project like the Mitochondrial Disease Biobank. Biobanking is a new type of research infrastructure, so many people are not familiar with what biobanking entails. According to federal regulations, for individuals to be able to enroll in the Biobank, they must understand the risks, benefits, and alternatives to Biobank participation. We have been working with a product designer and illustrator to produce graphic versions of our consent form for adults, and an agreement sheet (called assent) for teens in the hope that the combination of illustrations and text will make it even easier to understand the details of Biobank enrollment.

Conference News

At the recent American Society for Human Genetics National Meeting in San Francisco, Douglas Wallace, Ph.D., was honored with one of the Society's most prestigious awards, the Gruber Genetics Prize. The Genetics Prize is presented to a leading scientist in recognition of groundbreaking contributions to any realm of genetics research. For the past 30 years, Dr. Wallace has been one of the leading mitochondrial disease researchers in the world. He was the first to identify mutations in the mitochondrial genome that cause disease. If you have mitochondrial disease, odds are good that Dr. Wallace first identified it. Congratulations to Dr. Wallace for winning this prize! It is truly gratifying to see mitochondrial disease get top billing at the largest genetics meeting in the country.

The inaugural Mayo Clinic Individualizing Medicine Conference was held in October 2012 in Rochester. We staffed a booth and spoke to attendees about our new illustrated consent forms. (See article above for more information). Next year's Individualizing Medicine Conference will include a session will focused on mitochondrial disease. The speakers will include a few of the top mitochondrial disease researchers in the country.



Updates for Researchers:

The BAP Lab – Home of the Mitochondrial Disease Biobank

After a sample is collected for the Mitochondrial Disease Biobank, where does it go, and who is responsible for processing and storing it? The answer is the Biospecimen Accessioning and Processing Laboratory, also known as the BAP Lab.

The BAP Lab was created to meet the evolving needs of the Mayo Genomics Facility (MGF), the Center for Individualized Medicine, Mayo Clinic Cancer Center, and other Mayo researchers for specimen acquisition and processing. The roots of the BAP Lab lie in a clinical laboratory, the Molecular Genetics Laboratory (MGL) in the Division of Laboratory Genetics, Department of Laboratory Medicine and Pathology (DLMP). In 2010, the BAP Lab merged with the DLMP Special Studies unit, which processed serum and plasma for routing into the clinical laboratories and a research unit, the South Warehouse sample shipping and storage team. Thus, the BAP Lab now offers a complete range of services to the Mayo research community — sample accessioning, processing, DNA extraction, and storage at -80 degrees Celsius in a state-of-the-art 600,000-tube, robotically controlled freezer.

Over the past 12 months, the BAP Lab has received and accessioned over 56,000 samples, extracted DNA from 26,000, and made and stored over 215,000 tubes of plasma or cells from blood samples. At this time, the BAP Lab employs 50 individuals to process the samples that are collected for research at Mayo Clinic, including those collected for the Mitochondrial Disease Biobank.

Further Your Research by Utilizing Samples from the Mitochondrial Disease Biobank

Samples are available to mitochondrial disease researchers nationwide. You must submit a research proposal, which will be reviewed by our executive committee. If it is approved, we will share samples with you free of charge. Please contact us for additional information or a blank copy of our sample request documents.

Who used the Mitochondrial Disease Biobank in 2012?

We provided letters of support to the following projects, indicating that we are willing to give researchers access to stored specimens when their projects are approved:

Wolfdieter Springer, Ph.D. Title: Targeting Mitochondria Quality Control for Individualized Healthcare Solutions. Department of Neurology and Neuroscience, Mayo Clinic, Jacksonville, Florida.

> *Aim: To uncover the role that Parkin and PINK1 play in mitochondrial autophagy during health and disease.*

Stephen Ekker, Ph.D. Title: A TALEN-based Therapeutic Strategy for Treating Mitochondrial DNA Genetic Disorders. Department of Biochemistry and Molecular Biology, Mayo Clinic, Rochester, Minnesota.

> *Aim: To investigate whether genetic engineering can help repair large mitochondrial DNA deletions, such as those found in Kearns-Sayre syndrome (KSS).*

Studies that received approval from our Executive Committee to use Biobank specimens include:

Amir Lerman, M.D. Title: Humanin in Mitochondrial Disease. Division of Cardiovascular Diseases, Mayo Clinic, Rochester, Minnesota.

> *Aim: To assess the relationship between humanin protein levels and mitochondrial dysfunction.*

Norma Frizzell, Ph.D. Title: Protein Succinylation in Mitochondrial Disease. Department of Pharmacology, Physiology, and Neuroscience, University of South Carolina School of Medicine, Columbia, S.C.

> *Aim: To uncover novel protein post-translational modifications in Leigh syndrome and Leber's hereditary optic neuropathy.*

W.E. Highsmith, Ph.D. Title: Sequencing of the Mitochondrial Genome and Selected Nuclear-Encoded Mitochondrial Genes in Individuals from the Mayo Clinic Bipolar Disease, Mitochondrial Disease, and Vascular Disease Biobanks and Controls. Department of Laboratory Medicine and Pathology, Mayo Clinic, Rochester, Minnesota.

> *Aim: To discover genetic causes of mitochondrial, bipolar and cardiovascular diseases.*

Academic Activities

The main aim of the Mitochondrial Disease Biobank is to gather samples and medical records from people with mitochondrial disease or relatives of those with mitochondrial disease, but we also keep active in the research community by publishing articles and giving conference presentations about our work. Here is a sample of our 2012 activities:

Publication. LR Eisenberg and JR Anderson, Picture This: Illustrating the Future of HIPAA Documents, Atrium: The Report of the Northwestern Medical Humanities & Bioethics Program, Issue 10, Spring 2012.
Read it online at http://bioethics.northwestern.edu/atrium/pdf/atrium-issue10.pdf.

Presentation, American Society for Bioethics and Humanities Annual Meeting. Washington, D.C., Oct. 20, 2012.
"The tangled Web: The rare disease biobank and the internet age."
LR Eisenberg, CM Anderson, JL Hesemann, WE Highsmith, and D Oglesbee.

Presentation, Comics & Medicine: Navigating the Margins. Toronto. July 22–24, 2012. "A form of comic relief: HIPAA in graphic detail." LR Eisenberg and JR Anderson.

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CONTACT US

If you have questions or need information about the Mayo Clinic Biobank, please contact us at:

507-293-0203 (local)

1-866-613-2386 (toll free)

biobank@mayo.edu (e-mail)

mayoresearch.mayo.edu/biobank

Helping Support Groups

Are you a part of a mitochondrial disease support group? We would love to help members of your group enroll in the Mitochondrial Disease Biobank by sending you a box of enrollment materials to distribute at your next meeting. If this interests you, please contact us at 507-293-1386 or 1-877-594-2149 (toll-free) or email mitochondrialdb@mayo.edu.

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