HEART CENTRE BIOBANK REGISTRY

Participant Newsletter

ISSUE 06 · APRIL 2019

What's Trending?

Greetings from the Heart Centre Biobank Registry! You are receiving this newsletter because you are a Heart Centre Biobank participant and as always, we are grateful for your contribution to our registry and would like to update you on what your participation has helped us achieve in the past year! Here is an update on what is new and trending in our registry:

Our enrollment is trending up as we continue to recruit new and retain existing participants. Our patient population grows on average by **8%** each year.

9497

Data use is trending up as we have supported **50** new research studies within the past 3 years with the specimens and data that you have donated to the biobank.

Family participation is trending up! **370%** increase within this past year.

We recognize the importance of studying not just the child with disease but parents to help us determine if the finding is new or passed down. If you are a parent who has not yet participated and would like to, please contact us at heartcentre.biobank@sickkids.ca or 1-866-489-7711. Participation can be coordinated by mail.

Our Findings Have Reached the Clinic!

Researchers are motivated by the prospect of having their science jump from bench to bedside where discoveries lead to making a tangible difference in improving the care and quality of life of patients and their families. Here are some examples of how some work through the registry has made an impact clinically.

New clinical test offered for a gene we helped to identify

Through an international collaborative study involving biobank participants with atrioventricular septal defect (AVSD), we were able to identify a new gene, *NR2F2*, as the cause of the disease in several children. Findings from this study have led to this gene being offered as part of clinical testing. *Turki SA, Manickaraj AK, Mercer CL, Gerety S, et al. Rare variants in NR2F2 cause congenital heart defects in humans. Am J Hum Genet. 2014 Apr 3;94(4):574-8*

Clinically significant research findings returned to participants

Researchers using biobank samples may find something that could be important to you or your family's health. We believe it is important for families to learn about these findings in real time if there is a chance that it can help them now rather than years later. To date, 20 new genetic findings discovered through research have been communicated to patients so that they have an option to confirm these findings in a clinical lab and receive medical care accordingly. We are available to answer any questions you may have about new findings that you receive.

The Cardiac Precision Medicine Program

Precision Medicine is a new approach that individualizes medical care by taking into account what is unique about each person based on their genes, environment, and lifestyle. Read on to see how researchers are using precision medicine for better ways to manage their patients.

Early diagnosis

Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) is a genetic cardiomyopathy that causes heart muscle to be replaced with fiber and fat. The condition can be life threatening. The genetic cause remains unknown in 2/3rds of families. A research team led by SickKids discovered a new way to diagnose this heart condition. Read more at: **https://tedrogersresearch.ca/2018/11/at-sickkids-astounding-finding-may-lead-to-new-therapeutic-target-for-life-threatening-heart-condition/**

Preventing sudden death in cardiomyopathy

The sudden loss of a child or young adult is a devastating burden on a family. Patients with hypertrophic cardiomyopathy (HCM) are at risk for sudden death. Sudden death can be averted with the

continued on back page

Heart Centre Biobank

Thank you for participating and for your continued support!

For the most up-to-date news, check us out at www.heartcentrebiobank.ca

Phone: at 1-866-489-7711 Email: heartcentre.biobank@sickkids.ca

TED ROGERS CENTRE FOR HEART RESEARCH

If you are a parent that isn't already participating or you have parents that would like to, please call or email the Biobank team for information.

Let's build a closer working relationship – Participant Engagement Network

We believe that participants and families can play an insurmountable role in driving research forward. We are currently exploring ways to engage with individuals as partners to drive research forward. Please email us at heartcentre.biobank@sickkids.ca if you are interested in learning more about biobank engagement or a particular project. Parents and high school aged children are encouraged to inquire. Volunteer service hours are available for students.

Tell us more about our communication efforts!

Do you hear enough from us, often enough, and through the channels you prefer? We'd love to hear more from you so that we can improve on how best to communicate with you. Included with this newsletter is a short survey on communication and engagement, please tell us your thoughts at **https://redcapexternal.research.sickkids.ca/surveys/?s=MJ84ALR4JY**

Let's get Social!

Would you like to start receiving updates via email, receive our newsletters, and stay connected with important information and new discoveries? Tell us more on what you want! Email us at: heartcentre.biobank@sickkids.ca

The Cardiac Precision Medicine Program *continued from front page*

insertion of a device called an implantable cardioverter defibrillator (ICD). At this time, we cannot accurately predict who is at high risk for sudden death. This results in failure to provide an ICD in a timely manner. Researchers are developing a prediction tool that will use clinical and genetic information to determine which child is at high risk and needs an ICD. In the future, this will allow your cardiologist to make precise risk calculations for your child while in clinic and offer timely recommendations, preventing an adverse series of events that no family should have to endure.

Targeted therapies for cardiomyopathy

Cardiomyopathy is the most common cause of heart failure and sudden death in children. There are no long-term cures for cardiomyopathy. The biobank is supporting studies to find targeted therapies for cardiomyopathy. Through a process called reprogramming, we are converting a patient's skin/blood cells into stem cells and using these to generate heart muscle cells from patients with cardiomyopathy. We are using revolutionary new technology called Crispr gene editing to fix these defects in the patient cells and change them into healthy cells. We are also testing drugs that act directly on the defective protein in the heart muscle cell to see if they can help the cells beat normally. We expect that these targeted drugs will provide a better chance at recovering the heart in cardiomyopathy so that we don't lose children to this disease. Read more at: **https://tedrogersresearch.ca/2018/02/unveiling-first-million-dollar-visionary-projects/**

The More we Know, the More we can Do

Breaking new ground

The human genome has become a critical piece in the search for what causes human disease. The human genome contains over 4 million little changes, some of which are the cause of disease. We can now generate the sequence of an entire human genome to try and find that one little defect that is causing the disease. But, we need to look not just within genes but also within regions outside of genes that control how a gene functions.

Through funding from Ted Rogers Centre, we have launched large scale whole genome sequencing (WGS) of children with cardiomyopathy and congenital heart disease. We have many expert data scientists working on this data as it is generated to find what causes heart disease in children. This is uncharted territory but we are making significant breakthroughs. Using WGS, we were able to identify the likely cause of cardiomyopathy in almost twice as many children. As we do studies to confirm these findings, this information will become useful to help family members be tested for these gene defects. This will help physicians inform which family members have risk of developing cardiomyopathy, and offer treatment in a timely manner.

Expanding our reach: Personalized Genomics for Congenital Heart Disease (PROCEED)

This newly launched study with European partners are combining biobank populations of patients with tetralogy of Fallot and transposition of the great arteries from Canada, Germany and the Netherlands with the goal of developing tools to allow rapid analysis of whole genomes in these populations to find the underlying genetic cause. By collaborating, we will be able to find answers faster. We will be able to develop new and more accurate genetic tests. We will be able to counsel families about the severity of the disease based on the type of gene defect, and what to expect after a child is born. An informed patient is an empowered patient who can participate in shared decision making with the medical team for what is right for themselves and their families. Read More at: **https://tedrogersresearch.ca/**

A Message from the Heart Centre Biobank

The discoveries made through the research highlighted in this newsletter would not have been possible without your participation. Your original contribution through your participation in the Heart Centre Biobank Registry is a gift that keeps on giving as your sample(s) and data can be used to support multiple research studies. The Heart Centre Biobank is thankful to all its participants for their contribution to these discoveries.

Heart Centre Biobank Team: Phone: at 1-866-489-7711 Email: heartcentre.biobank@sickkids.ca Twitter: @hcbiobank