



BIG News

Biorepository and Integrative Genomics (BIG) Initiative Newsletter

Thank you again for your participation in the BIG Initiative!

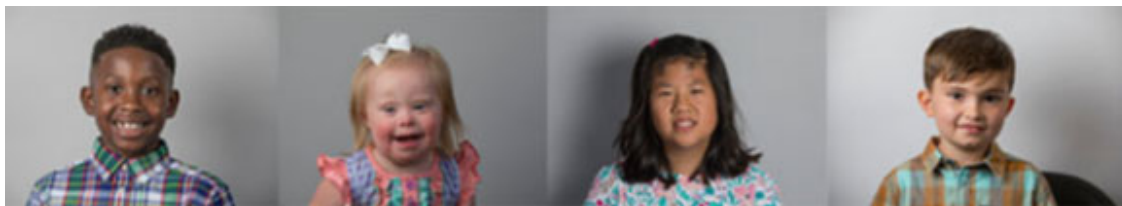
As you know, the BIG Initiative is a collaboration among Le Bonheur Children's Hospital, the Children's Foundation Research Institute and the University of Tennessee Health Science Center (UTHSC) to help doctors and scientists study the influence of genes, or the genetic "blueprints" in each of the body's cells, on what diseases we get, how well we respond to treatment and to also help develop new treatments and better tests for what ails us.

When you enrolled in this project, you shared your email address with us, and we promised to keep you informed of our progress. We are excited to share these updates with you about the initiative.

Current Enrollment

Since our start in November 2015, more than 42,000 participants have joined the BIG Initiative, and BIG has collected DNA from more than 15,000 participants! These numbers are huge milestones, and we want to thank each and every one of you for such an outstanding level of participation and support. As a result, we now have a resource that truly reflects the breadth and diversity

of kids in our community. As BIG continues to expand its enrollment efforts, this resource continues to grow each day.



Some have asked why we haven't collected DNA from every participant. You may remember that BIG never asks for extra blood samples from participants and instead uses whatever blood is leftover in blood draws taken for clinical tests ordered by your doctor. Not everyone has a suitable blood draw for collecting DNA when they enroll in BIG, and sometimes not enough blood remains even when they do. So, this means that BIG doesn't always get a sample from every participant when they enroll.

Nevertheless, we prefer doing it this way so that our participants and their families are not subjected to any additional stress or discomfort.

In our previous newsletter, we described how BIG had expanded enrollment areas around Le Bonheur beyond inpatient hospital floors adding the Neonatal Intensive Care Unit, the Emergency Department and various locations in the Outpatient Center. We are excited to announce that BIG has since expanded its efforts again.

Since May 2022, we have included Regional One Health Medical Center as a site for recruitment. Now, many individuals who receive care at Regional One will have the opportunity to participate in the BIG Initiative. Kito Lord, MD, physician lead of BIG at Regional One Health, has been encouraged by the success. "We are absolutely thrilled to have enrolled more than 5,600 patients at Regional One Health. Each volunteer represents an opportunity to learn how genes impact medical conditions. We believe the future of health care treatments will focus heavily on precision medicine and are excited to extend the BIG program to our patients."



Current Studies Partnering with BIG Initiative

Now that we have begun to receive data from the acquired samples, we have been able to share data with approved researchers. The scientists who wish to have access to this data must submit detailed proposals to the BIG Research Oversight Committee and the UTHSC Institutional Review Board for review and final approval. Though we anticipate a varied and extensive list of research topics, currently, BIG has proposals and studies related to:

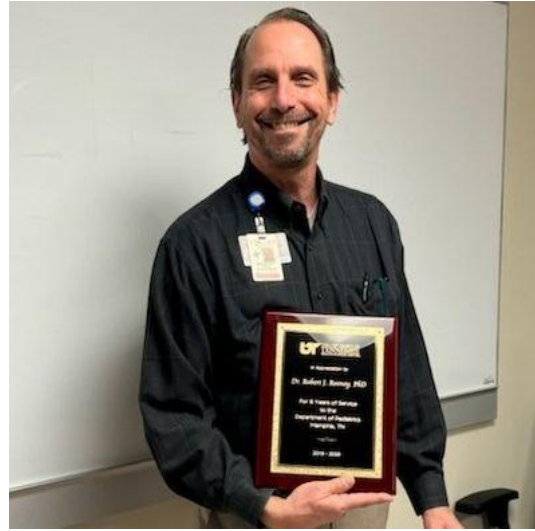
- Kidney disease
- Sickle cell disease
- Pediatric interstitial lung disease*
- Ear, nose and throat (ENT)
- Rare genetic diseases
- Seizure and epilepsy
- Diseases of the GI tract
- Asthma
- Disorders of the urinary tract
- High blood pressure
- Susceptibility to HIV and other infectious diseases
- Obesity

*A project soon to be funded by the National Institutes of Health

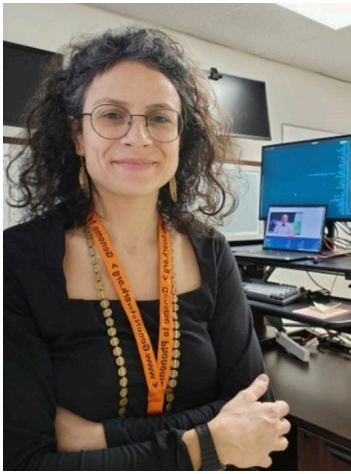
Departmental and Staff Updates

In 2015, Robert Rooney, PhD, joined the BIG Initiative as the biorepository lab director. Rooney skillfully guided the inception and growth of the BIG Initiative and has been invaluable in its success. He designed the biorepository from the ground up and was instrumental in creating and instituting BIG Initiative operational infrastructure and policies that governed many BIG Initiative activities. Chester Brown, MD, PhD, chief of Genetics at Le Bonheur, states, "Dr. Rooney was one of the founders of the BIG initiative and made extraordinary contributions to the effort. Without his expertise and advice over

the years, BIG would not have reached the point that it has today. Our patients and families stand to benefit for many years to come as a result of his efforts.” Thank you, Dr. Rooney for your many years of service. We wish him all the best in his retirement!



The Biorepository lab will now be led by Vicenza Colonna, PhD. Colonna is a faculty member in the departments of Genetics, Genomics & Informatics and Pediatrics, and is internationally known in the fields of population genomics and pangenomics. Welcome Dr. Colonna!



"I am thrilled to be a part of the BIG Initiative, a groundbreaking project that promises to make genomics more inclusive. I look forward to contributing to this dynamic team, leveraging state-of-the-art pangenomic techniques to analyze data in ways never before possible. Together, we are at the edge of transforming our understanding of genetic diversity and its impact on human health." – Enza Colonna, PhD

We would also like to welcome Terri Finkel, MD, PhD, to the BIG team. "Terri Finkel, MD, PhD, interim chair of the department of Pediatrics, has recently joined the BIG advisory board. Dr. Finkel is also chief of Rheumatology and is deeply involved in genetics research, including projects with the Genomics Information Commons. She led the statewide effort to successfully pass legislation that will make large-scale, rapid genetic testing (whole genome sequencing) accessible to all hospitalized, critically ill children." – Chester Brown, MD, PhD, principal investigator of the BIG Initiative



Congratulations are in order for Chester Brown, MD, PhD, and Sandra Arnold, MD. They have provided excellent leadership for the BIG Initiative, and we are proud to announce their recent accolades.



Le Bonheur Chief of Infectious Diseases Sandra Arnold, MD, was named one of the Health Care Heroes by the *Memphis Business Journal*. This award salutes companies, individuals and organizations for their contributions to improving health care in Memphis and the Mid-South. Arnold is a professor at UTHSC and medical co-director of Antimicrobial Stewardship at Le Bonheur. Though Arnold is ending her direct involvement with BIG, her years of leadership and guidance of the consent team have been invaluable and much appreciated!

Chester Brown, MD, PhD, Genetics division chief at Le Bonheur Children's Hospital and professor of Pediatrics and Genetics, Genomics and Informatics at UTHSC, was also listed as a Health Care Hero. Additionally, he was listed among "1,000 Inspiring Black Scientists in America," by the journal, *Cell Mentor*. In addition to serving as the principal investigator for the BIG Initiative, Brown is also the St. Jude Chair of Excellence in Genetics at UTHSC.



For more information about BIG, please see the **BIG Consent videos** below:

[Le Bonheur Participants](#)

[Regional One Health Participants](#)

And learn more through our website on the **FAQ page** for BIG participants and families.

[Le Bonheur Participants](#)

[Regional One Health Participants](#)

