CARDIOLOGY 2024

Data Acquisition: Challenges and Opportunities from the Frontline in Development of a Birth Defects Biorepository

Stacy Woyciechowski, MS, LCGC February 15, 2024



IN THE BEGINNING...

CHOP Annual Report, 1914

"There is no branch of the modern scientific care of sick children that has not been carefully considered in the formulation of the future plans, and indeed it may here be suggested that this institution might well consider the possibility of a maternity department as a necessary part of the hospital. Among the class from which we draw our patients prenatal work is of vital importance, and so closely allied with pediatrics that the line of demarcation between them is exceedingly narrow."

- Edward S. Sayers, President, Board of Managers, 1914





WHAT IS A BIOREPOSITORY?

- A centralized place to collect and store biological samples to be used for future research studies
- A way to obtain a complete dataset of de-identified clinical and research data linked to each enrolled subject and specimen
- An infrastructure built to remove barriers to research and simplify the process
- A way to provide re-useable data



AIMS OF THE BIRTH DEFECTS BIOREPOSITORY

- The goal of the BDB is to improve the health and well-being of all children with birth defects by creation of a **sustainable resource** to support:
 - investigations into the *etiologies* of birth defects
 - the advancement of personalized medicine with development and implementation of <u>tailored treatments</u>, and
 - the understanding of <u>long-term outcomes</u> for children with birth defects



NEED THE DATA TO ACHIEVE THE AIMS

- A comprehensive dataset consisting of data elements obtained from <u>existing clinical and research datasets</u> and core analyses of samples
- A <u>biorepository of samples</u> available for non-human subjects research and to serve as a resource for future access through IRB-approved studies
- A system that will <u>permit linkage</u> of biorepository samples to longitudinal clinical data, including prenatal data, for future studies
- A scalable infrastructure to **combine and enhance biorepositories** at CHOP





ENROLLMENT



Updated 2.13.2024





SPECIMEN COLLECTION





SEQUENCING





SOCIAL DETERMINANTS OF HEALTH







HOW MANY PATIENTS AT CHOP HAVE HEART DISEASE?



PHENOTYPIC CHARACTERIZATION

- Hierarchy of data sources and comprehensive phenotyping
 - Curated clinical registries
 - Reconciled diagnoses with expert input
 - Manually abstracted genetics physical examinations
 - Electronic health record (EPIC)
 - Problem lists
 - Billing

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DATA STANDARDIZATION FOR INVESTIGATORS

- Standardized ontology
 - HPO (Human Phenotype Ontology)
 - CUI (Concept Identifier)
 - SNOMED (Systematized Nomenclature of Medicine Clinical Terms)
- Provide the data sources
- Uniform data packages

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CHOP ENTERPRISE SYSTEMS

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Roberts Individualized Medical Genetics Center (RIMGC)

Birth Defects Biorepository

CC/CVI Biorepository

Investigators

Common

Infrastructure

-IRB language for sharing
-Standard Operating Procedures
-Project-specific Team of coordinator(s), abstractionist(s), analyst(s)
-Enrollment Tracking/Billing in OnCore
- REDCap data entry and data dictionary Sample and relationship linking in the BRP
-Samples stored in BioRC with common

labeling system

Neuroscience Biorepository

Additional Biorepositories

STREAMLINED PROCESS FOR COLLABORATIVE BIOBANKING

- Increased **sample size** (multiple ways in)
- Increased **specimen type** (for overlapping subjects)
- Enhanced capability for **sharing and future use**





COLLABORATIVE DASHBOARD

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WHERE WE ARE AND WHERE WE ARE GOING

- Created a Birth Defects Biorepository with a scalable infrastructure
- Enrolled over 1200 probands and over 1000 complete trios
- Sequenced over 2500 individuals
- Approved 25 projects
- Supported several grants



WHERE WE ARE AND WHERE WE ARE GOING

- Built collaborative relationships across CHOP, including with the newly initiated Institutional Biobank
- Increased enrollment across Cardiac Center together with Laura Mercer-Rosa and Ryan Callahan, with support from the Cardiovascular Institute and the Cardiac Center Executive Committee to collect tissue in the OR
- Highlighted what is possible with this kind of resource
- Looking forward to future growth

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Data Team

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Study Coordinators

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Translational Informatics (TRIG)

Monica Molina Alex Gonzalez Edward Krause

Genetic Counselors

Erica Schindewolf Natalie Burrill Lisa Pilchman Renee Wright Haley Crane

Steering Committee

Holly Hedrick Natalie Rintoul Ingo Helbig Jack Rychik Rebecca Linn Michael Padula Rebecca Ahrens-Nicklas Monique Gardner Laura Mercer-Rosa Akira Nishisaki Danielle Traynor Carrie Coleman-Campbell **David Stokes Gregory Tasian** Gregory Heuer Stephen Zderic

Juliana Gebb Tom Reynolds Jesse Taylor

Specialty Departments

Special Delivery Unit

Center for Fetal Diagnosis and Treatment

Pathology

Biorepository Resource Center (BioRC)

Cardiac Center/CVI/CICU

Cardiac OR

NICU

