Collecting the Data:
The Essential Value of a Birth Defects Biorepository

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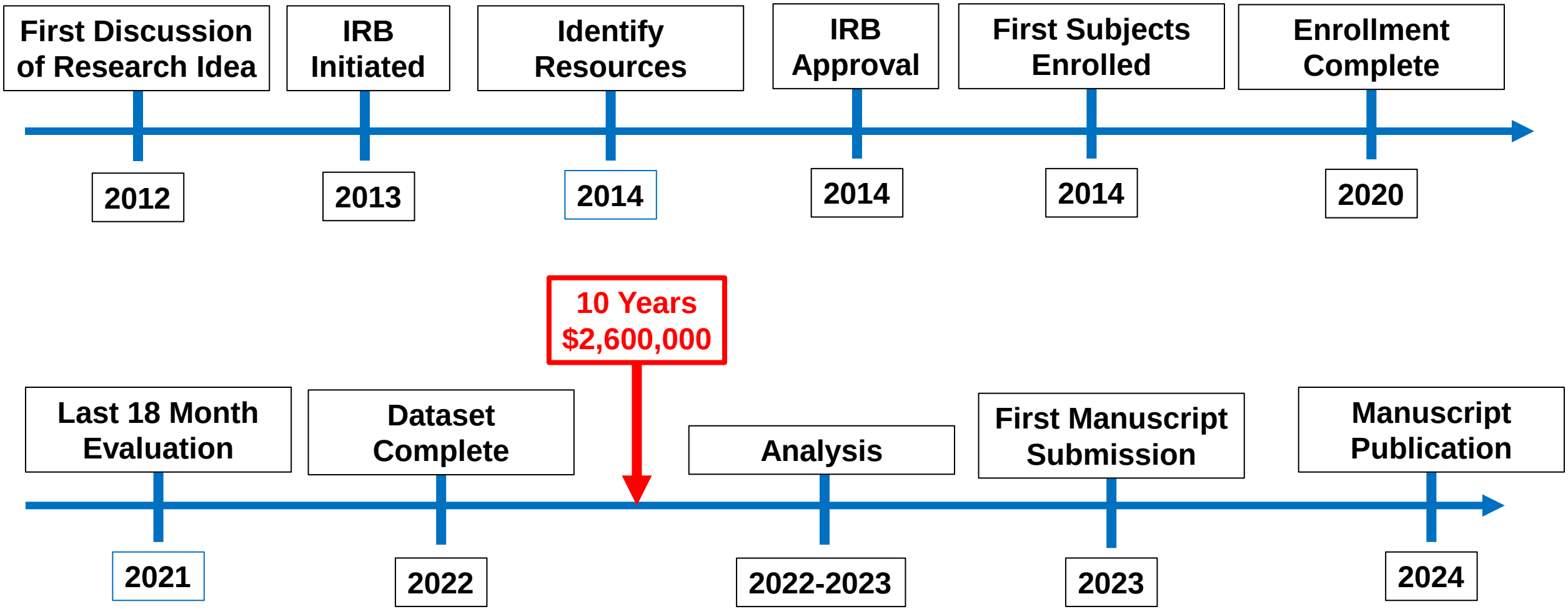
Disclosures

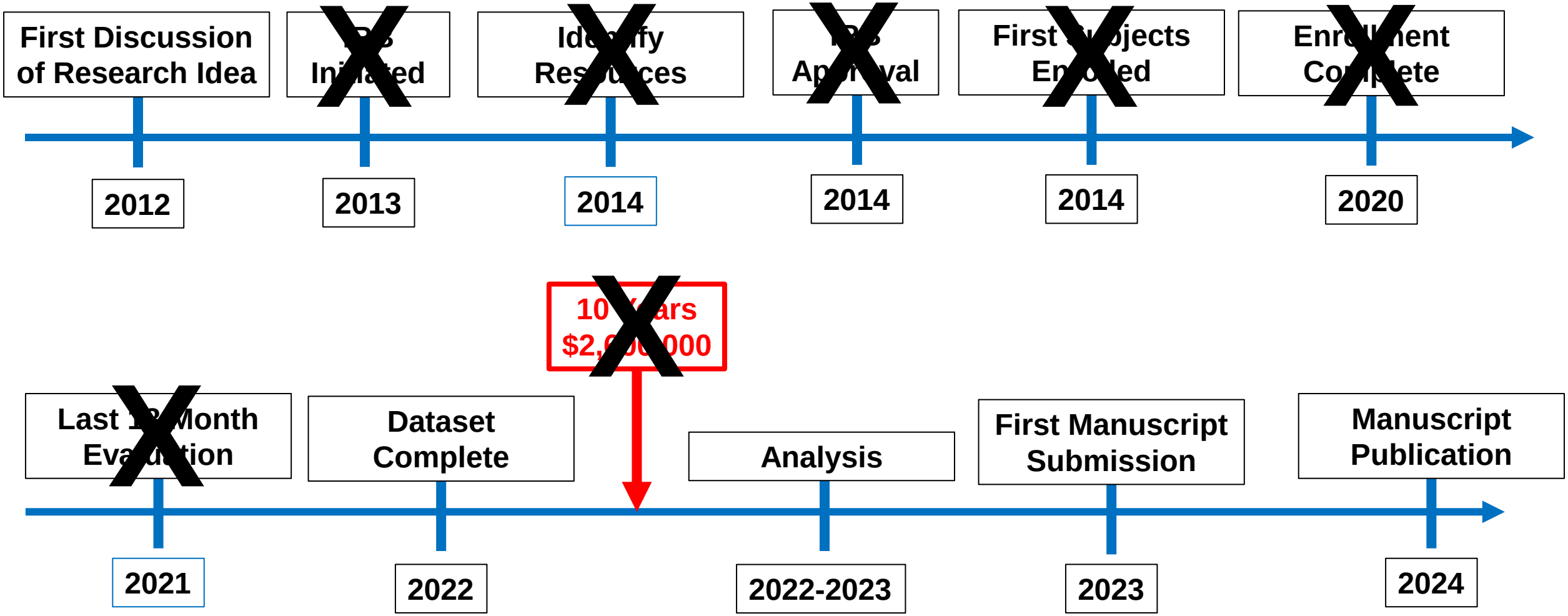
Conflicts: Co-PI of the Birth Defects Biorepository

Off-label Use: None

Typical Study Timeline *(Maternal Progesterone Study)*

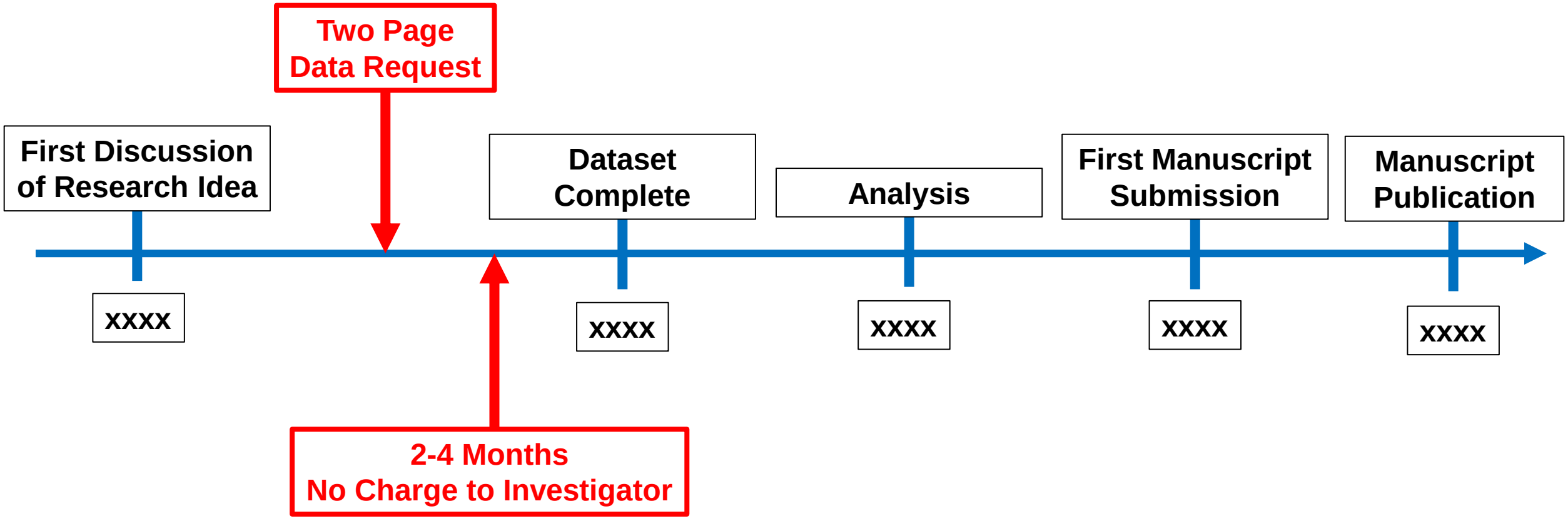


Study Timeline *(What If?)*



Study Timeline *(Using Birth Defects Biorepository)*

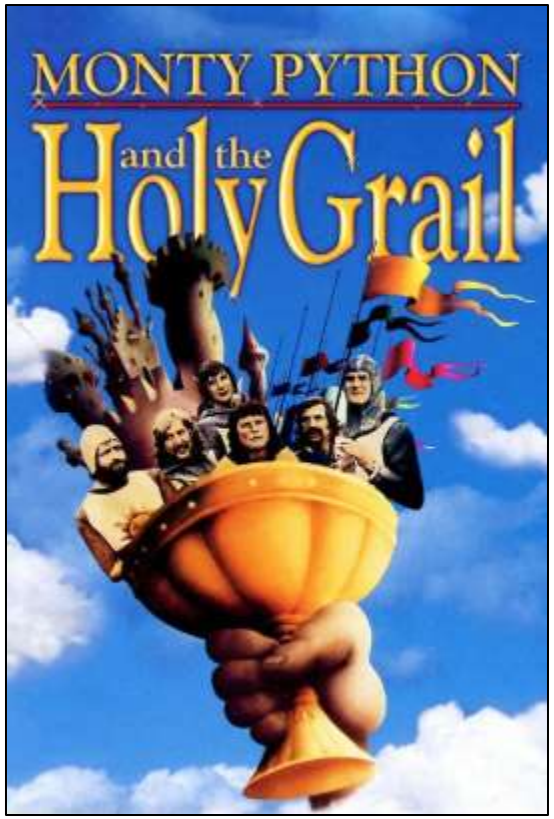
The goal is to dramatically shorten the time from research idea to discovery.



The Essential Value of a Birth Defects Biorepository

Precision Medicine

Development of tailored, individualized therapeutic strategies for patients based on understanding their unique characteristics, risk profiles, and responses to therapies.



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Precision Medicine

Development of tailored, individualized therapeutic strategies for patients based on understanding their unique characteristics, risk profiles, and responses to therapies.

1. Creation of large patient cohorts across the Lifespan.

2. Collection of comprehensive data including: patient characteristics, details of therapy, fetal development, genetic variants, socioeconomic factors, and long-term outcomes.

3. Collection of human biological samples and integration with associated data in a systematic way for research purposes.

4. Decrease time from research idea to discovery.

5. Steward scarce resources (ROI).

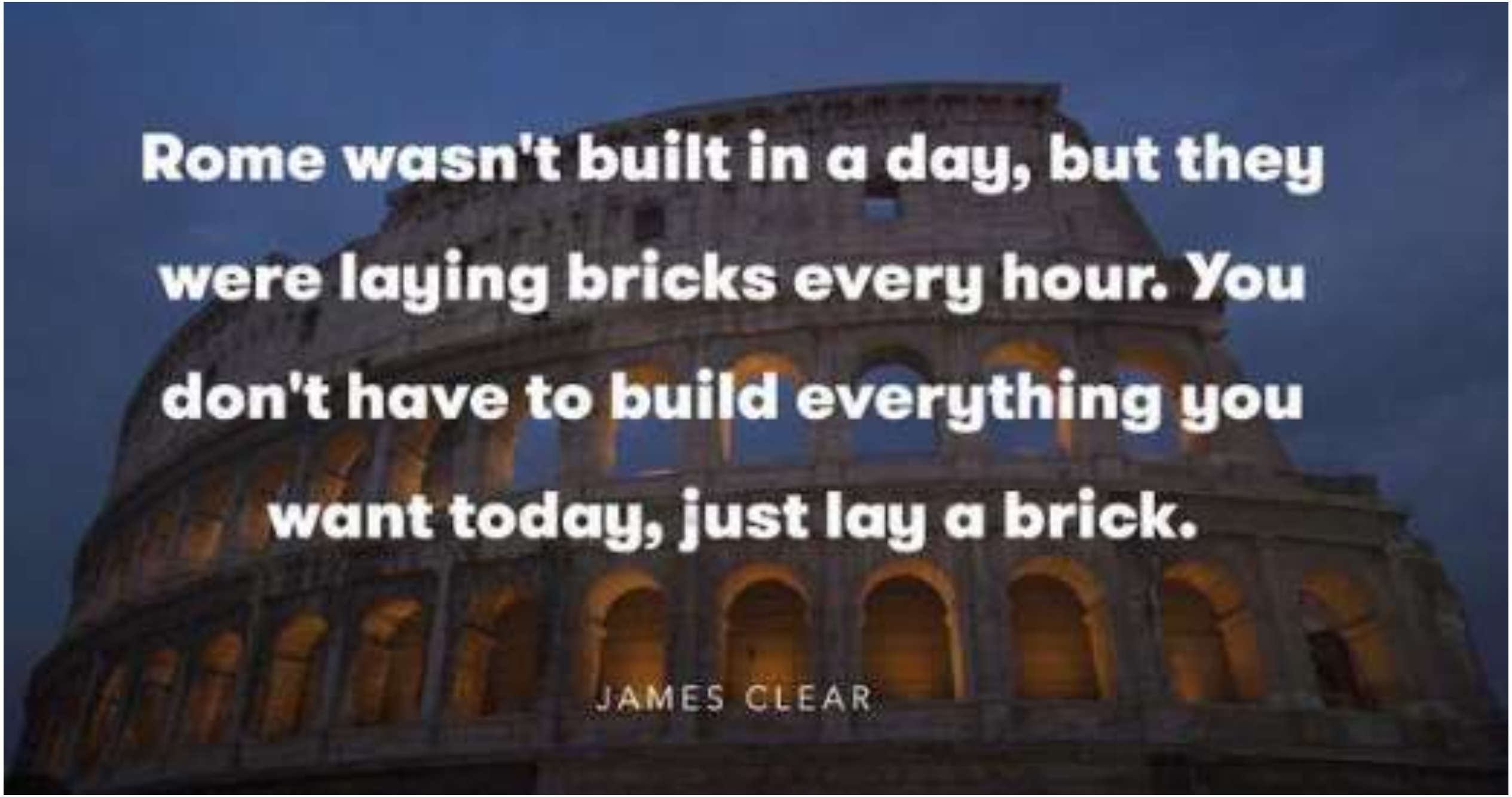
The Birth Defects Biorepository

The Birth Defects Biorepository (BDB) was established in June 2019 by the Center for Fetal Diagnosis and Treatment (CFDT) and the Department of Surgery in collaboration with families at CHOP.

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:

1) investigations into the etiologies of birth defects,
2) advancement of personalized medicine, and
3) understanding of long-term outcomes for children with birth defects.

In 2023, in collaboration with the Cardiac Center and the CHOP Cardiovascular Institute, the BDB expanded to initiate the CC/CVI/BDB.


Rome wasn't built in a day, but they were laying bricks every hour. You don't have to build everything you want today, just lay a brick.
JAMES CLEAR

Cardiology 2020

Design, Development, and Benefits of a Birth Defects Registry

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The Birth Defects Biorepository

Cardiology 2020

Children's Hospital of Philadelphia
Department of Biomedical & Health Informatics

BIRTH DEFECTS BIOBANK
Enrollment and demographics

Total affected child: 182	Family trios: 143
Total Mother/Surrogate: 176	Family duos mom - baby: 38
Total enrolled: 504	Family duos father - baby: 0
Multiple Gestations Enrolled: 11	Singleton babies: 1
Both Twins Affected: 6	Agreed to relinking PHI: 409
One Twin Affected: 5	Agreed to future contact: 356

Cardiology 2025

Enrollment and Demographics

Enrollment | Ancestry | Specimen | Birth & Deliveries | Maternal Factors | Placenta | Diagnoses | Procedures

Last Updated: 2/18/2025 1:01:10 PM

All Enrollment | Proband Enrollment

Total Probands	1,830
Total Mother/ Surrogate	1,663
Total Enrolled	4,757

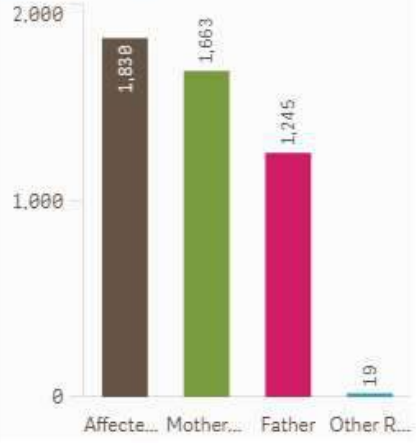
Multiple Gestations	115
Triplets	4
Twins	111
Sets of Conjoined Twins	8

Family Trios	1,327
Family Duos: Mom-Baby	465
Family Duos: Father-Baby	17
Singleton Babies	21
Agreed to Relinking PHI	3,069
Agreed To Future Contact	2,856

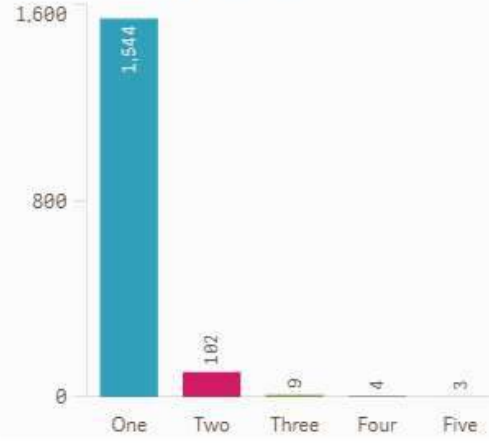
Enrollment Sources
CFDT includes Fetal Reunion

CFDT Probands	1,434
Cardiac Center Probands	186
Pulmonary Vein Clinic Probands	20
EAT Clinic Probands	3

Familial Roles



Proband Count per Family



The Birth Defects Biorepository

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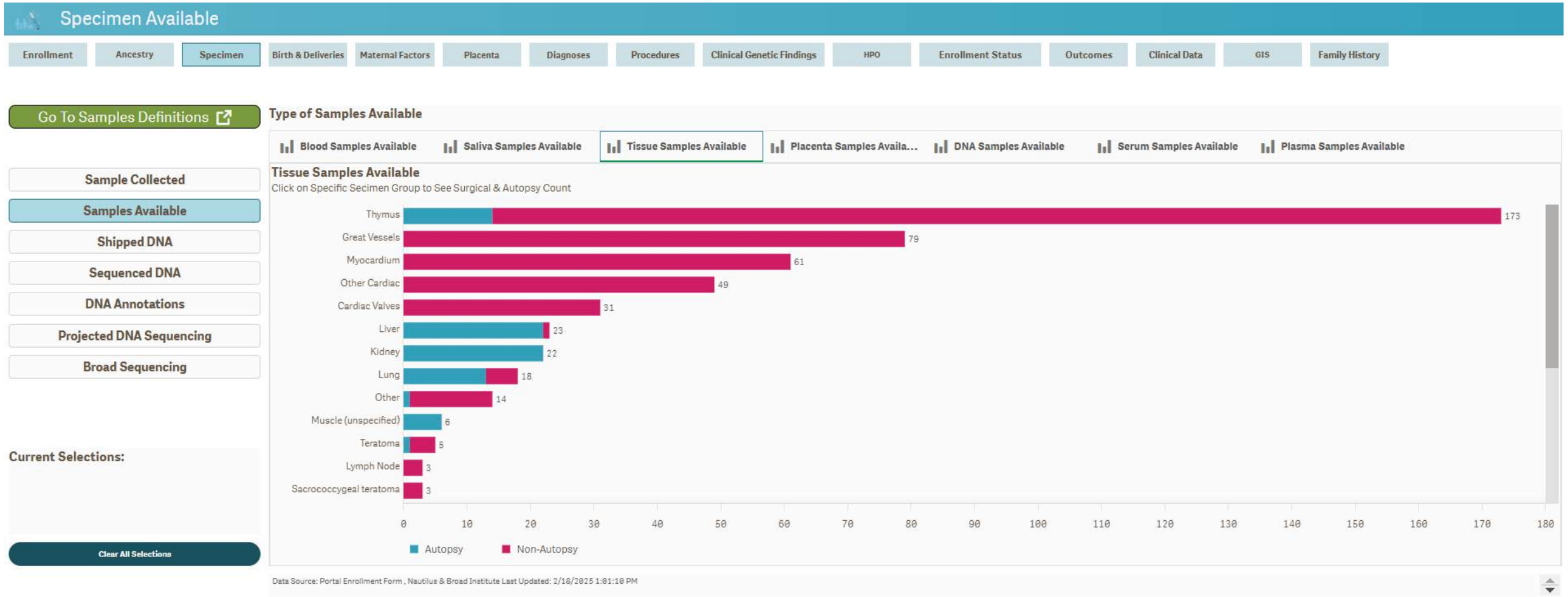

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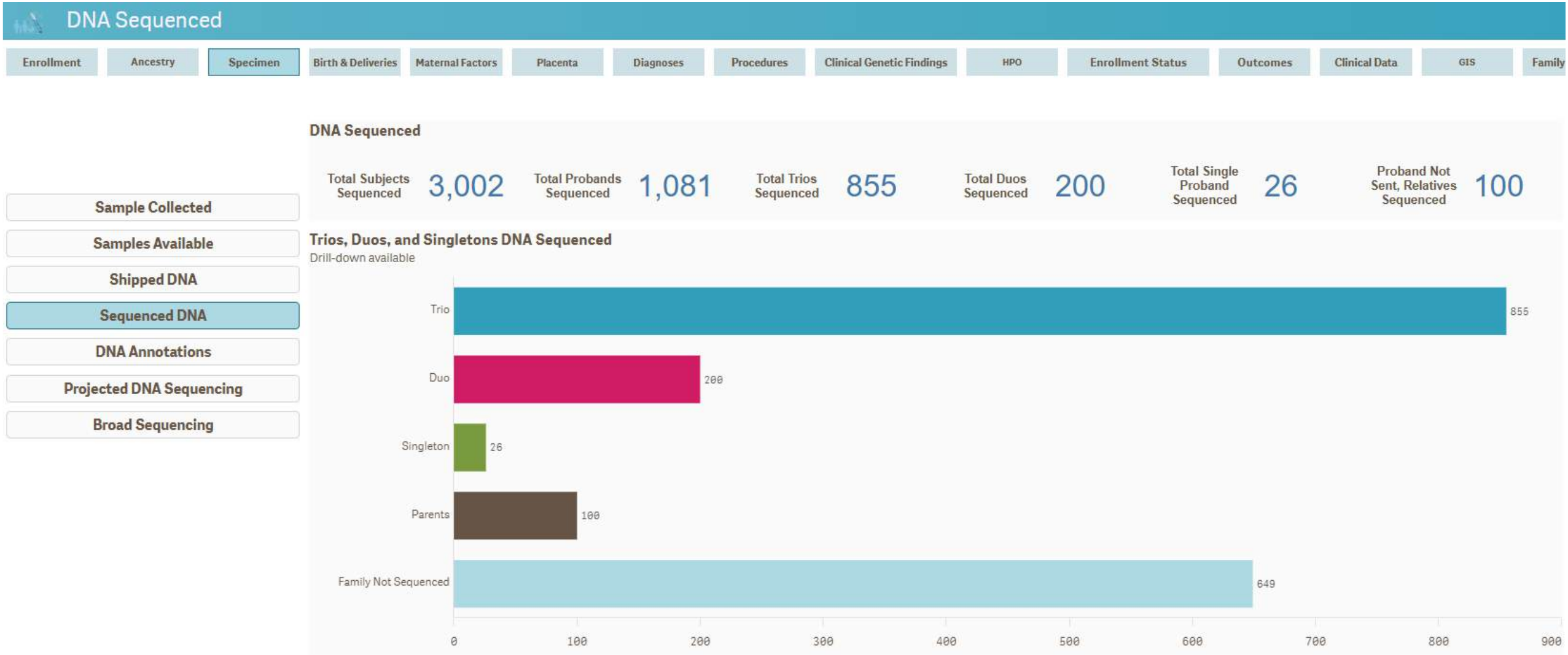

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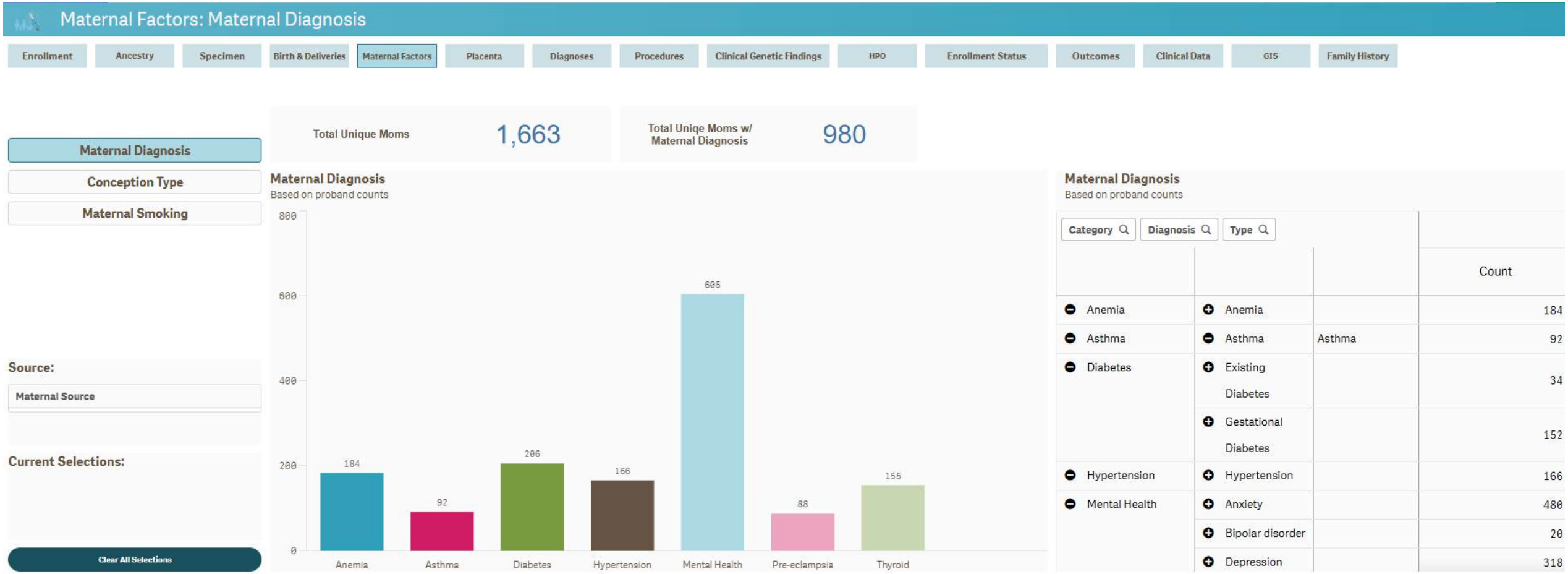

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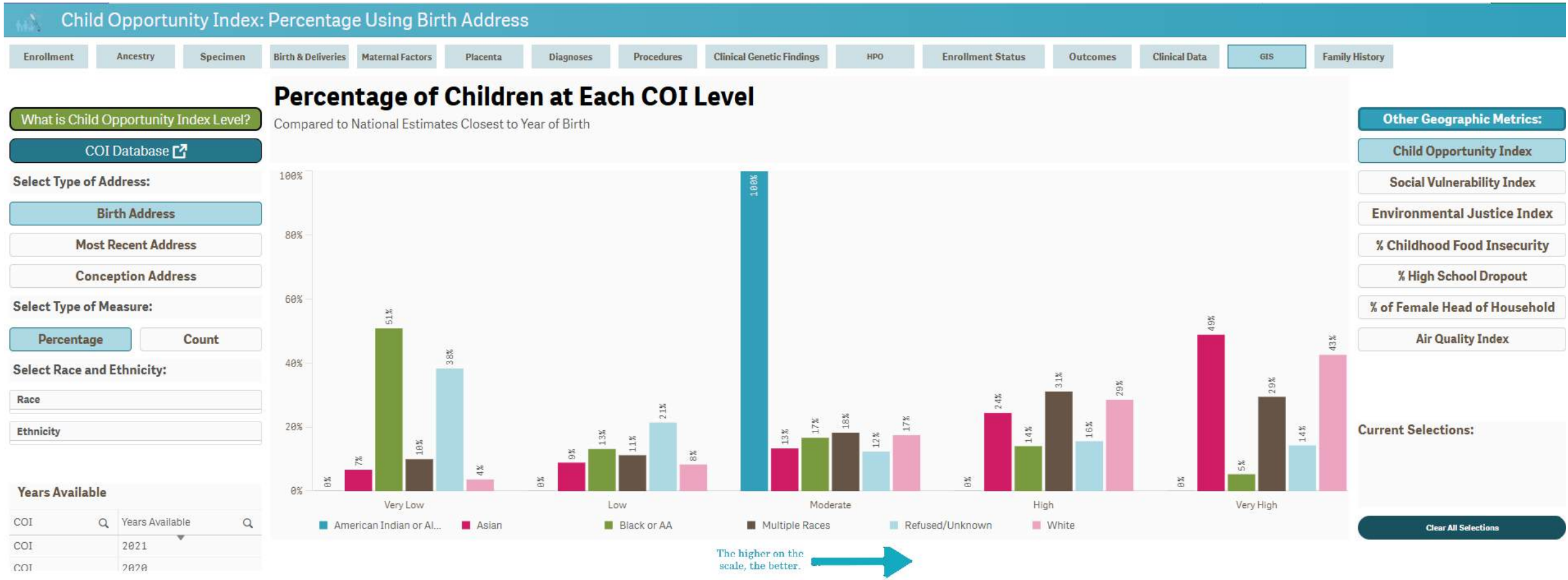

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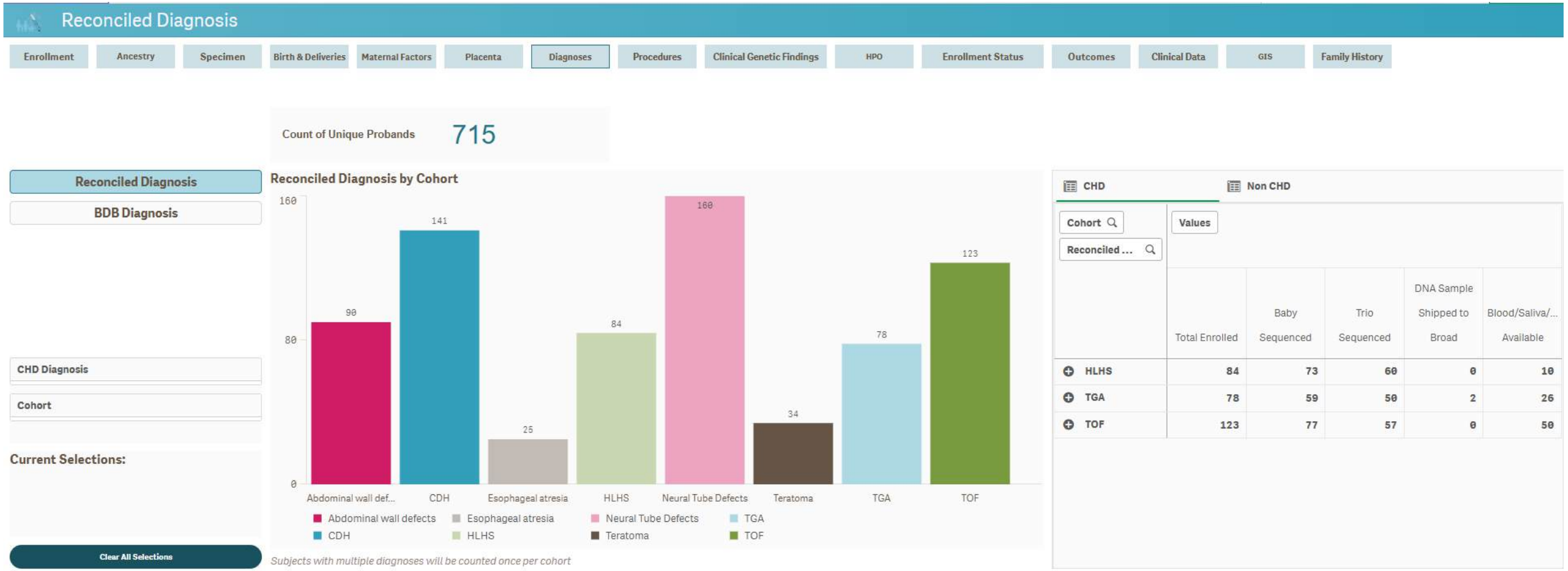

The Essential Value of a Birth Defects Biorepository

Project Number	Investigators	Project Title
1	Gaynor, JW and Ahrens-Nicklas, Rebecca	Sarcomeric Variants in Congenital Heart Disease
2	Xing, Yi and Helbig, Ingo	Cis Variation in RNA Processing and Regulation across the Birth Defects Biorepository
3	Gaynor, JW and Ahrens-Nicklas, Rebecca	Homeodomain-interacting protein kinase 2 (HIPK2) and Congenital Heart Disease
4	Jacobwitz, Marin	Interaction of Placental Pathology, Genomics, and Neurodevelopmental Outcomes in Children with Prenatally Diagnosed Birth Defects
5	Moldenhauer, Julie and Heuer, Gregory	Candidate Genes Associated with Prenatally Diagnosed Spina Bifida
6	Strong, Alanna	Novel gene discovery for ciliopathy-spectrum disease
7	Pratap, Nick	Development and Validation of a Scalable Multidimensional Organ Recovery-Based Framework ("PODIUM C") to Understand Pediatric Cardiac Surgical Outcomes
8	Hayeck, Tristan	Improving diagnosis and interpretation of genomic variants
9	Kienzle, Martha and Sutton, Robert	Adrenoreceptor Polymorphisms Among Pediatric Patients with In-hospital Cardiac Arrest
10	Liao, Eric	12Development of a Collaborative CHOP Craniofacial Anomaly Cohort
11	Punchak, Maria	Sociodemographic, geospatial variables among spina bifida patients treated at CHOP and their association with patient outcomes
12	Gacita, Anthony and Kelly, Daniel	Identifying Disease Associated Variants in Estrogen Related Receptor (ERR) Binding Sites
13	Gordon, Scott	Defining cellular and molecular consequences of congenital viral infection at the maternal-fetal interface
14	Coker, Eric	A cohort study investigating the associations between prenatal particulate matter air pollution and adverse birth outcomes among newborns with birth defects.
15	Gardner, Monique and Erikson, Lori	Second surgical site validation of SNARE-ing the reason for post-cardiac surgery cortisol steroid insufficiency

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16	**Rand, Elizabeth**	Investigation of genetic contributors to cholestasis in complex congenital heart disease
17	**Avitabile, Catherine**	The association between maternal-fetal environment and ventricular dysfunction in congenital diaphragmatic hernia (CDH)
18	**Ross, Margaret**	Understanding genetic complexity in spina bifida
18	**Josowitz, Rebecca**	Genetic variation in fetuses with congenital heart disease and placental abnormalities
20	**Krantz, Ian**	The molecular landscape of congenital diaphragmatic hernia
21	**Spinner, Nancy**	The Genetic Etiology of Esophageal Atresia and Tracheoesophageal Fistula
22	**Levy, Robert**	Serotonin transporter polymorphism (5-HTTLPR) genotype in patients with bicuspid aortic valve with moderate to severe aortic stenosis
23	**Raffini, Leslie**	Impact of genetic variation in thrombophilia related genes on placental structure, fetal growth, complications of pregnancy, and post-natal outcomes in CHD
24	**Campbell, Ian**	Assessing the Pharmacogenetic Landscape at CHOP
25	**Morton, Sarah**	Impact of Parental Origin of Genetic Variants on Placental Function and Outcomes Among Infants with Congenital Anomalies
26	**John, Audrey and Gaynor, JW**	Fecal Volatile Organic Compounds and Necrotizing Enterocolitis in the CICU
27	**Gaynor, JW; Moldenhauer, Julie; Rychik, Jack; Linn, Rebecca; Logsdon, Glennis; and Ittenbach, Richard**	Placental Senescence and Adverse Outcomes in Hypoplastic Left Heart Syndrome (HLHS)
28	**Kahn, Mark**	Genetic mutations in TGFB signaling pathway components contributes to giant omphalocele in children
29	**Hayeck, Tristan; Allen, Andrew; and Landstrom, Andrew**	Improving diagnosis and interpretation of genomic variants related to sudden cardiac arrest
30	**Gaynor, JW; Moldenhauer, Julie; and Russell, Mark**	Examining Vascular Signaling in Pregnancies Affected by a Fetus with a Heart Defect
31	**Logsdon, Glennis and Spinner, Nancy**	The role of centromere dysfunction in trisomy and monosomy disorders
32	**Grant, Struan**	Identifying Rare Pathogenic Noncoding Variants in a Pediatric Cohort
33	**Campbell, Ian**	Development of Pediatric-Specific Pre-Trained Large Language Models
34	**Wang, Kai**	Multimodal identification of causal genes for rare diseases

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35	**Alexander-Bloch, Aaron**	Imaging-genetics of clinically-acquired brain MRIs and relationship to neuropsychiatric risk
36	**MacFarland, Suzanne**	Novel Genomic Drivers in Juvenile Polyposis Syndrome
37	**Cotney, Justin**	Identification of rare non-coding variants in craniofacial abnormalities
38	**Jacobwitz, Marin**	Placental Pathology in Down Syndrome
39	**Levy, Robert, Stachelek, Stanley, and Pei, Liming**	Serotonin (5-HT) transporter (SERT) mechanisms that affect the progression of pulmonary regurgitation (PR)
40	**Helbig, Ingo**	HPO-based analytical framework to identify novel genetic etiologies.

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Presentations and Abstracts:

Accepted: 7

Submitted: 1

Grants Utilizing BDB Data and Biospecimens:

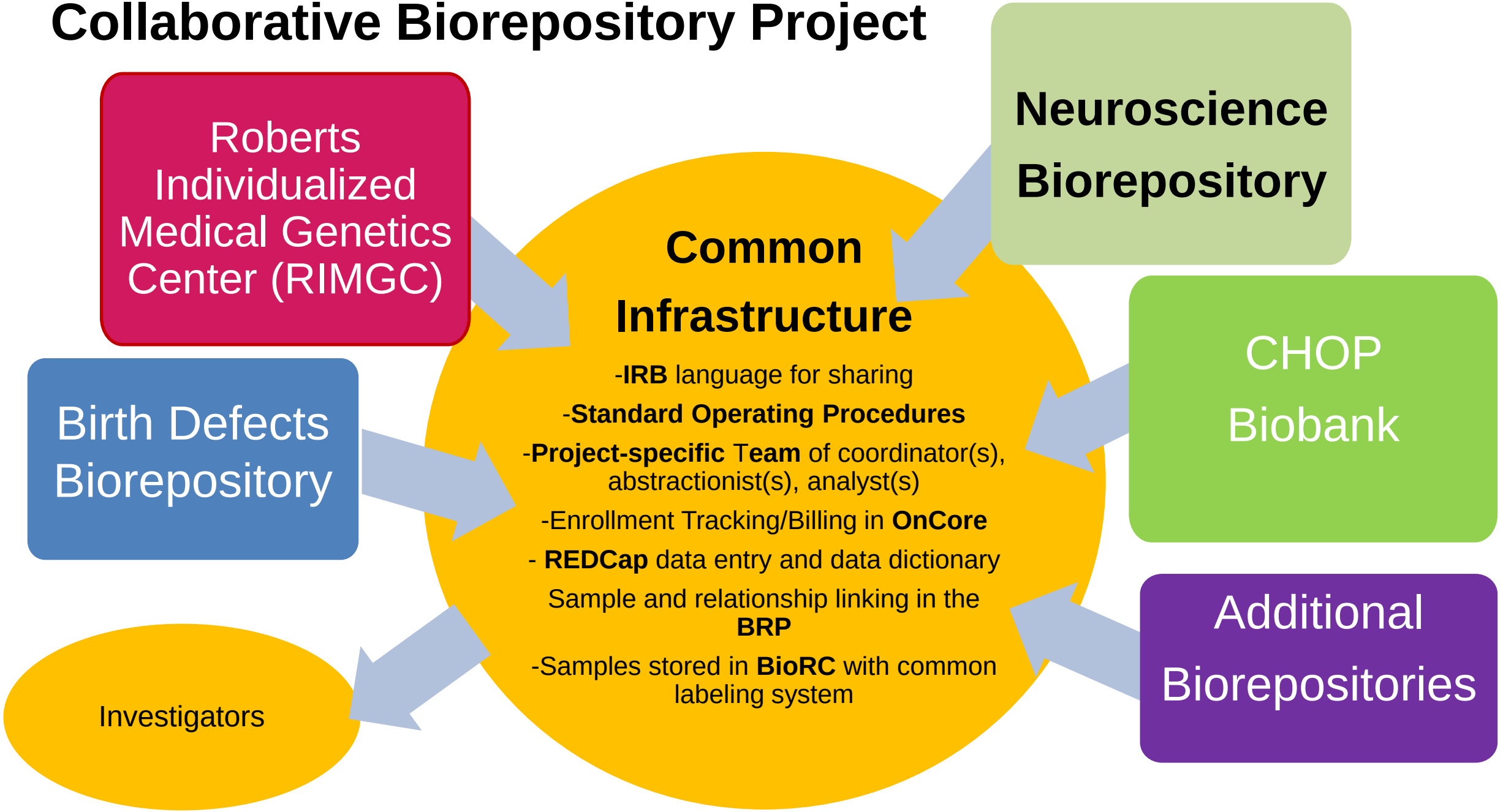
Funded: 9

Submitted: 2

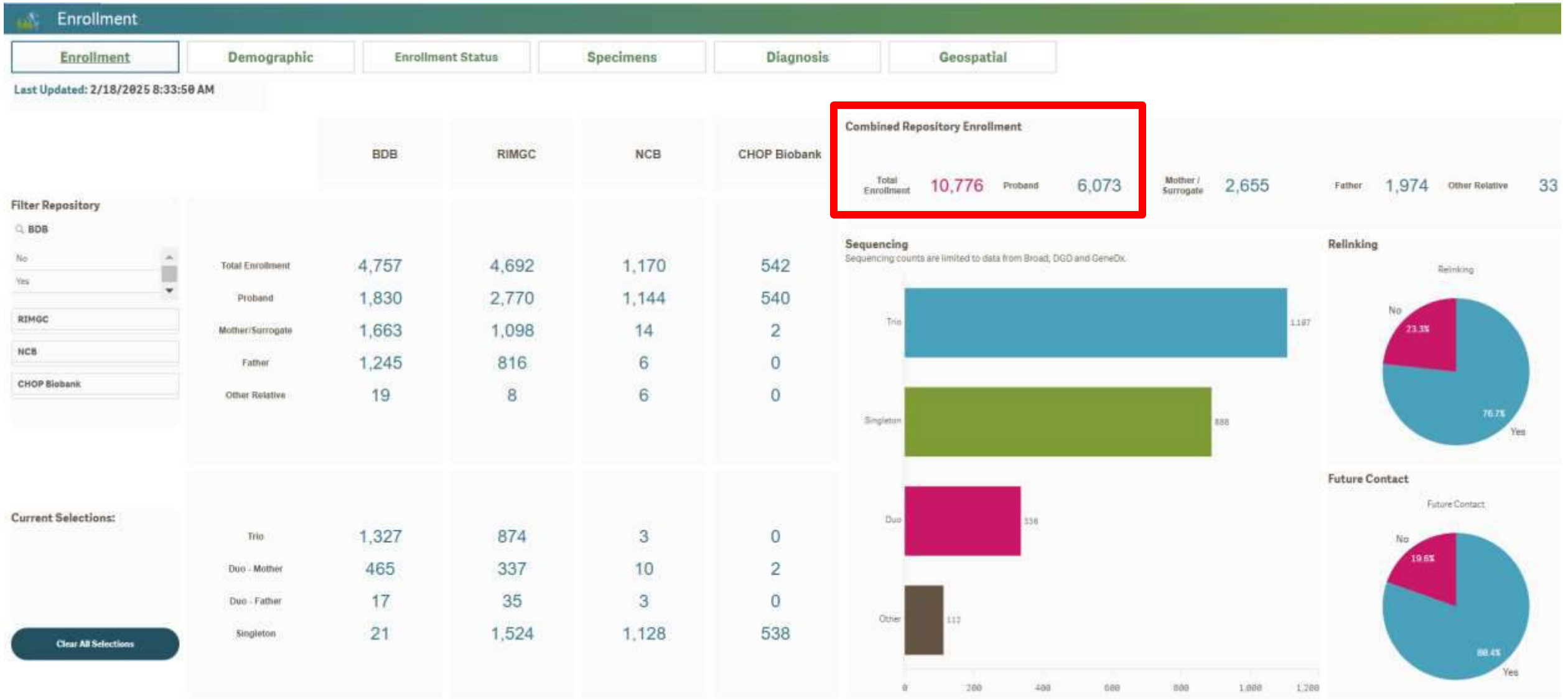
Manuscripts:

Submitted: 1

Collaborative Biorepository Project



Collaborative Biorepository Project



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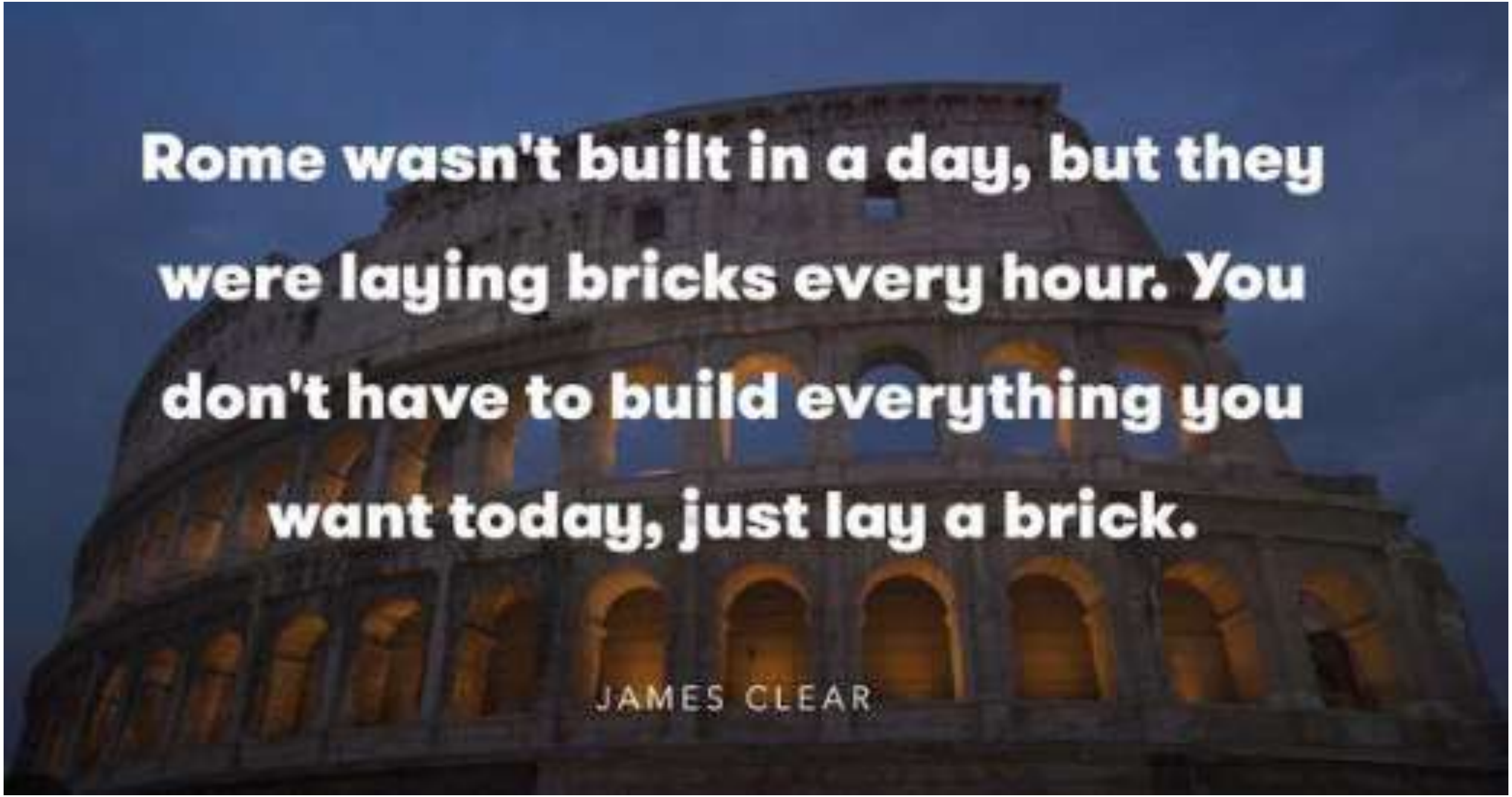
MARCH 2025 EMERGING INNOVATORS IN COLLABORATIVE RESEARCH

"Birth Defects Biorepository at CHOP"

Speaker: Stacy Woyciechowski, MS,
Clinical Research Program Manager II
Department of Surgery

"Genetic variation and outcomes: The role of the placenta in fetal congenital heart disease"

Speaker: Rebecca Josowitz, MD, PhD,
Physician/Medical Fellow
Department of Pediatrics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