

BEACONS_{NBS}

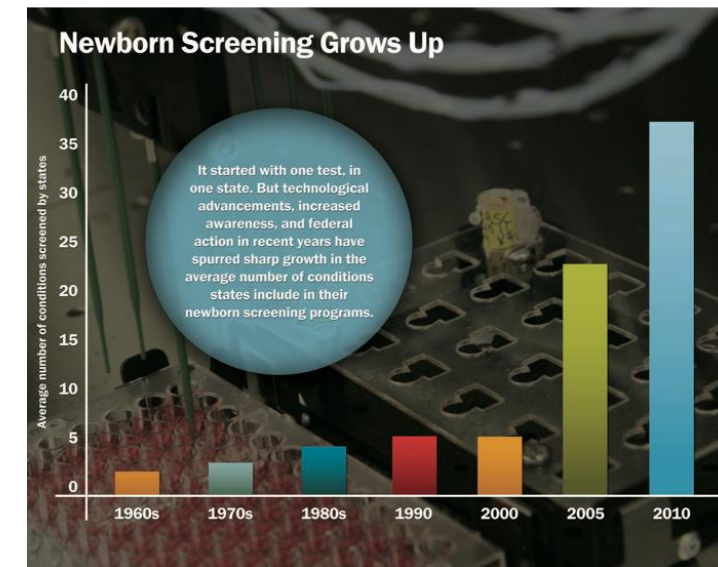


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Newborn Screening (NBS)

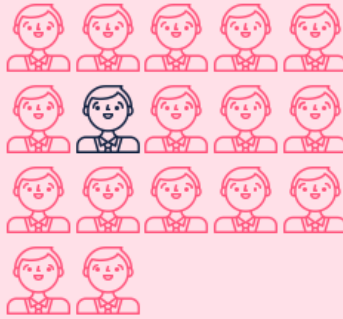
A Highly Successful Public Health Program

- Recommended Uniform Screening Panel (RUSP) – 40 core conditions
- NBS regulated by states
- Most states report $\geq 99.9\%$ participation
- Mandatory and unconsented
- 1 out of every 300 newborns diagnosed with a core condition



1/17

people develop
a rare disease



400m

people with
rare diseases



>7000

different types



25%
adults

75%
children



28%

neonatal intensive
care deaths



80%

are genetic
in origin



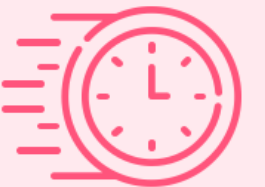
30%

of affected children
never reach their
5th birthday



5years

on average
to diagnose



There are >800 Treatable Genetic Disorders

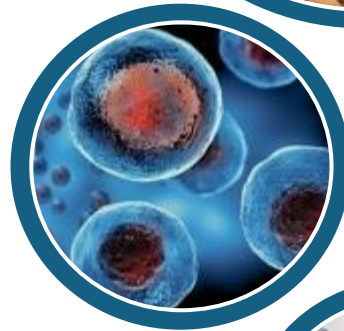
328 Specific medication



270 Special diet or supplements



175 Stem cell or organ transplant



70 Specialized proteins (enzymes or immunoglobins)



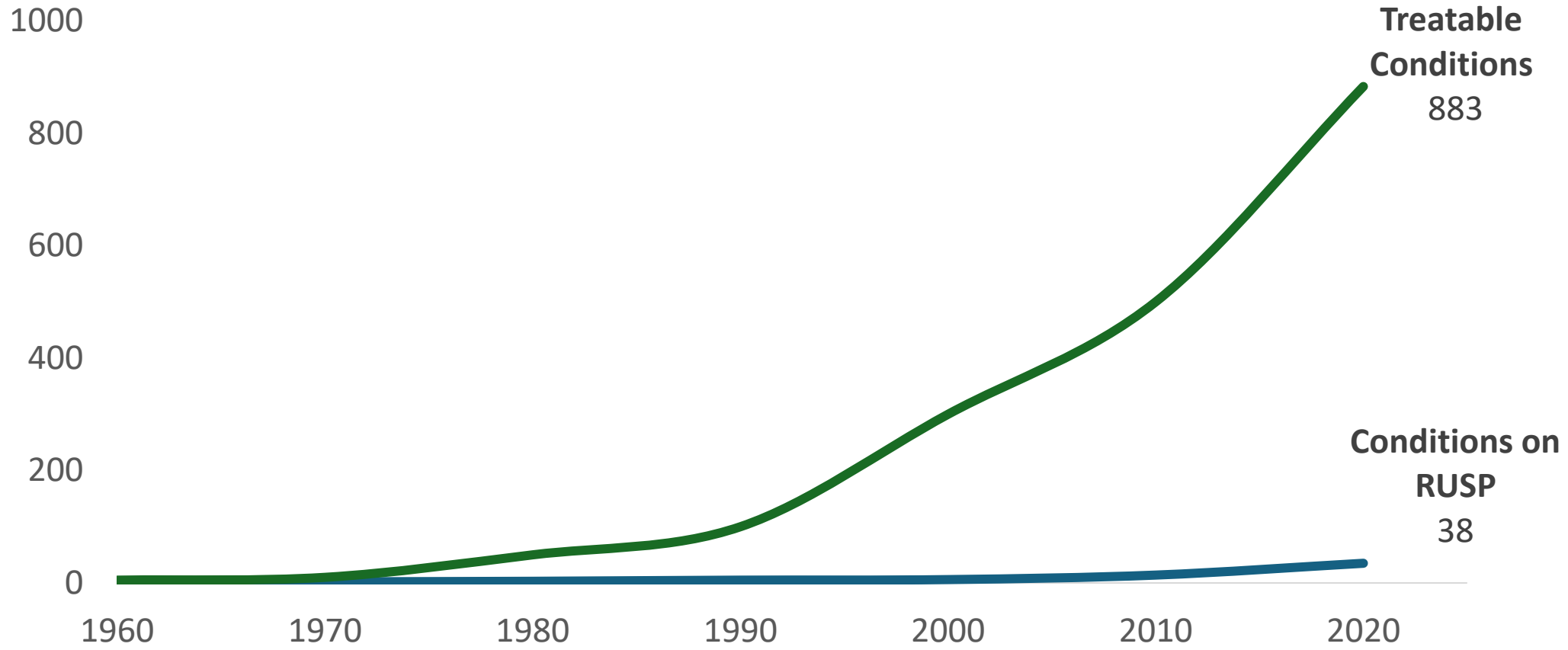
36 Medical procedures



4 Gene therapy (many more in development)



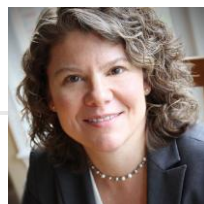
Yet the RUSP Identifies only 38 of those Treatable Conditions





BABYSEQTM

NIH HD077671 / TR003201
(2013-2025)



 Mass General Brigham

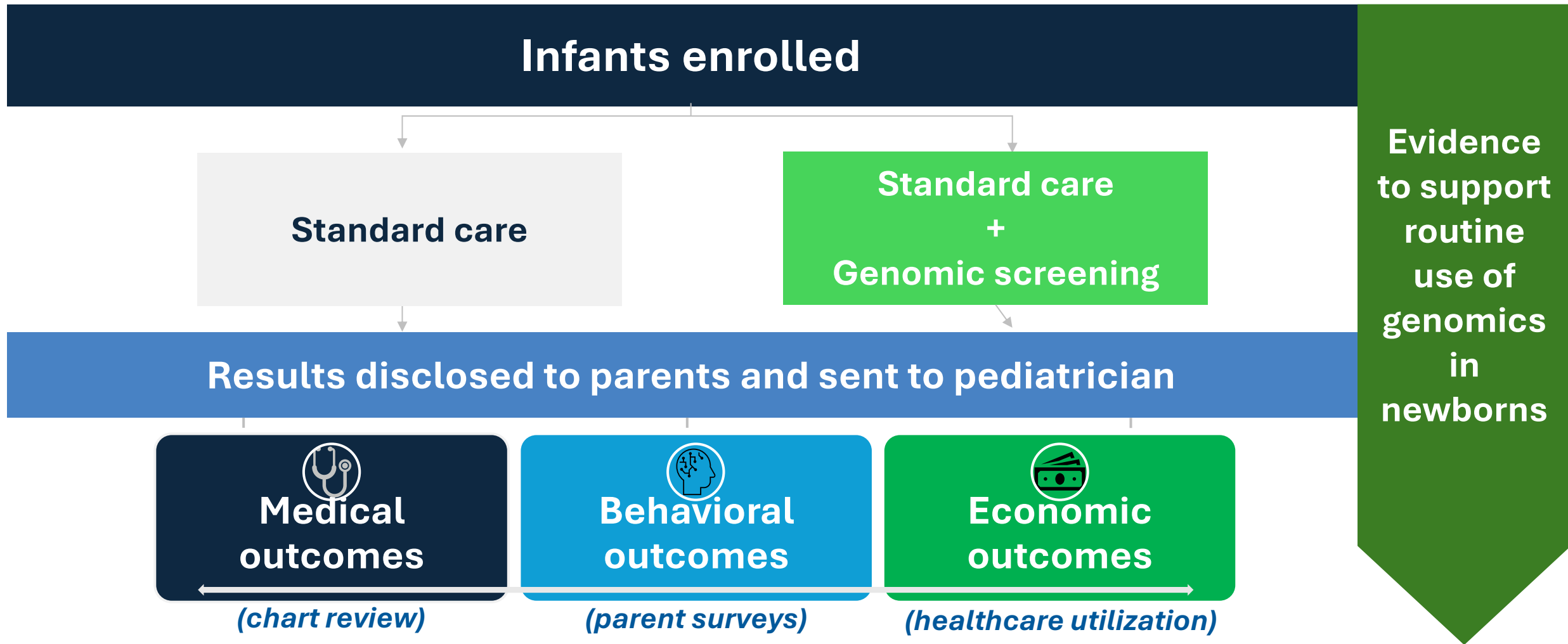


UAB

BCM
Baylor College of Medicine



BabySeq: The Only RCT of Sequencing in NBS



BabySeq Reporting



Childhood-onset and highly actionable
adult-onset disease



Carrier status



Pharmacogenomic (PGx)
(pediatrics)

Studies of Genomic Sequencing in NBS



NBSxWGS Collaboratory

- NIH Venture Initiative (2025-2028)



Analysis of WGS results for **5-10 state public health labs**



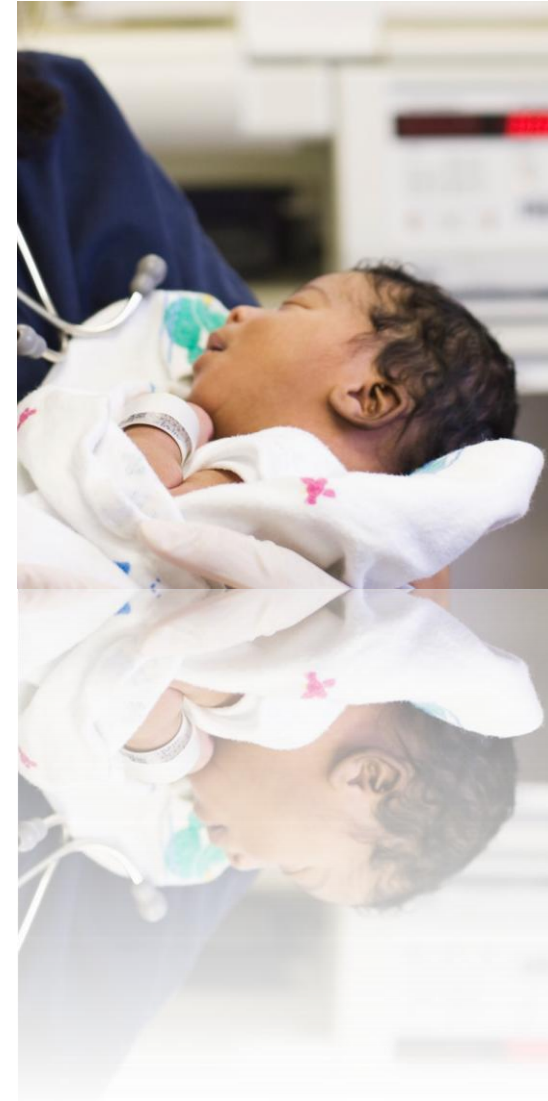
Enable **broad access to WGS** as a **screening tool** in the **newborn** period



Gene panel of **serious rare diseases** with available **early treatment options**



Examine ethical, legal, and social implications (**ELSI**) of population-wide NBSxWGS



BEACONS_{NBS}

Building Evidence And Collaboration for
GenOmics in Nationwide Newborn
Screening



A Team of Teams



Children's Hospital
at Montefiore Einstein



7 State PHLs
Iowa
Minnesota
New York
Oregon
Puerto Rico
South Carolina
Texas

A Study of Feasibility

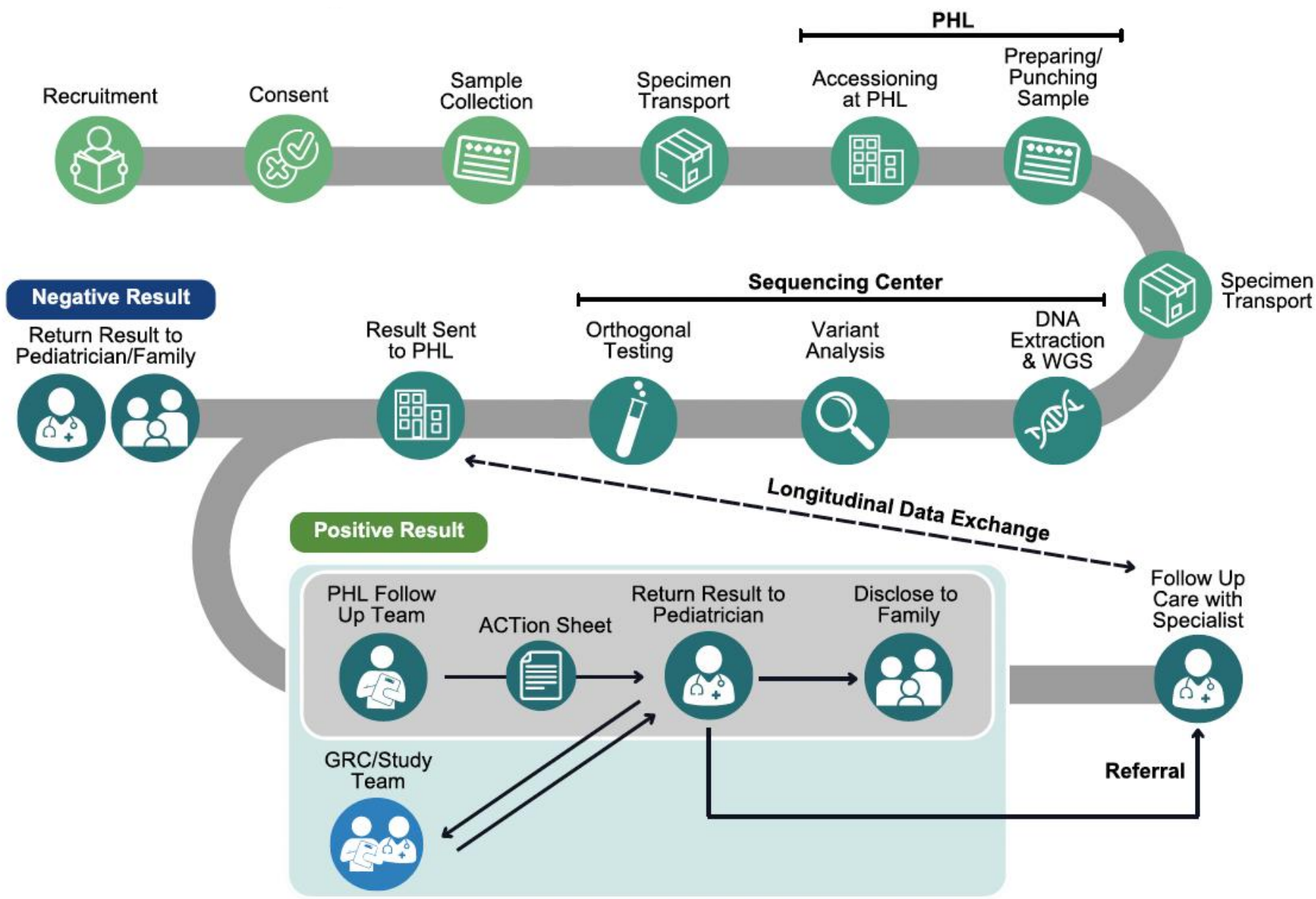


Scientific

Programmatic

Human

<ul style="list-style-type: none">• Gene list	<ul style="list-style-type: none">• Efficient	<ul style="list-style-type: none">• Consent
<ul style="list-style-type: none">• WGS pipeline	<ul style="list-style-type: none">• Scalable	<ul style="list-style-type: none">• Uptake
<ul style="list-style-type: none">• Variant interpretation, confirmation	<ul style="list-style-type: none">• 7-10 multi-state PHLs	<ul style="list-style-type: none">• Acceptability
<ul style="list-style-type: none">• Medical followup	<ul style="list-style-type: none">• Birthing hospitals	<ul style="list-style-type: none">• Communication



Steering Committee

MPIs

Robert Green, MD, MPH

Ingrid Holm, MD, MPH

Nina Gold, MD, MS

Melissa Wasserstein, MD

Aaron Goldenberg, PhD

Jelili Ojodu, MPH

Stacey Pereira, PhD

Sikha Singh, MHS, PMP

Britt Johnson, PhD

Swaroop Aradhya, PhD

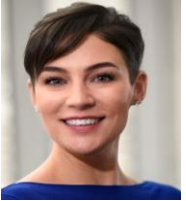
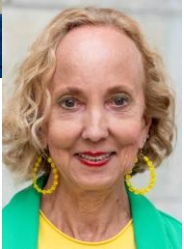
Carrie Zawatsky, MS, CGC

Stephanie Coury, MS, CGC

State PHLPs

Community Advisory Board (CAB)

NIH representatives



The Team

