



Nebraska Newborn Screening

ADVISORY COMMITTEE

JULY 28, 2026



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Agenda

- I. Open Meeting Act Announcement
- II. Introduction
- III. Review/approval of minutes from the April 2026 meeting
- IV. Laboratory update (Revvity Omics)
- V. Legislation, Regulation, and Policy
- VI. Intern Presentation on Physician Manual Updates
- VII. Break
- VIII. Intern Presentation on Physician Manual Updates
- IX. Programmatic Updates
- IX. Data Presentation
- X. Other Business
- XI. Adjourn

I. Open Meeting Act Announcement

WELCOME TO THE JULY 26 MEETING OF THE NEBRASKA NEWBORN SCREENING ADVISORY COMMITTEE. IN ACCORDANCE WITH THE “OPEN MEETINGS ACT”, THIS MEETING WAS ANNOUNCED TO THE PUBLIC BY POSTING ON THE STATE OF NEBRASKA PUBLIC MEETING CALENDAR BEGINNING ON NOVEMBER 4, 2025. A COPY OF THE “OPEN MEETING ACT” AND AN AGENDA FOR THIS MEETING CAN BE FOUND ATTACHED TO THE MEETING NOTICE IN THE ONLINE CALENDAR.

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II. Introductions



III. Review/approval of minutes from the April 2026 meeting

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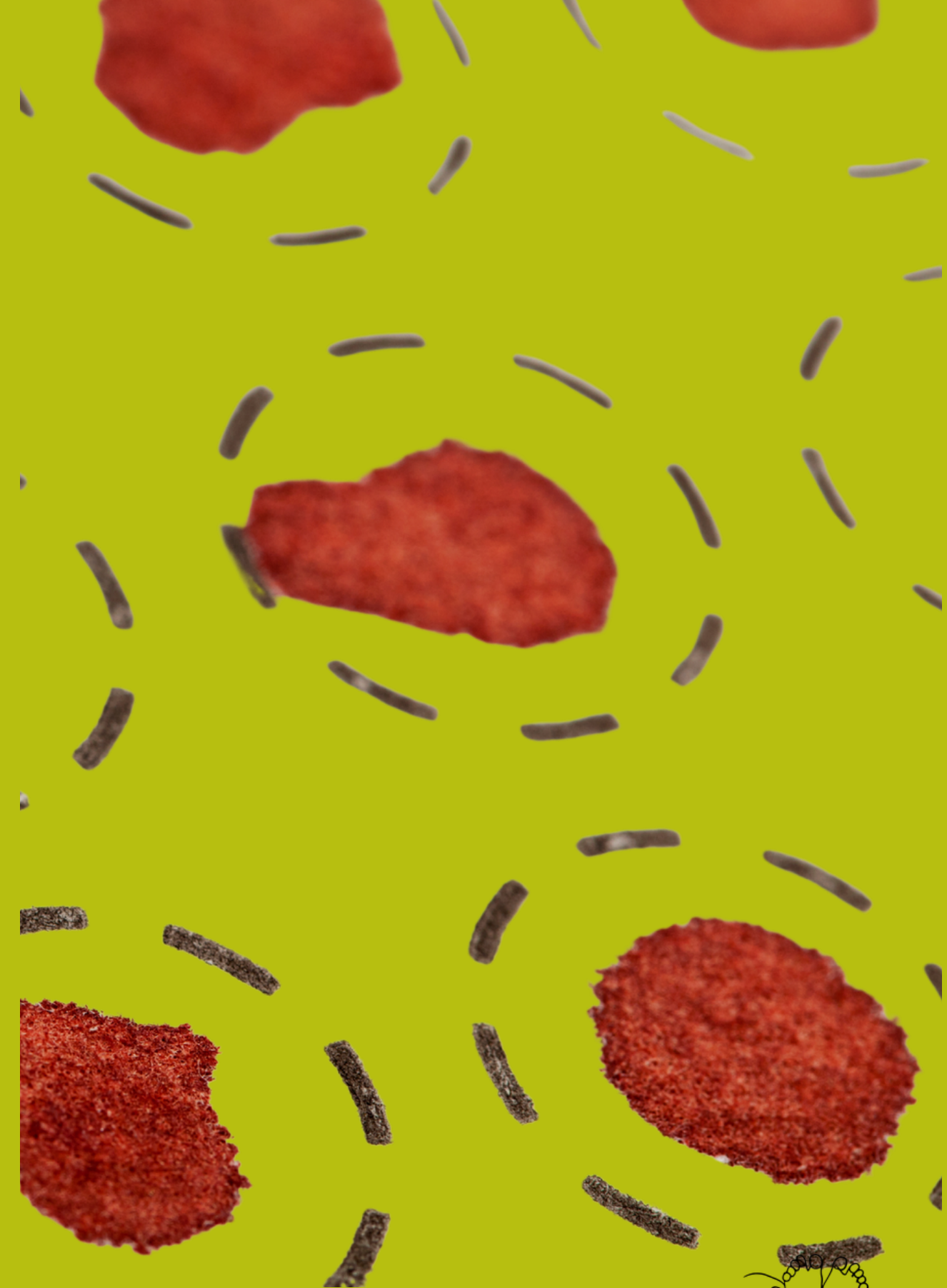
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IV. Laboratory update

a. (Revvity Omics)

V. National and State Updates:



State Updates:

1. MPS II and CCHD regulatory hearing held on July 16th. Three comments submitted, all were comments in favor of the changes. Next step is Board of Health Meeting in September.
2. GAMT still being reviewed by leadership (as of 7/20/26).
3. MLD, DMD, and IKD submitted for leadership review and approval.



Whole Genome Sequencing Pilots in Newborn Screening (NBS)

ICONS and emerging genomic newborn screening initiatives



Our Agenda for Today

GENOMIC NEWBORN SCREENING OVERVIEW

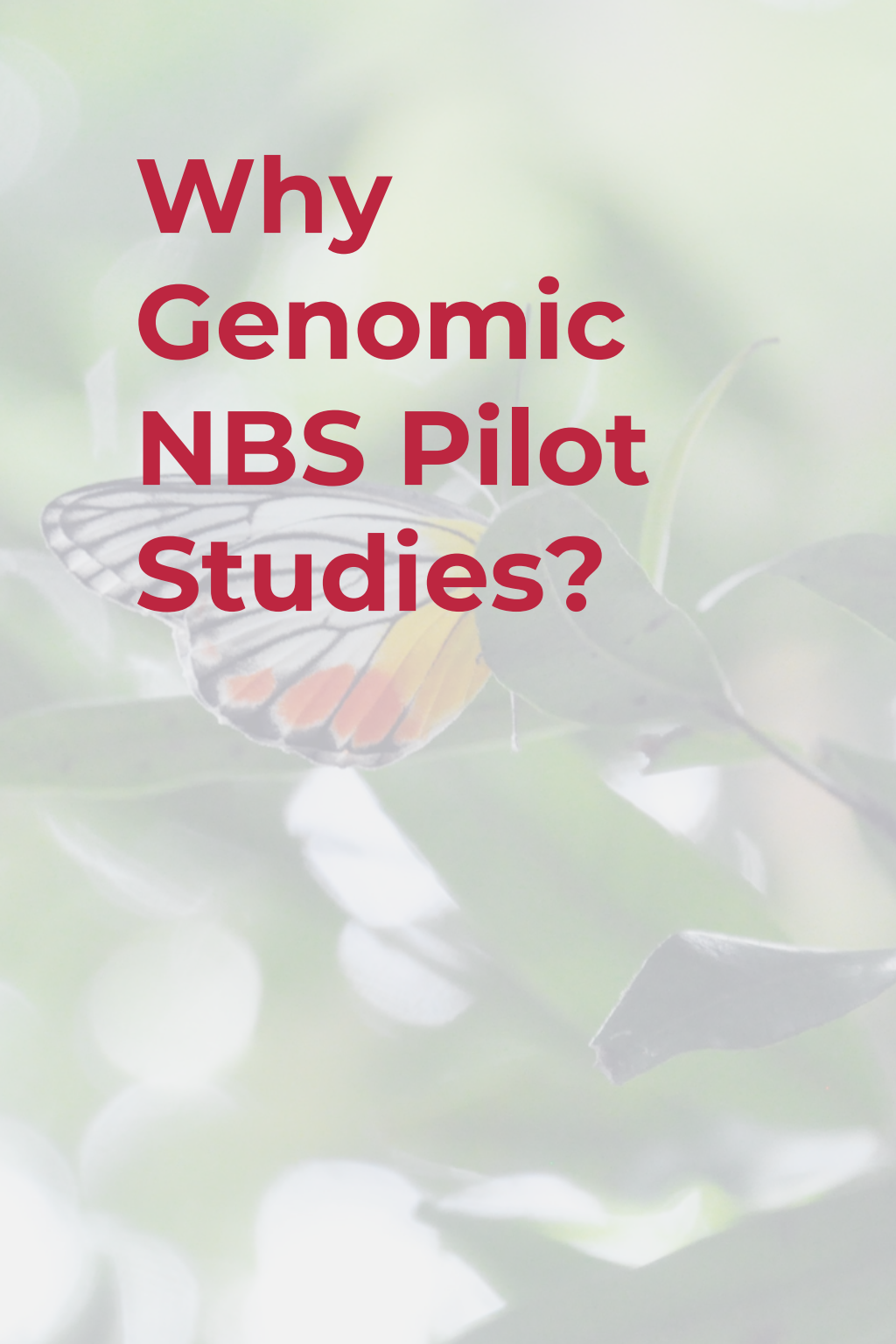
Pilot Projects

ICONS

Key Lessons & Future Directions

Newborn Screening (NBS) in the U.S.:

- Established in the 1960s, the U.S. has built a comprehensive public health system for newborn screening.
- Nearly all of the 3.6 million newborns each year are evaluated for serious, treatable conditions.
- Each state manages its screening panels independently, often aligning with the federal Recommended Uniform Screening Panel (RUSP), which includes 40 core conditions.
- The system initially focused on phenylketonuria (PKU).
- Over time, it has evolved to incorporate molecular testing, second-tier assays, and long-term follow-up care.



Why Genomic NBS Pilot Studies?

Early detection

Expands detection beyond traditional biochemical screening

Expansion of NBS

- Focus on actionable, childhood-onset disorders
- Intended as a complement —not replacement—to standard NBS
- Evaluates feasibility, ethics, equity, and implementation
- Multiple studies taking place simultaneously world-wide

BeginNGS

(Rady Children's, San Diego)

FIRST GENOME SCREENING PILOT

How it Began:

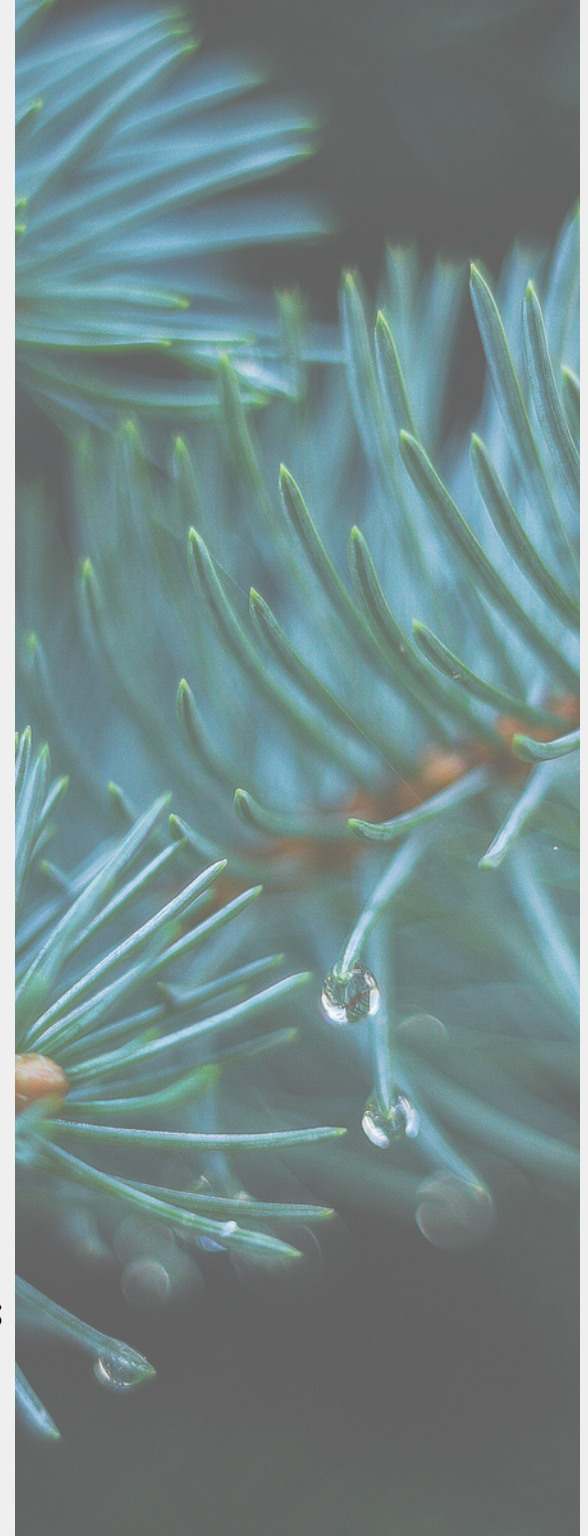
Launched by the Rady Children's Institute for Genomic Medicine in 2015 as a proof of concept of whether rapid WGS (goal of < 24-hour results) could lead to better outcomes in sick infants admitted to NICUs without a definitive diagnosis.

Current Goal:

The clinical success of rWGS paved the way for the development of BeginNGS, a system currently aimed at scaling rapid whole-genome sequencing as a universal, preventative newborn screening tool.

Screening Scope:

Currently evaluates approximately 400+ actionable genetic conditions, focusing primarily on childhood-onset disorders with available interventions



GUARDIAN (Genomic Uniform- screening Against Rare Diseases In All Newborns) Study

(New York, USA)



SCOPE

Uses genome sequencing alongside routine NBS



NUMBER OF CONDITIONS SCREENED

Screening >450 childhood conditions



RECRUITMENT GOAL

Goal up to 100,000 infants



METHOD

Uses existing dried blood spot workflow

Generation Study

(England)

WHO?

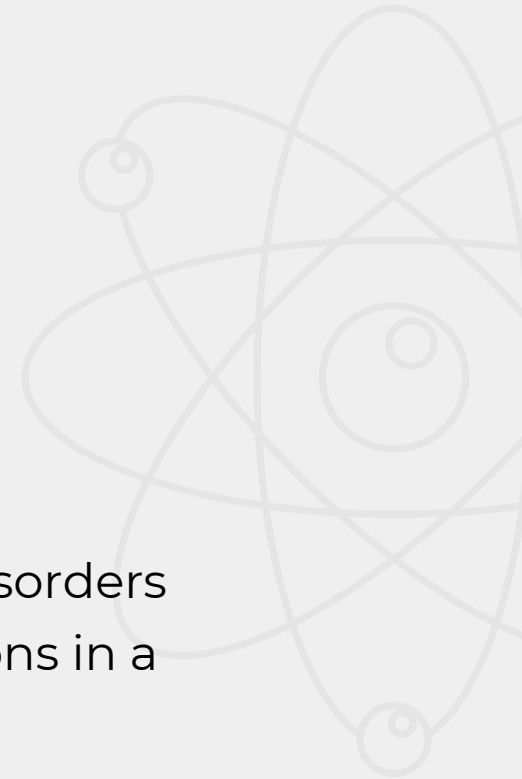
Genomics England + NHS initiative

Enrollment

100,000 newborns

Scope

Evaluates WGS for actionable childhood-onset rare disorders
Also studies long-term feasibility and policy implications in a single payer system.



Early Check

(NORTH CAROLINA, USA)



Voluntary
research
expansion to
state NBS



Parent consent
model



Evaluated
additional
disorders and
genomic
approaches



Important
implementation
model for future
genomic pilots

BEACONs (Baby's Early Assessment and Care through Omics in Newborn Screening)

(Iowa, Minnesota, New York, Oregon, Puerto Rico, South Carolina, and Texas)

Funding

NIH funded multi-state genomic newborn screening research initiative evaluating integration of sequencing and other omics technologies into newborn screening programs

Enrollment Goal

Up to 300,000 newborns during the 3-year project period.

Scope

Initially sought to conduct study across 10 states, but currently only 6 states and one territory are enrolled.

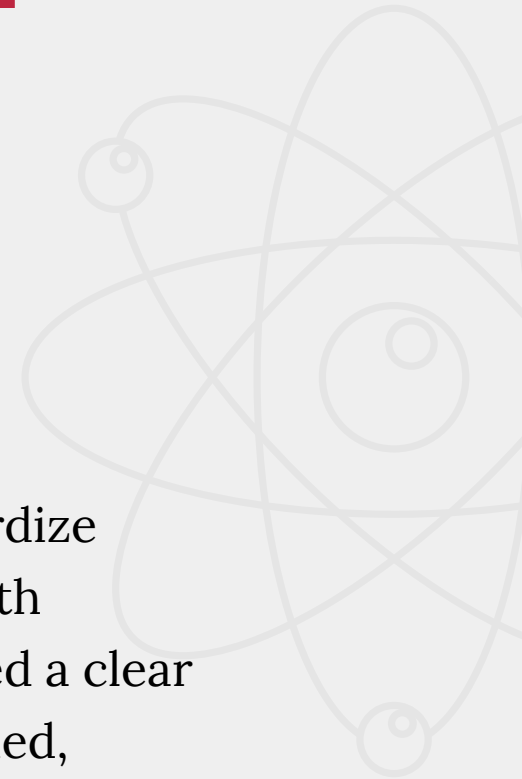
Number of Conditions Screened

777 conditions associated with 743 genes

ICONS (Informatics for Clinical and Public Health Genomic NBS)

Standardization for the future

ICoNS brings together global experts to speed up and standardize newborn genome sequencing so it can become a routine health check. Since launching in 2022, the consortium has established a clear governance structure and now leads the development of unified, evidence-based guidelines for global use.



Key Questions for Public Health



NEW OR EXISTING FRAMEWORK?

Which conditions meet screening criteria?



ECONOMICS

What is the cost, scalability, and workforce impact?



PUBLIC TRUST

Data governance and privacy



ETHICS

- Equity and access



FEASIBILITY

Integration with existing follow-up systems

VI. Intern Presentation on Physician Manual Updates



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VII. Break

VIII. Programmatic Updates (Jillian)



IX. Data Presentation



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X. Other Business





XI. Adjourn

**Next Meeting
October 27,
2026**

**Children's Home
Society Omaha**



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