

Newborn Screening Data Collection Challenges and Opportunities

National Academies Committee Meeting on Newborn Screening: Current
Landscape and Future Directions
May 16, 2024

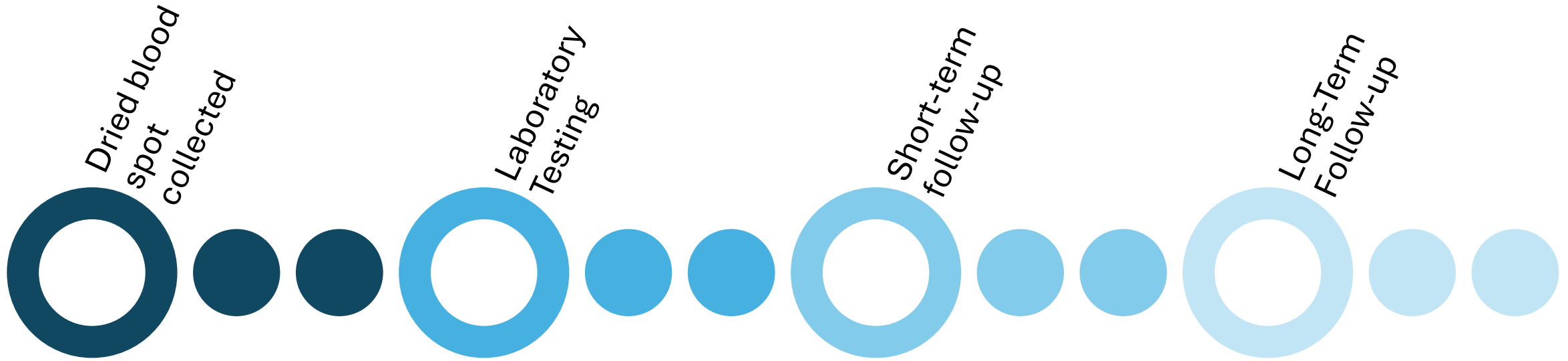
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Introduction

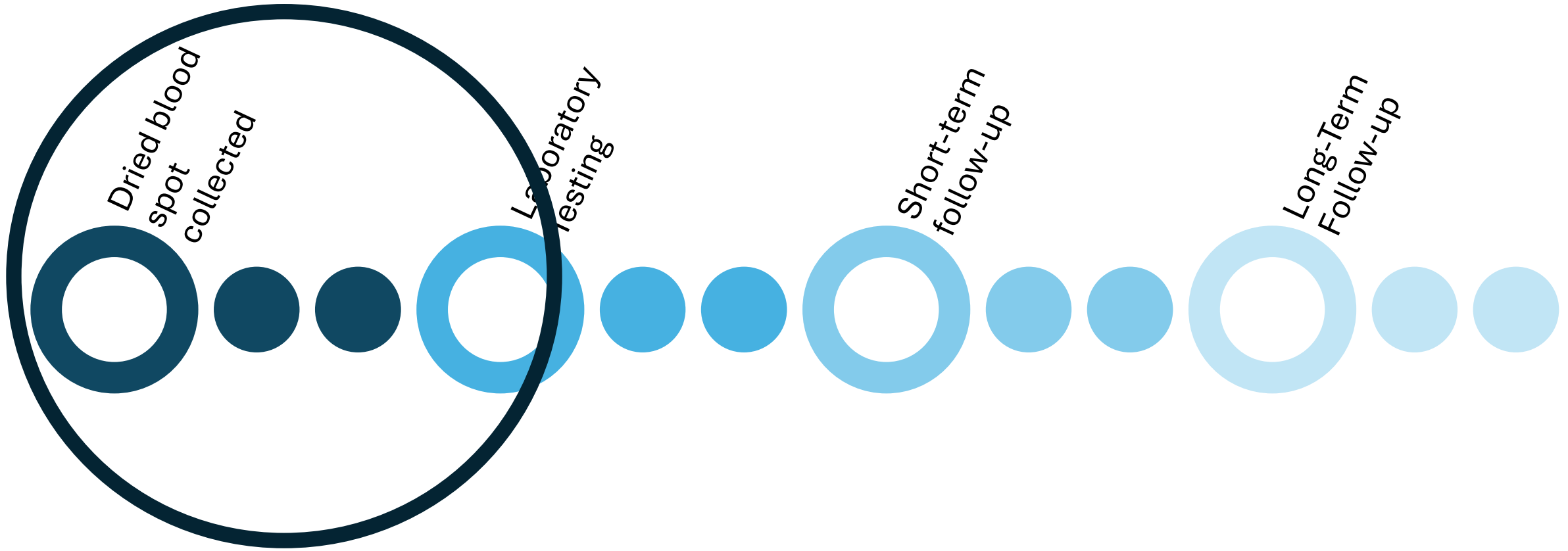
- Description of newborn screening data challenges from dried blood spot collection through long-term follow-up
 - Overview of challenges
 - Ideas for solutions: some that are being implemented (or soon will be), others that are possible
- Foundation of this presentation



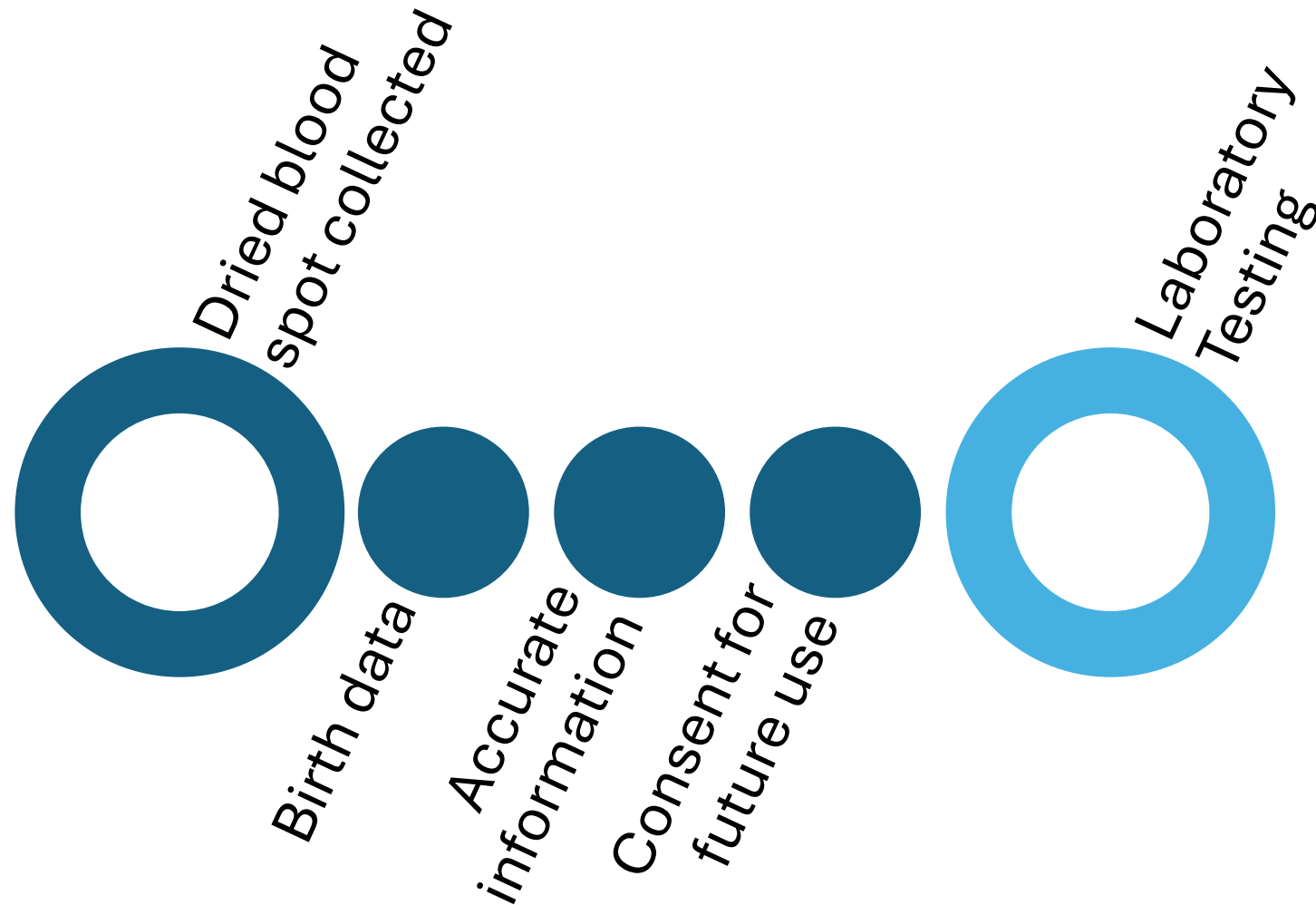
The Spectrum of Newborn Screening Data



Starting at the Beginning



Collection Through Receipt at Laboratory



Specimen and Infant Data



Birth Data

Challenge:

- Unable to assess impact of screen
- Timely true denominator not available

Opportunities:

- Link to Vital Records
- Newborn Admission Notification Information (NANI)



Accurate Information from Specimen Card

Challenge:

- Handwritten specimen cards, Data entry
- Missing information leads to delays
- PH Lab Systems aren't built the same as EHRs

Opportunities:

- Labels with printed data
- Electronic Test Orders and Results (ETOR) – HL7 messages
- Collect data via integrated data systems for demographic, PCP info, contact information



Consent for Future Use

Challenge:

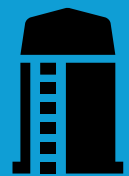
- Recording consent for use for other purposes
- Collecting and tracking

Opportunities:

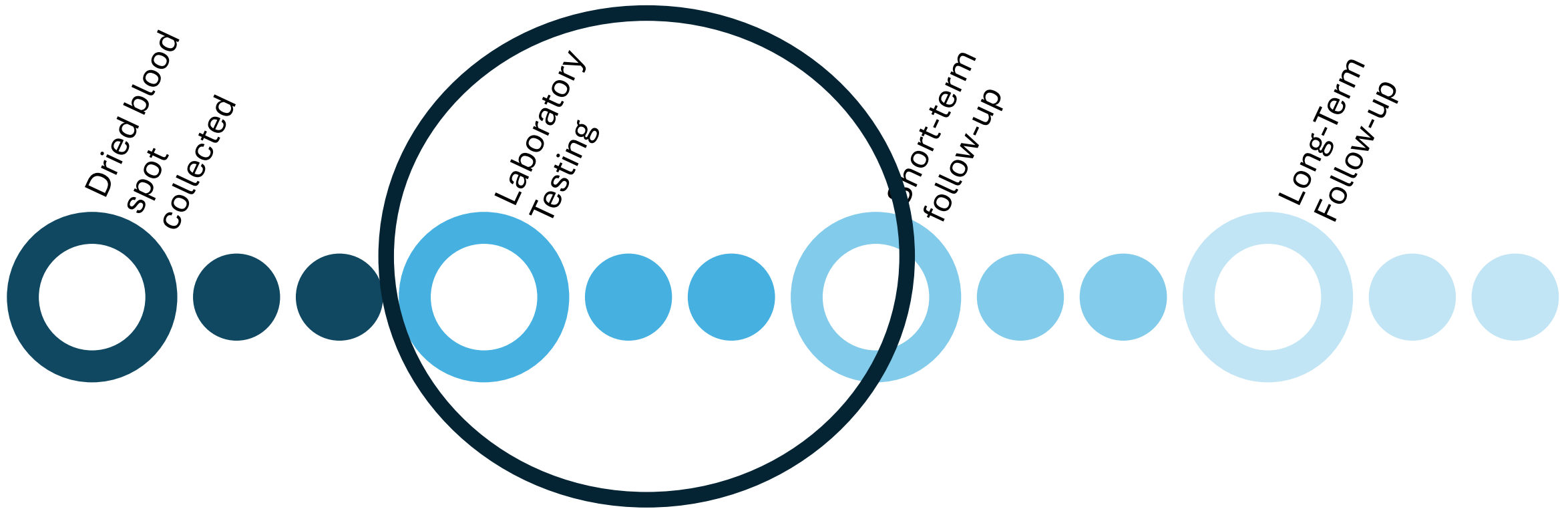
- Electronic consent models

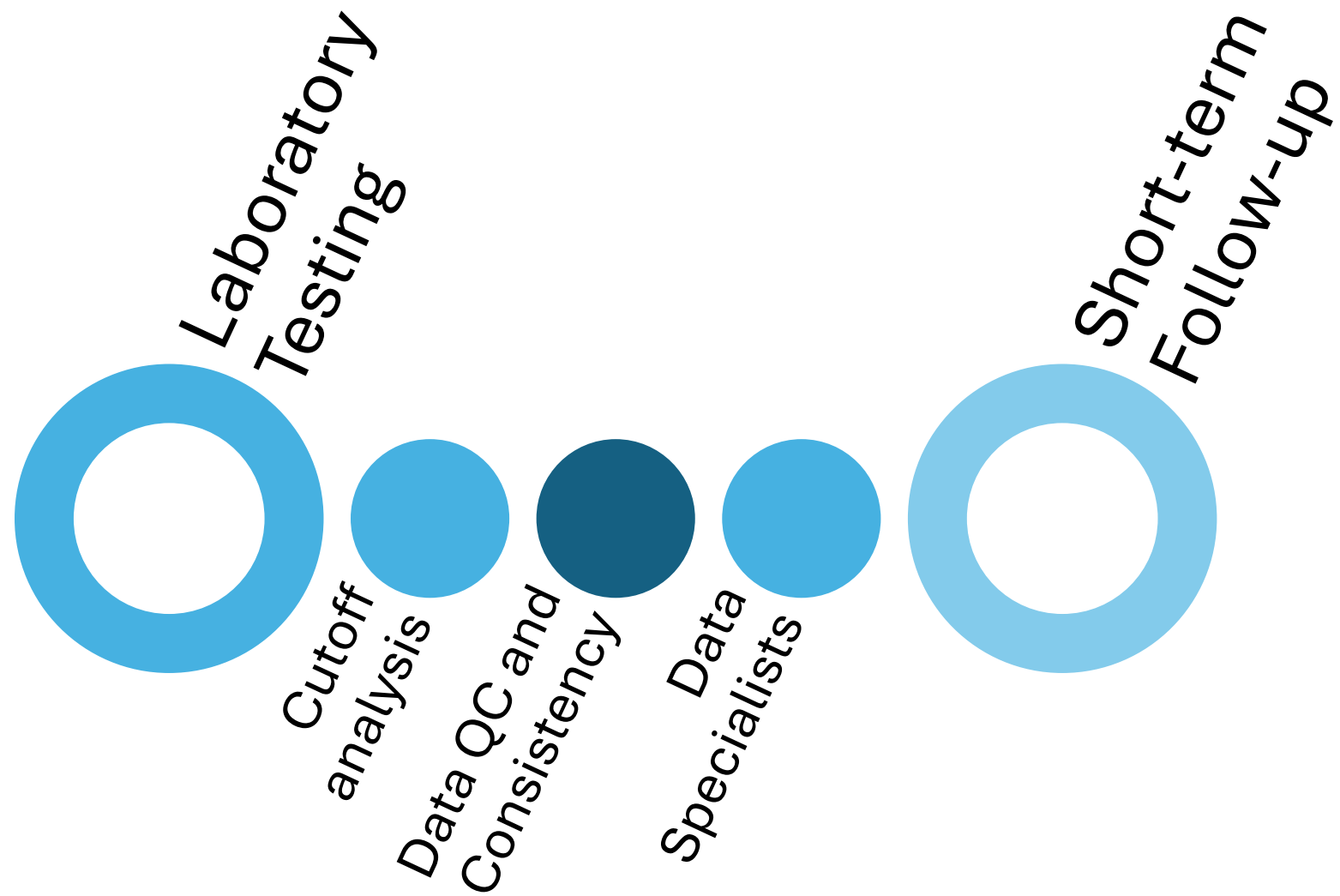
Key Opportunities: Collection Through Receipt at Laboratory

- Need to be able to reliably link to other systems
 - Public Health Newborn Screening Laboratory and Follow-up
 - Electronic Health Records
 - Vital Records
 - Electronic Birth Certificate
 - Vaccination Registries
 - Medicaid
- Broader ETOR implementation
- *These linkages would help other parts of NBS as well*



Specimen Testing at the Laboratory





Specimen Testing and Reporting



Cutoff Analysis

Challenges:

- Small numbers of affected infants
- Complex analytes and ratios
- Different instrumentation
- Numerous covariates

Opportunities:

- CLIR (Collaborative Laboratory Integrated Reports – MAYO)
- dbRUSP (Yale)
- ED3N – Enhancing Data Driven Disease Detection in Newborns (CDC)
- Proficiency Testing Results (CDC)



Data Quality and Consistency

Challenges:

- Differing nomenclature and formats between programs
- Inconsistent data collected across programs

Opportunities:

- Agreement and harmonization of nomenclature
- Result reporting format (results, interpretations)
- Application of consistent data elements



Qualified Data Specialists

Challenges:

- Lack of qualified NBS data specialists
- Data specialists, epidemiologists, and informaticians often shared across public health programs in a state

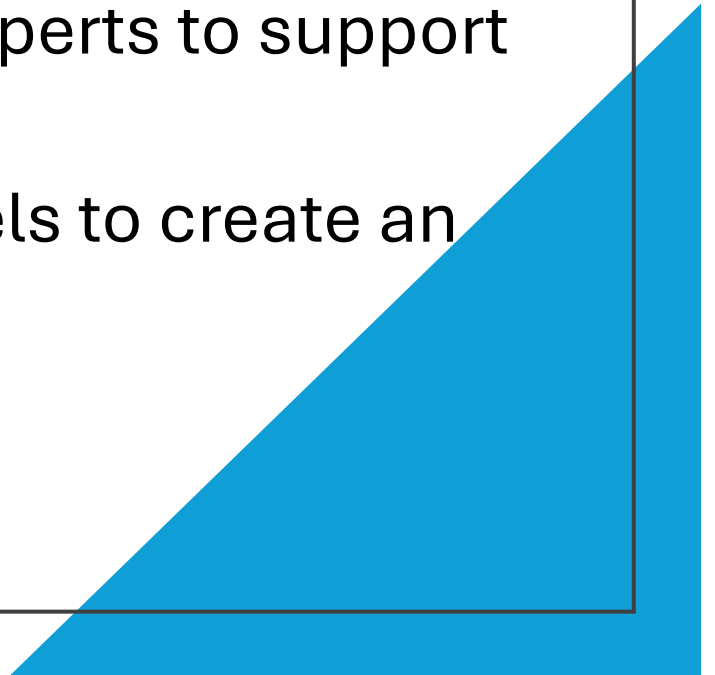
Opportunities:

- Funding for data specialists and training for NBS data
- Shared regional NBS data experts
- ED3N
- Technical assistance for data analysts

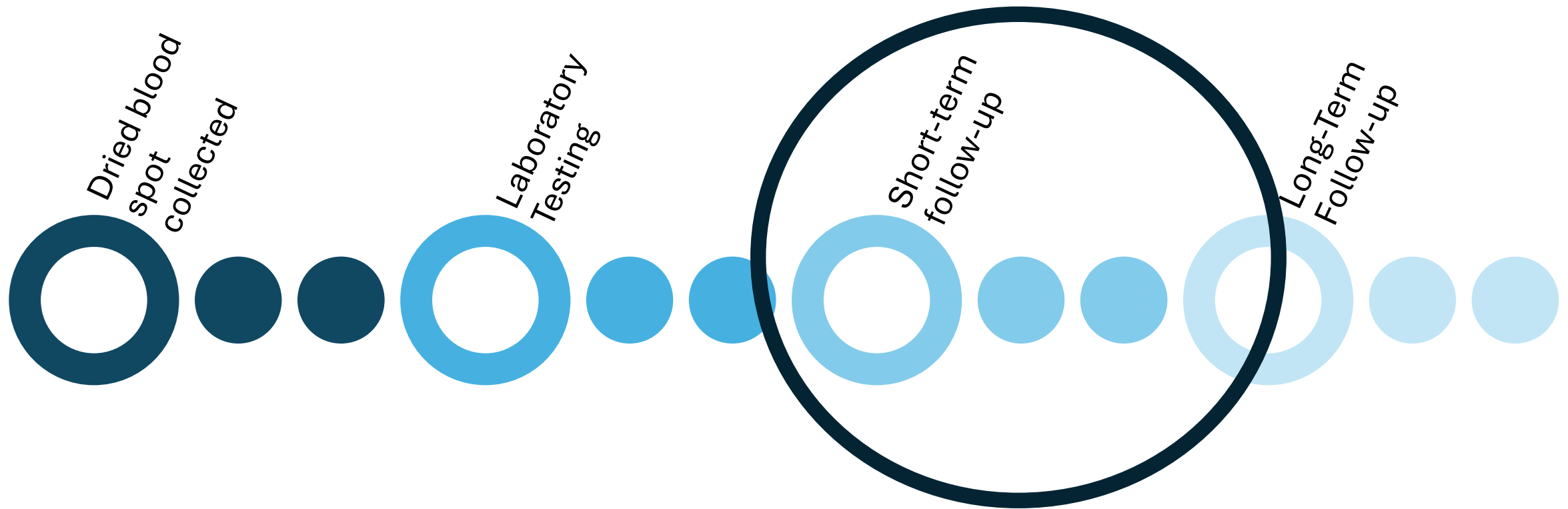


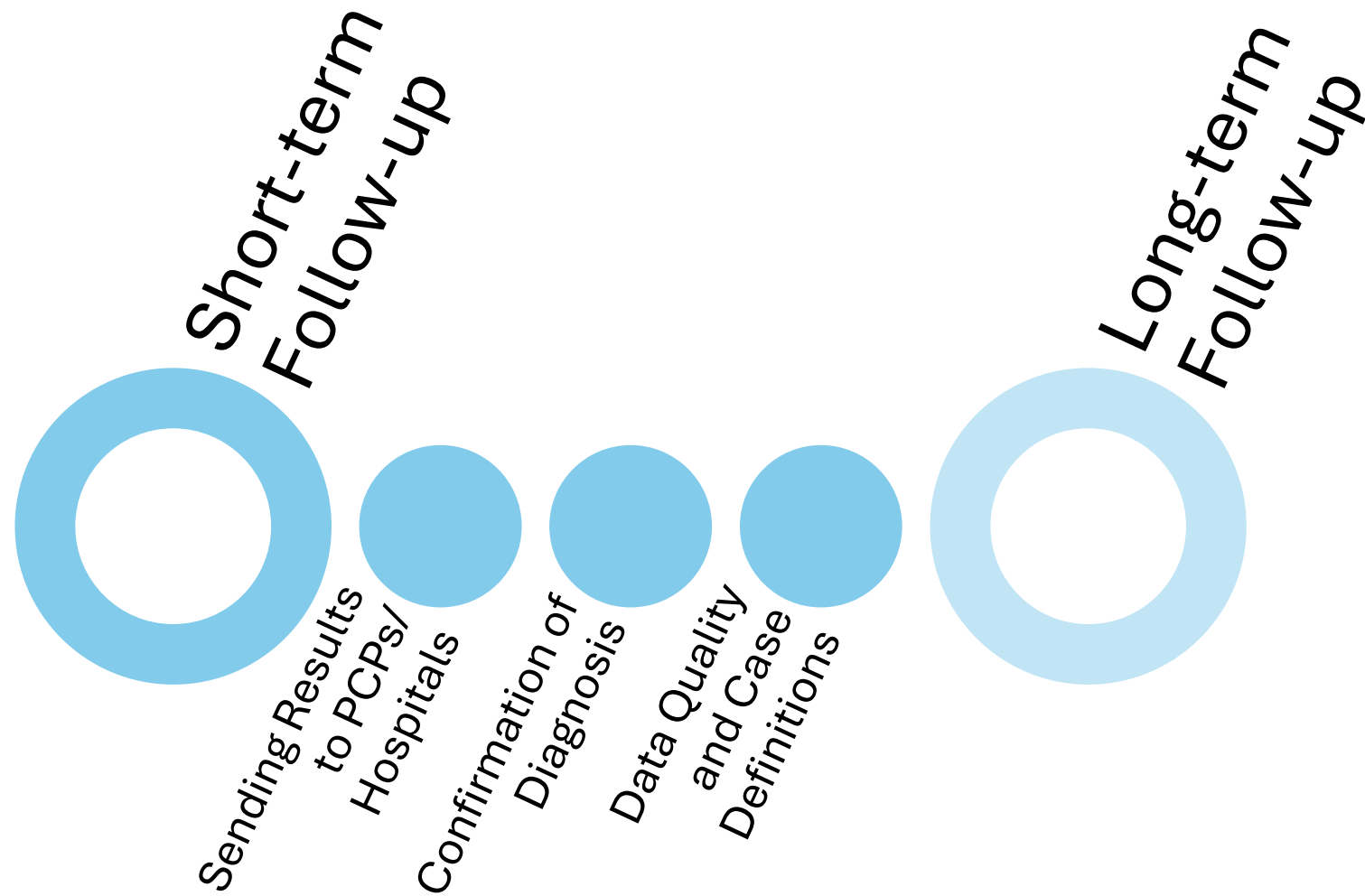
Key Opportunities: Laboratory Testing

- Continue to support and build national data analytic efforts
- Build a network of newborn screening data experts to support states
- Build upon existing definitions and data models to create an agreed upon data set



Result reporting and short-term follow-up





Short-term Follow-up



Sending Results to PCPs/ Hospitals

Challenges:

- Identifying the right PCP
- Ensuring reports are received and acted upon

Opportunity:

- Electronic Test Orders and Results (ETOR)
- Web portals of results



Data Quality and Consistency

Challenges:

- Differing nomenclature and formats between programs
- Case definitions are not consistently applied

Opportunities:

- Agreement and harmonization of nomenclature
- Result reporting format different
- Application of consistent case definitions



Confirmation of Diagnosis

Challenges:

- Hard to get case data from PCPs
- EHRs and HIEs are not integrated with follow-up systems, requiring manual communication
- Some children are lost to follow-up before diagnosis
- Some disorders may take years to resolve

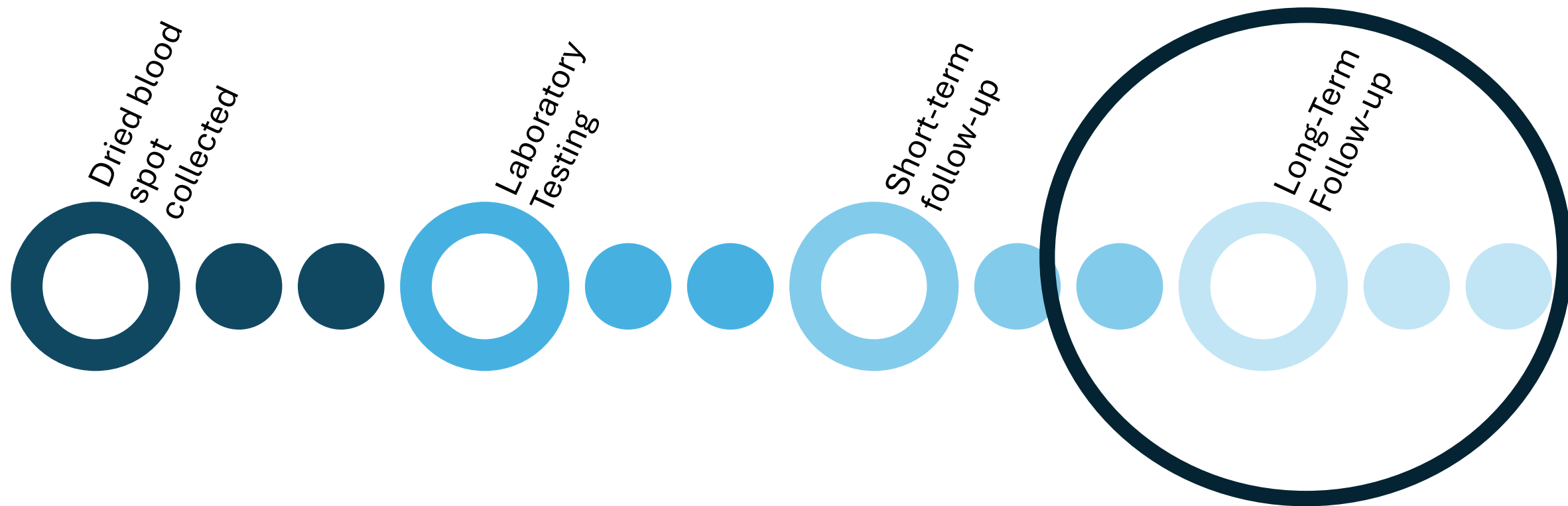
Opportunities:

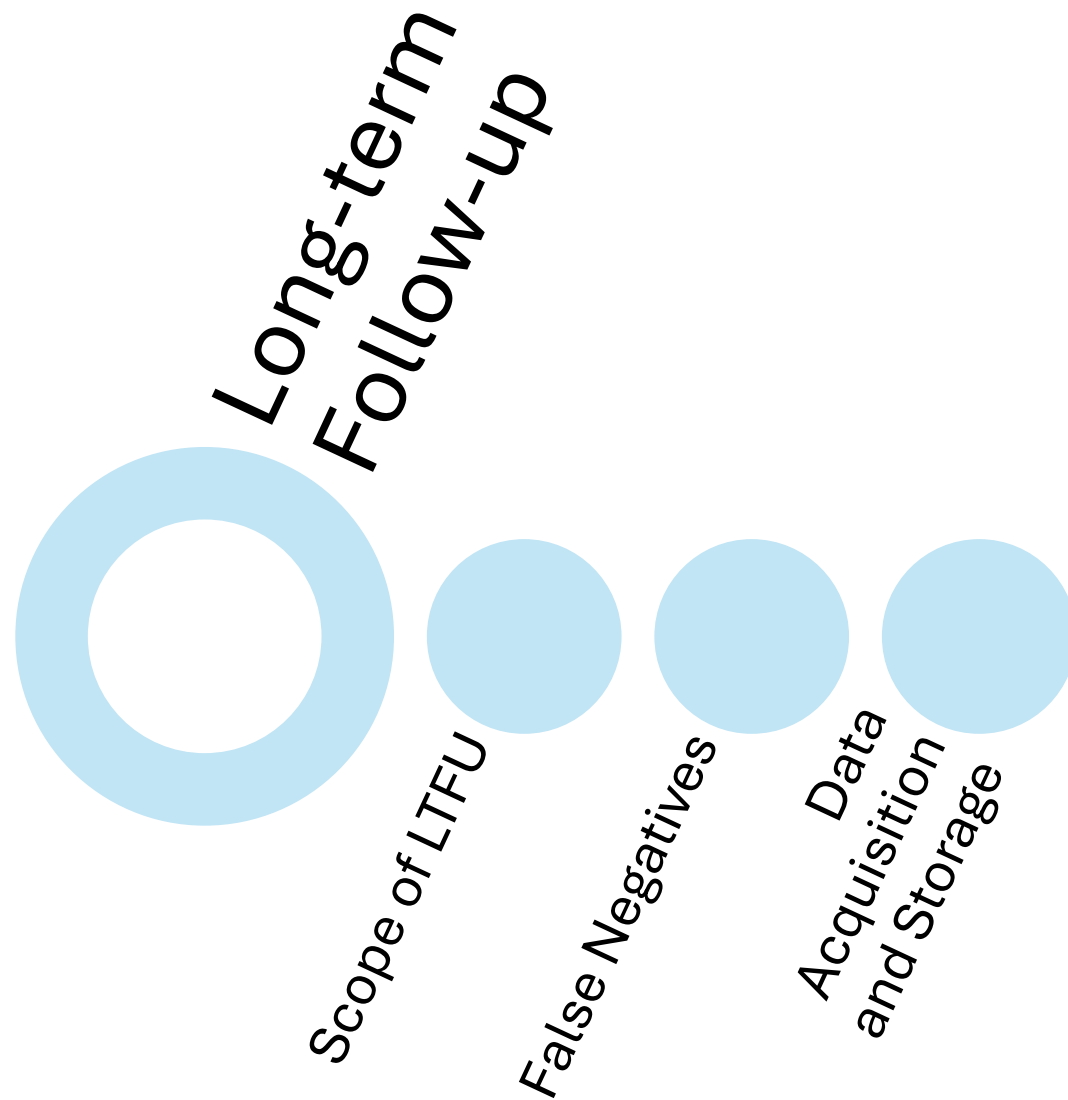
- Utilize data integration tools: eCR, FHIR queries to get case data
- Update case management systems
- Data portals that are shared by public health and PCPs

Key Opportunities: Short-term follow-up

- Build upon existing definitions and data models to create an agreed upon set
- Implement shared case management systems between specialists and NBS program
- Capitalize on current data modernization initiative efforts to support newborn screening
 - eCR, FHIR Queries, QHINs, Data portals







Long-term Follow-up

Scope of LTFU

Challenges

- Families who aren't engaged in clinical care are missed by many data systems
- What data are needed to ensure we meet goals? Equity?
- Who has the responsibility of LTFU?
- What has authority?
- Who is going to monitor and analyze the data?

Opportunities:

- Build upon successes of HRSA awardees and efforts of HRSA Propel and soon to be funded Co-Propel grantees.
- Seek collaborations with others in PH
- Partner with clinical specialists to build LTFU network, capture QOL outcomes



False negatives

Challenges:

- Identifying false negatives and reporting them to Public health programs is based on partner networks

Opportunities:

- Capitalize on data modernization opportunities (e.g., apply eCR for case notification of late diagnoses)



Data Acquisition and Storage

Challenges:

- Some states don't have authority for LTFU data
- Consent for collection vs. public health authority
- Data extraction, collection, storage are hard to sustain

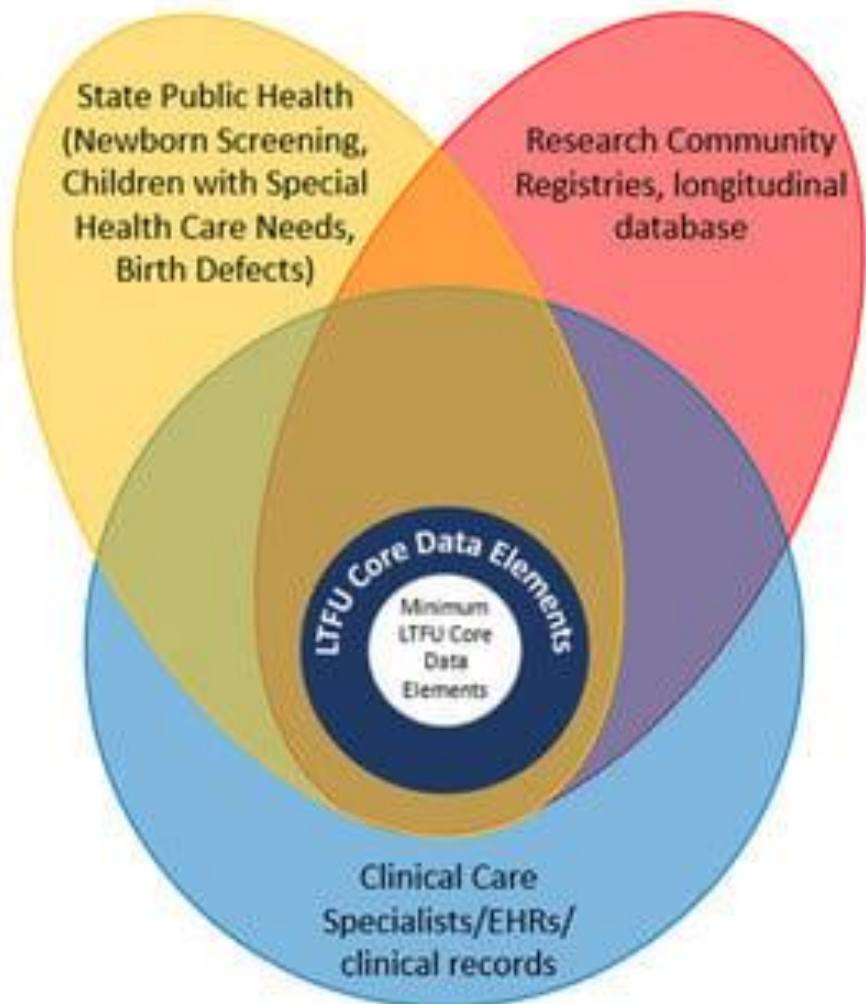
Opportunities:

- Consider alternatives for LTFU data storage (birth defects, hospital systems, EHR modules)
- Start with minimal data sets and build from there
- Use DMI/interoperability opportunities to get data, minimize data entry, and improve timeliness, consistency, improve equity



CPHI

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State Public Health LTFU Questions (examples)

- Treatment? (Yes/No; Reason reported for no treatment in the last 12 months?)
- Alive? (Cause of death: injury, medical, unknown, unasked, other, other)
- Developmentally appropriate milestones? (Yes/no/unknown/unasked by category)
- Healthcare utilization: How many ER visits in the last 12 months? How many hospitalizations in the last 12 months? How many clinic visits in the last 12 months?

Clinical LTFU Questions (examples)

- Clinical trial participation
- Quality of Life
- Patient Reported Outcomes

Research LTFU Questions (examples)

- Disorder specific outcomes
- Demographics
- Medical home status
- Family satisfaction?

Suggested NBS LTFU Core Data Set*

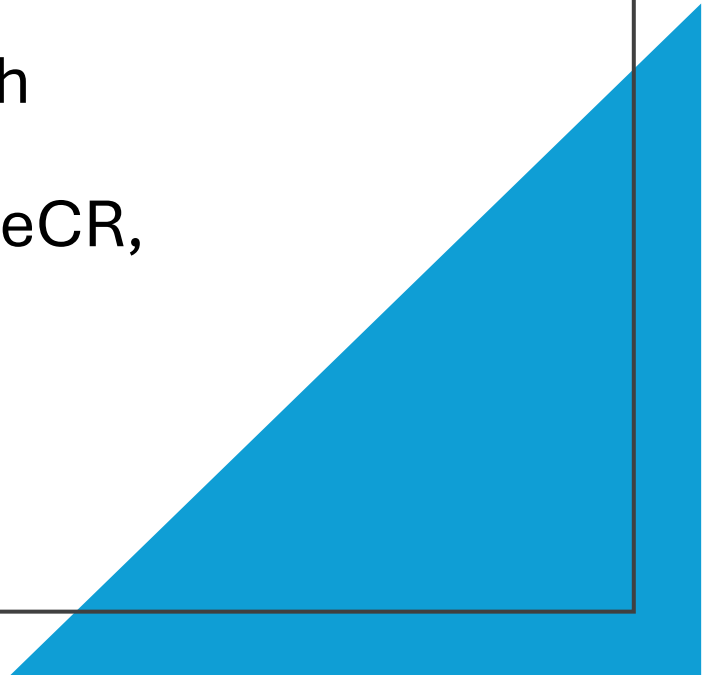
- Diagnosis
- Date of appropriate first intervention
- Alive?
- Within the last 12 months did the child receive care and treatment specific to the diagnosis? Type of care provider?

* Suggested by a workgroup of NBS professionals organized by the National Coordinating Center (NCC) of the Regional Genetics Network and the Newborn Screening Translational Research Network (NBSTRN), 2016.

Kellar-Guenther, Y.; et al *Int. J. Neonatal Screen.* 2024, 10, 37.

Key Opportunities: Long-term Follow-up

- Utilize knowledge of current programs building LTFU systems, partner with experts in data collection, quality of life, equity
- Build on existing data opportunities for LTFU: Birth Defects, Clinical Systems
- Capitalize on existing and new DMI applications: eCR, FHIR Queries



Key Partnerships to Support Data Activities



Conclusions

- The data challenges are numerous and span the breadth of the system
- Opportunities are also numerous, have been proven, yet complex to implement
- States NBS programs solving problems one at a time is onerous and slow. Need systems level, collaborative solutions

Thank you

Acknowledgements:

- Members of the newborn screening community who responded to my queries

Contract information:

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Select References

- Case definitions:
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- Electronic Case Reporting: <https://www.cdc.gov/ecr/index.html>
- ED3N: <https://www.cdc.gov/nceh/dls/ed3n.html>
- Special Issue on CLIR applications in newborn screening:
https://www.mdpi.com/journal/IJNS/special_issues/clir
- Long-term follow-up
Hinton, C. F., et al *Genet Med* 16, no. 6 (2014): 484-90.
Hinton, C. F., et al *Mol Genet Metab* 118, no. 4 (2016): 221-9.
Kellar-Guenther, Y.; et al *Int. J. Neonatal Screen*. **2024**, 10, 37.