

ED3N: Enhancing Data-driven Disease Detection in Newborns

Better Data. Better Decisions. Healthier Newborns.



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Creating a Vision to Enhance Data Analysis

Laying the Foundation for ED3N

Anticipating Domestic NBS Program Needs

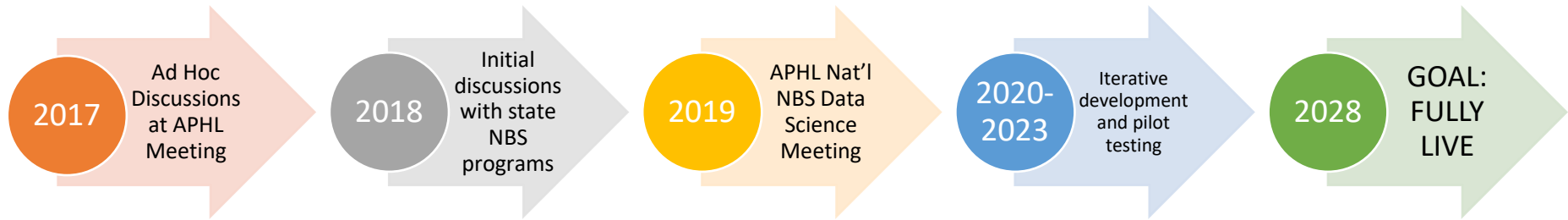


What do current challenges tell us about
what we should expect of the future?

- More diseases on RUSP, increased complexity, nuanced risk assessment
- New or more sensitive testing platforms and emerging technologies
 - multiplexing, metabolomics, NGS
- Increased data generated

**The Best Way to Predict the
Future is to CREATE IT.**

The Long Road to ED3N



Identified Gaps in Newborn Screening Programs

- Lack of harmonization between state programs in testing practices and data output and capacity
- Inadequate number of data analysts and interoperability specialists within NBS programs
- Disparate ability and resources to review and analyze screening data to improve performance
- Data silos and one-off connections within and between NBS programs and other relevant health programs

2019 National Data Analytics Meeting

Programs were Supportive of ED3N Development as a possible solution

79%

Feel it is important to have a NBS Data Platform

N=61

67%

Feel they will utilize a NBS Data Platform at least weekly

N=58

88%

Feel the NBS Data Platform should be housed at CDC

N=53

96%

Feel case level clinical data should be in the Data Platform

N=53

ED3N: *Enhancing Data-driven Disease Detection in Newborns*

A nationally representative NBS Platform to support Decision-Making

Aims to:

- **Improve detection of at-risk newborns**, allowing for more timely diagnosis and intervention for an increasing number of diseases
- **Decrease disparities** across state NBS programs and family experiences

Achieved By:

- **Increasing capacity and infrastructure** to collect, aggregate, and analyze newborn screening data across federal, state, and healthcare systems
- **Providing a national data solution** to remove burden on NBS programs and ensure equal access to high-level data analytics

CDC's Data Modernization Initiative

ED3N selected for Accelerated Modernization

Data saves lives. Better data saves more lives.

TOGETHER, WE ARE FOCUSED ON THE
DATA · PEOPLE · POLICIES
WE NEED TO *MOVE THE COUNTRY FORWARD*



“At CDC and throughout public health, we are in a pivotal moment for data and surveillance — one marked by opportunities, challenges, and the need for change.”

Dr. Rochelle P. Walensky, CDC Director

The ultimate goal of CDC's Data Modernization Initiative (DMI) is to get better, faster, actionable insights for decision-making at all levels of public health.

Our vision is to create one public health community that can engage robustly with healthcare, communicate meaningfully with the public, improve health equity, and have the means to protect and promote health.

Accelerated Modernization of Select Systems

Two systems from each Non-Infectious Disease Center have been designated for accelerated modernization.

**Enhancing Data-driven Disease
Detection (ED3N)
Is one of eight Systems selected.**

ED3N Structure

ED3N

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Enhancing Data-driven Disease Detection in Newborns (ED3N)

ED3N serves as a secure, centralized resource for newborn screening partners to analyze and share biochemical and molecular data. ED3N supports laboratory best practices through standardized workflows and robust, validated analytical tools.



Evaluate

Routine testing workflows for decision support

View and analyze your newborn screening data at the individual patient level to enhance disease detection.



Explore

Data repository for exploring correlations and trends

Explore de-identified, aggregate data. Ask what-if questions. Try out new workflows before they are validated and moved to the evaluate module.



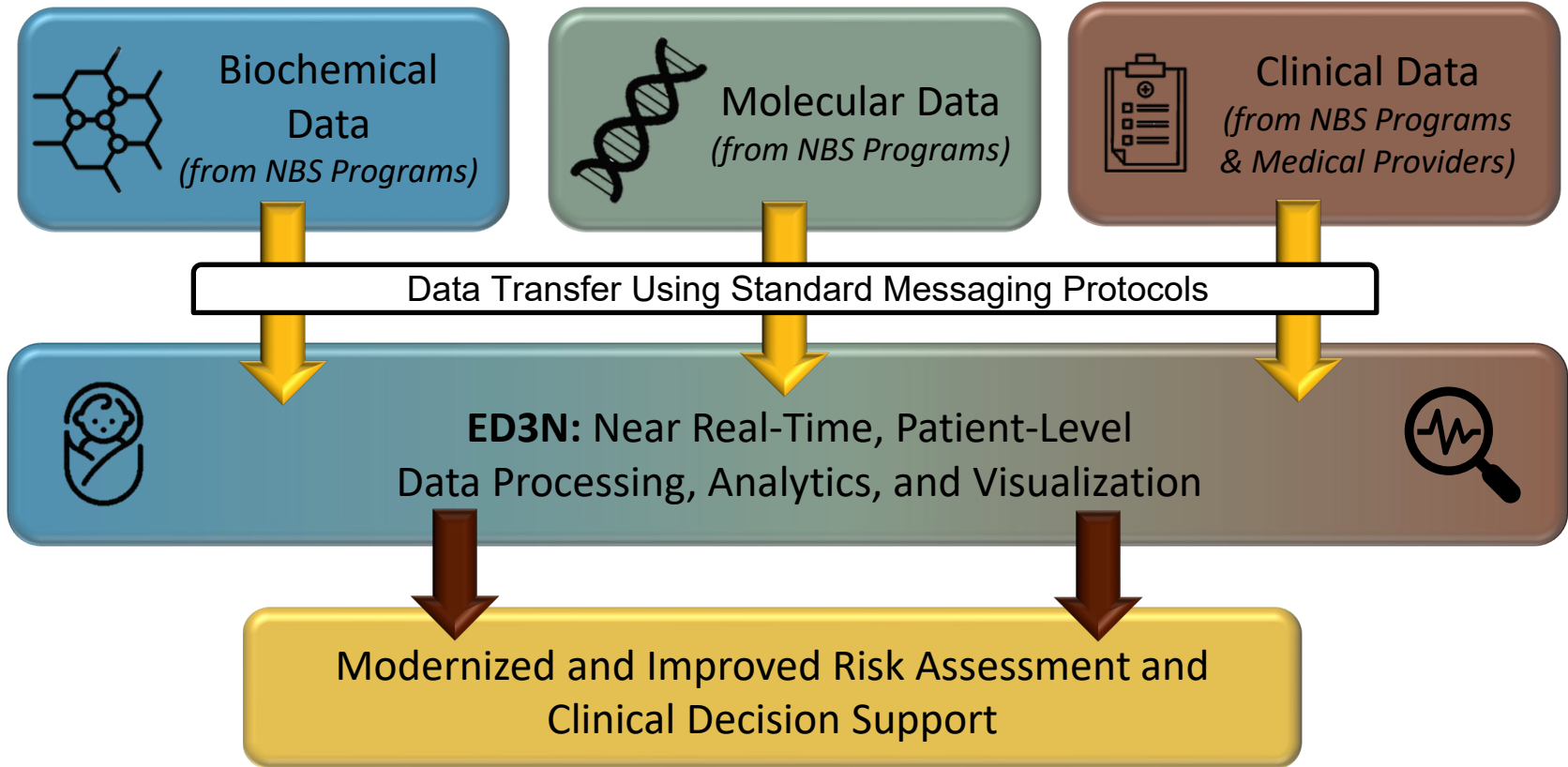
Educate

Helpful tutorials and training materials

Learn how to navigate ED3N. Improve your data analysis skills. Find materials to train newborn screening personnel in best practice workflows.

ED3N Can Lead to Improved Public Health Decision Support

Proposed high-level structure and workflow



Proposed Molecular Module

IDENTIFIED CHALLENGES IN MOLECULAR ANALYSIS

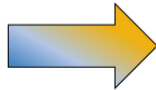
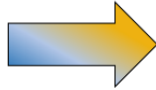
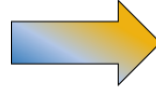
- Identification and curation of variants across NBS programs to inform interpretations



- Lack of access to and maintenance of a collaborative database of interpreted variants



- Inability to link molecular, biochemical, and clinical data across NBS programs



PROPOSED SOLUTIONS IN IN ED3N

- Guided NBS-specific variant interpretation, re-interpretation, and reporting tool



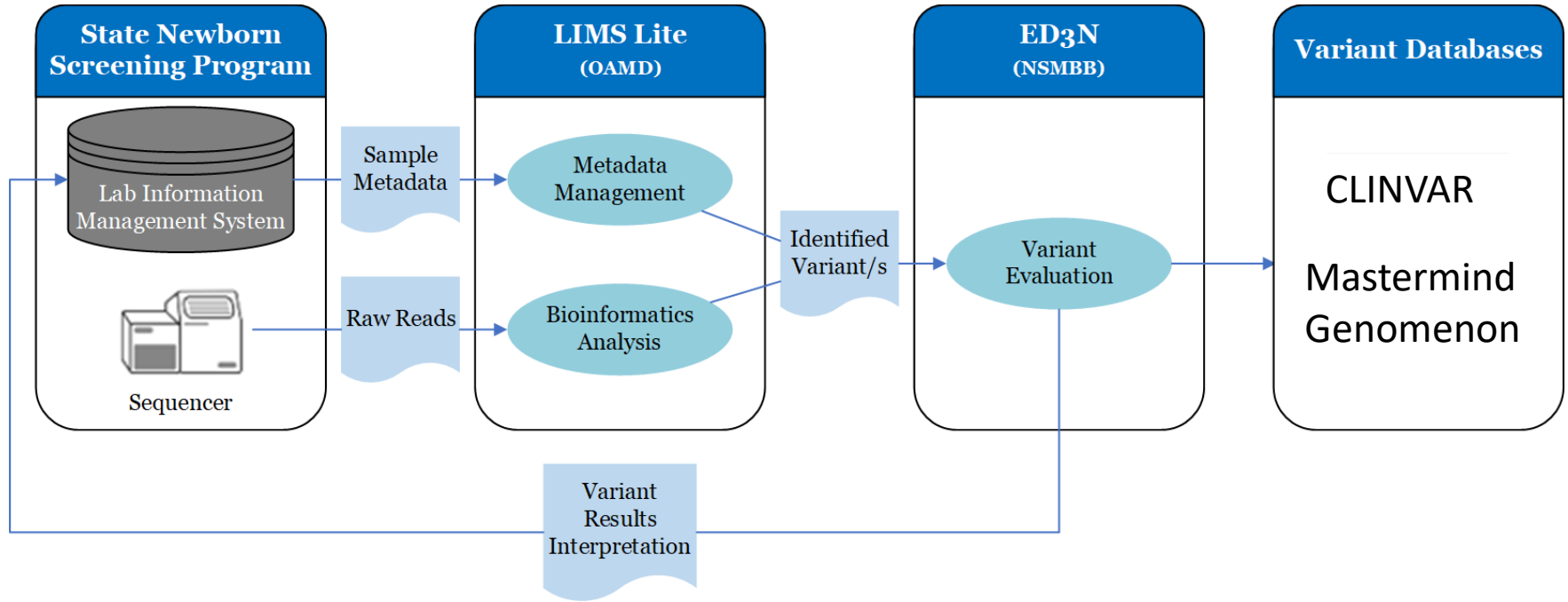
- A collaborative platform to interpret variants and compare across NBS programs



- Secure pipeline to support querying of variant data in conjunction with other NBS data



Molecular End-to-End Solution in ED3N



Proposed Biochemical Module

IDENTIFIED CHALLENGES IN BIOCHEMICAL ANALYSIS

- Variability in cut-off determination
- Challenges in harmonizing data



- Rarity of diseases limit ability to develop robust detection algorithms within each program



- Minimize false negative cases while keeping false positive rate low



PROPOSED SOLUTIONS IN ED3N

- Apply data harmonization techniques to allow comparability between programs



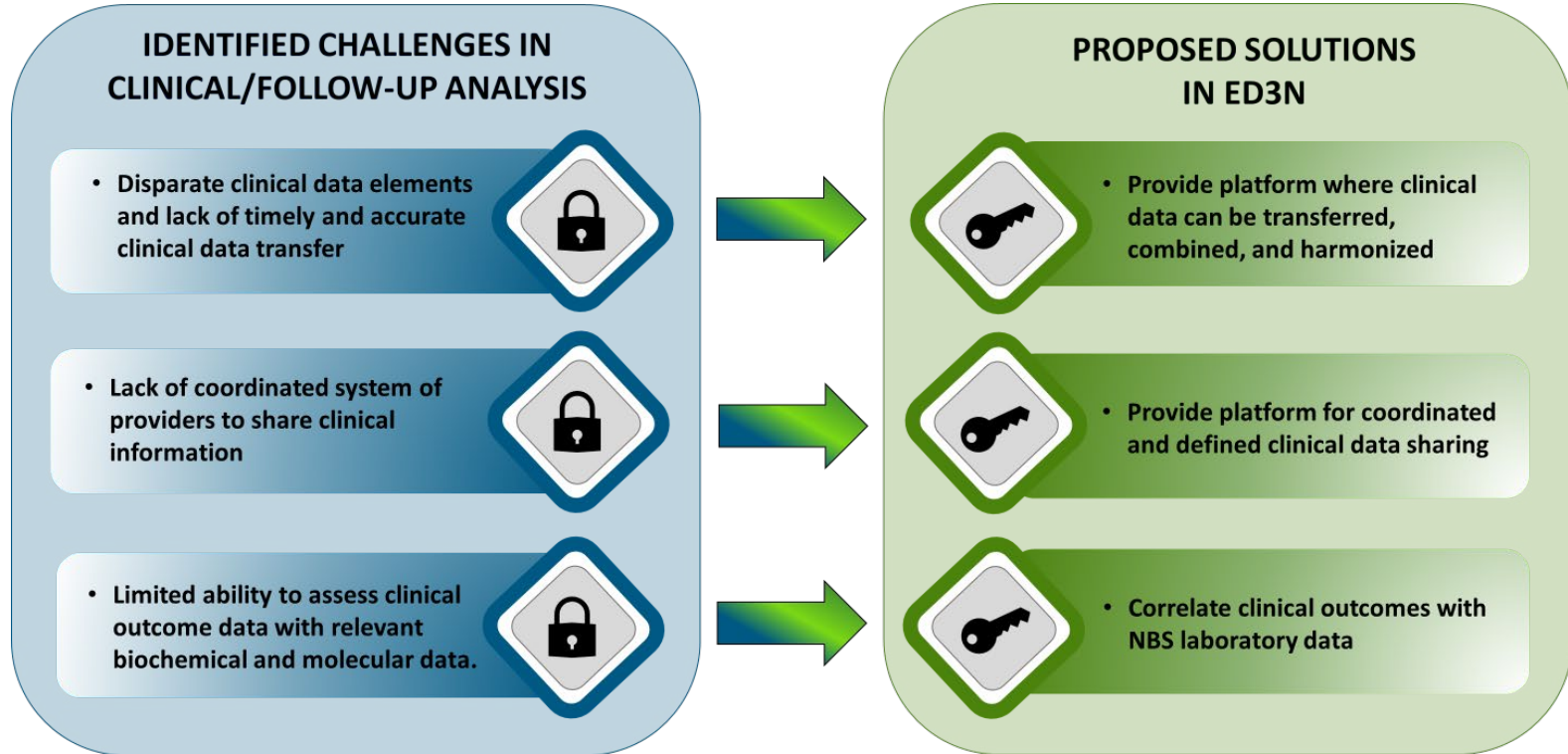
- Improve risk assessment for rare diseases with secure data sharing, analysis and visualization



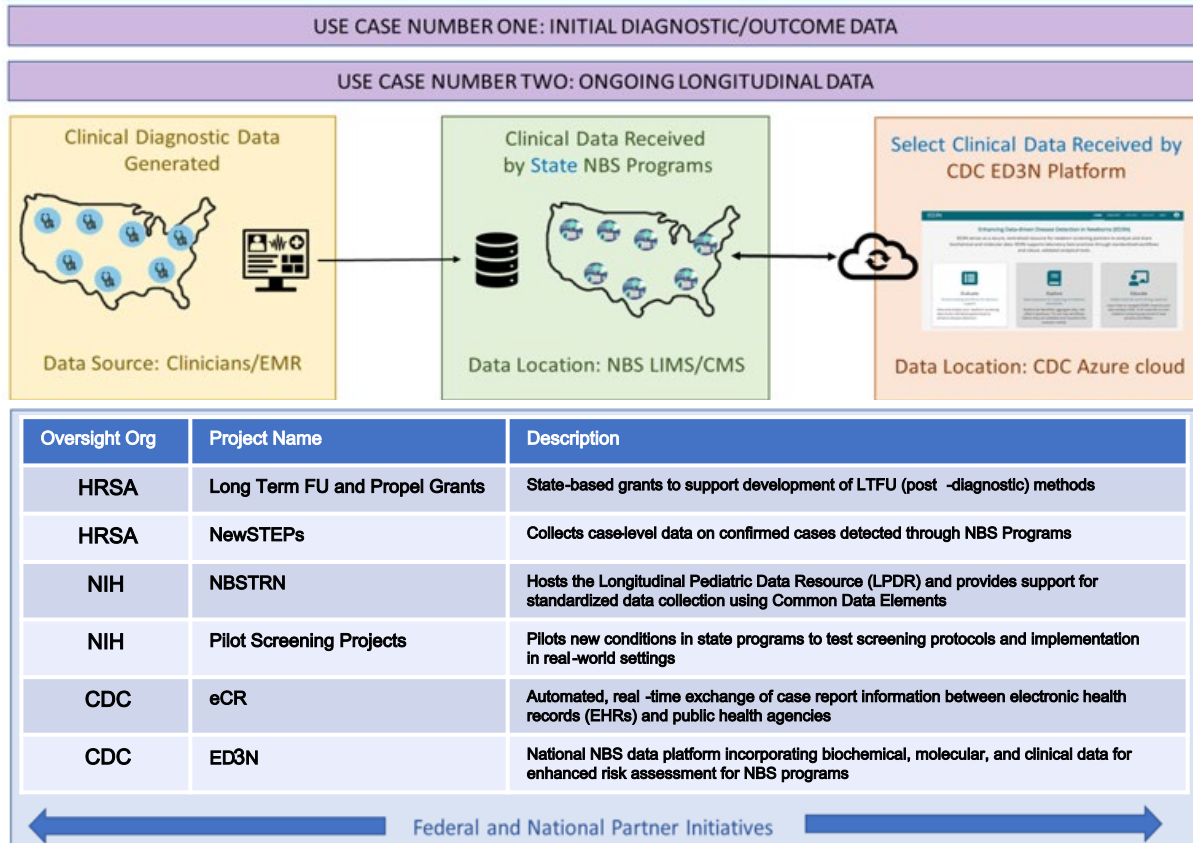
- Apply machine learning models to improve screening performance and effectiveness



Proposed Clinical Module



Developing a sustainable follow-up data collection model



Data Use and Privacy

- **Data Use Agreements**

- Approved by CDC's Office of General Council
- In ratification process with identified NBS pilot programs
 - < 9 for each of biochemical and molecular pilots

- **Paperwork Reduction Act**

- Approved as of April 2023

- **Privacy-Preserving Record Linkage**

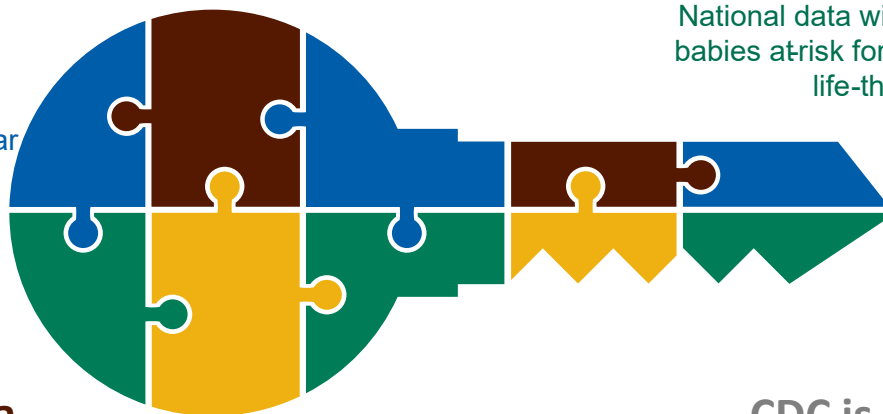
ED3N: The Key to NBS Data Modernization

3,800,000 babies/year

More than 1 in 550 babies affected with NBS diseases each year.

Over 60 diseases currently need enhanced screening and surveillance

Nearing 500 million data points across disparate data sources each year



Aggregated Data Analytics = Enhanced Disease Detection

Novel predictive analytics using aggregated National data will allow for better detection of babies at risk for an increasing number of rare, life-threatening diseases

A National NBS Data Infrastructure is Needed

Individual programs are unable to build and sustain robust data transfer and analysis mechanisms on their own – leading to risks for more false positive and false negative results

CDC is a Trusted Host for NBS Data Modernization

NBS Programs are looking to CDC to take the lead on an interoperable data solution to address increased complexities in NBS

Thank You!

Questions?



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For more information, contact NCEH

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