Innovations in Long-Term Follow-Up Panel

Data Tools and Resources from the Newborn Screening Translational Research Network

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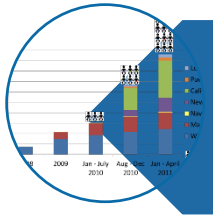
Hunter Kelly Newborn Screening Research Program

Identify, develop, and test the most promising screening technologies

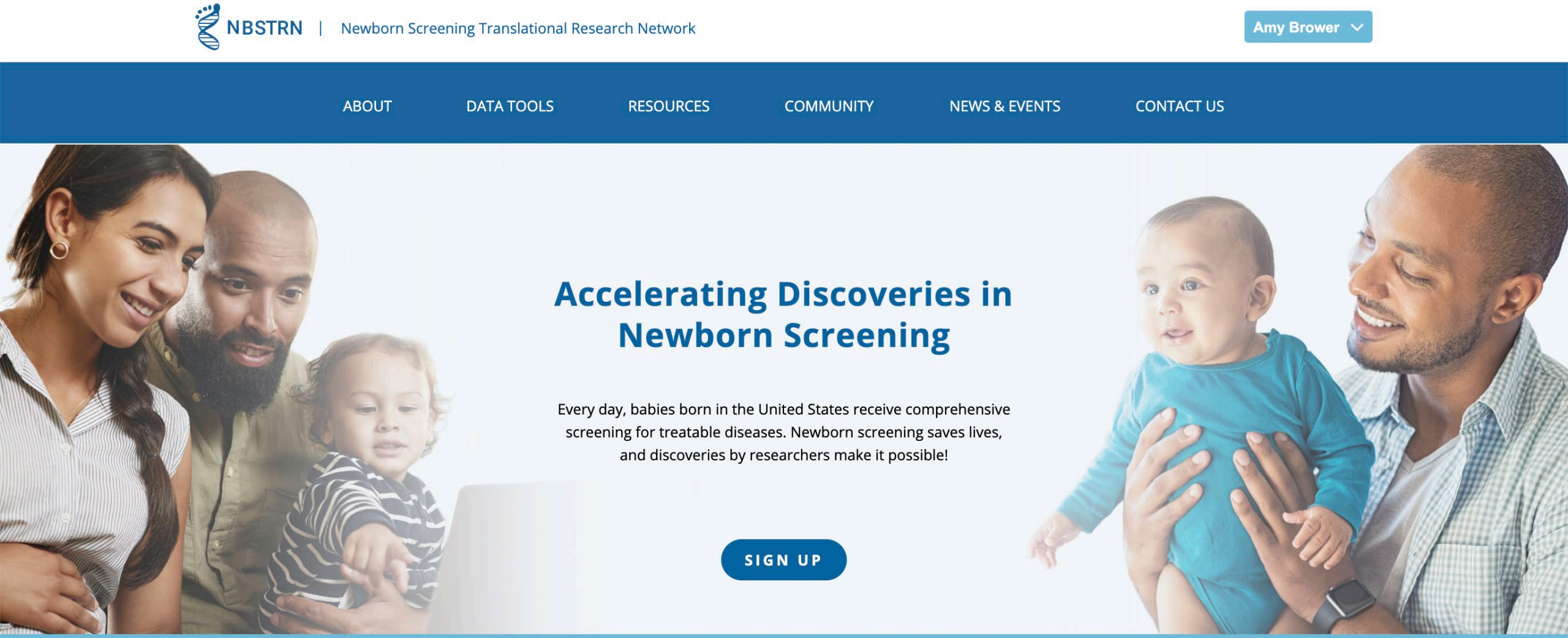
Develop treatments and management strategies for conditions that can be detected through NBS



Generate and provide research findings and data for conditions under review by the ACHDNC

Conduct pilots of conditions recommended or candidates for pilots or nationwide screening

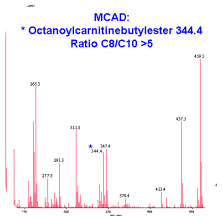
NBSTRN is a key component, operated by the American College of Medical Genetics and Genomics



NBSTRN leverages the multi-component, multi-stakeholder system of NBS to facilitate research

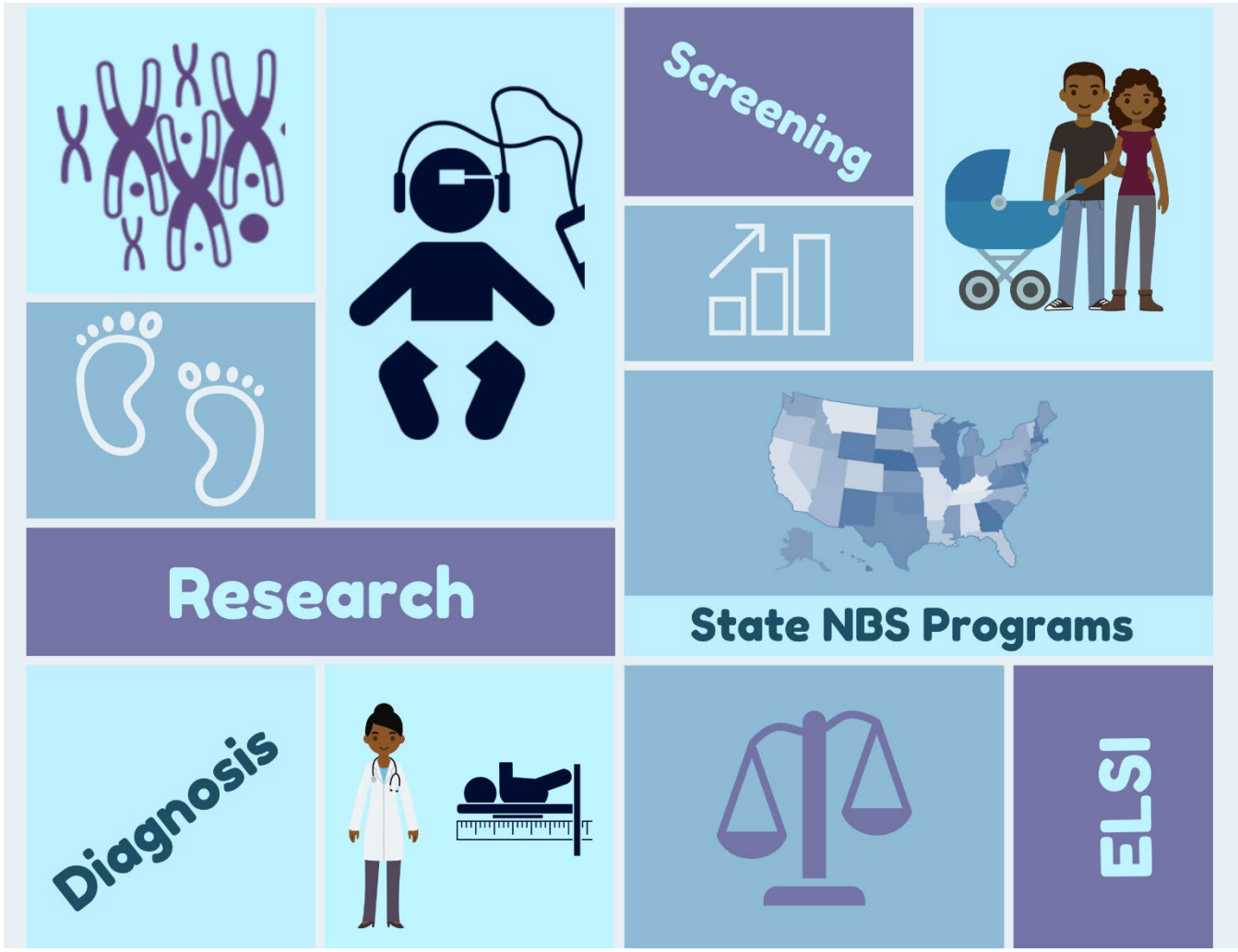
Prenatal Engagement

Neonatal Screening



Diagnosis and Clinical Assessment

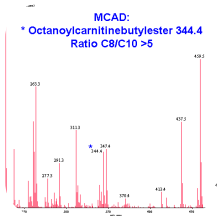
Clinical Care and Longitudinal Follow-Up



The NBS system offers a unique opportunity to conduct long-term follow-up

Prenatal Engagement

Neonatal Screening

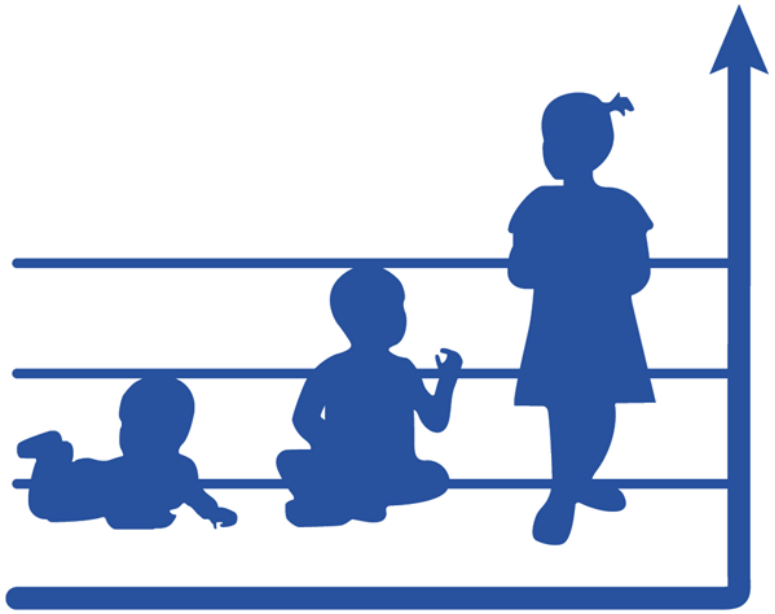


Diagnosis and Clinical Assessment

Clinical Care and Longitudinal Follow-Up

Clinical Care and Longitudinal Follow-Up

NBSTRN worked with researchers, healthcare professionals, families, advocacy groups, state newborn screening programs, and partners to create a data tool to facilitate long-term follow-up.



LPDR

Longitudinal Pediatric Data Resource

The ability to capture clinical information early in the clinical course of a disease, even before clinical symptoms appear, advances disease understanding, helps to establish the efficacy of new treatments and management approaches, informs the community at large about the value of early identification and treatment through newborn screening, and identifies areas for improvement in disease management throughout the lifespan.

Informs Assessment of the Benefit of Early Identification Through Newborn Screening

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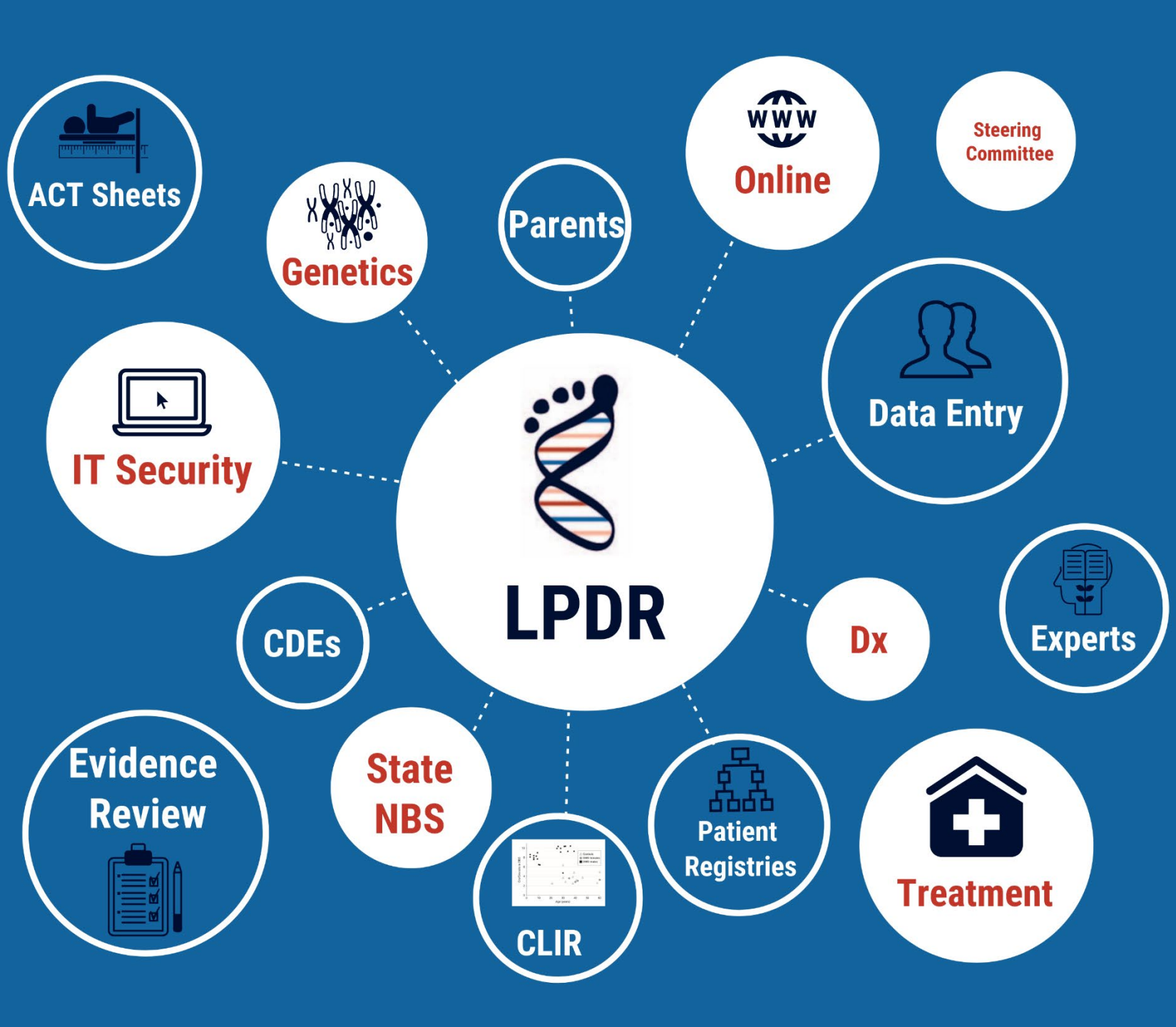
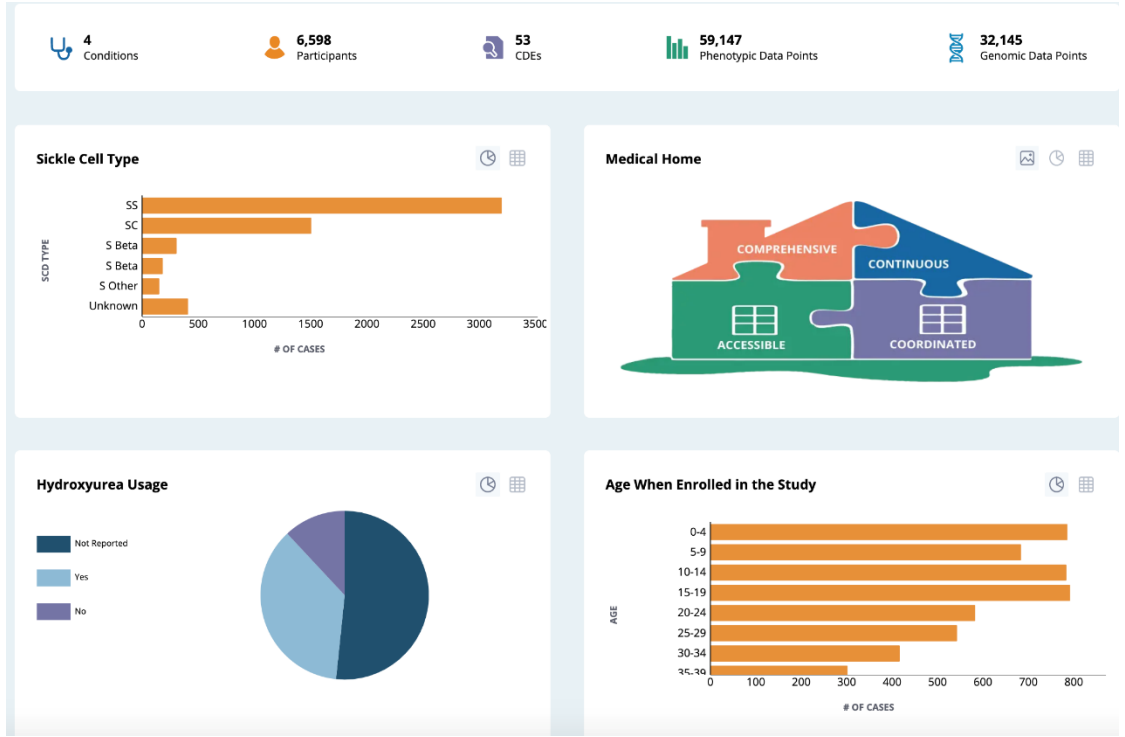
Case Level

Care Guidelines

Secondary Use and Research



	Conditions	Participants	CDEs	Phenotypic Data Points	Genomic Data Points
BabySeq	10	159	30	1,522	159
Project Cure Spinal Muscular Atrophy (SMA) - Natural History Data Repository	5	701	4,303	4,183,776	1,402
IBEMC MCAD Cohort	1	380	2,477	298	1,181,721
IBEMC PKU Cohort	2	488	2,649	1,361,100	121
World Wide Krabbe Registry	5	196	187	40,811	72
Sickle Cell Disease NBS Follow-Up Program	4	6,598	53	59,147	32,145
SPOT SMA (Spinal Muscular Atrophy) - Longitudinal Outcomes Repository for NBS-identified SMA cases	5	419	4,650	1,515,160	970,589



Long-term follow-up tools designed for all stakeholders and including data dictionaries, electronic case report forms, HPO mappings, NIH CDE repository, case level data dashboards

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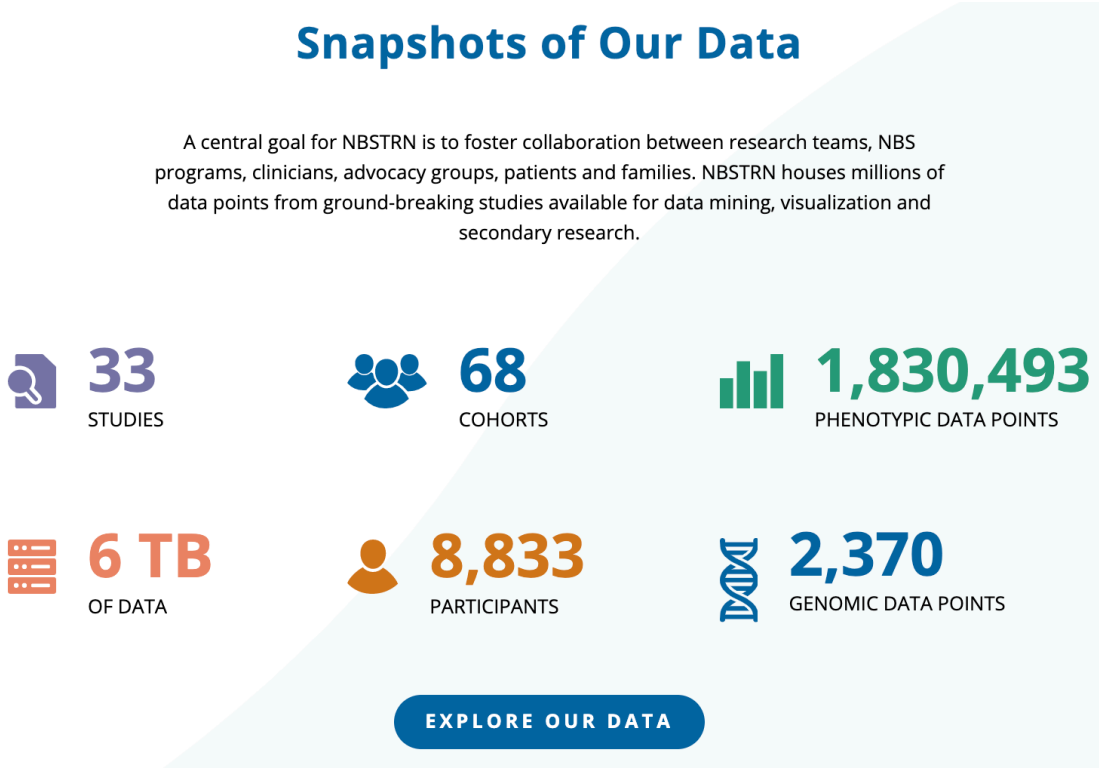
Efforts Utilizing NBSTRN

- 10-year Natural History Study
- State NBS Program Follow-Up
- Pilot of Conditions Recently Recommended to RUSP
- Pilot of Candidate Conditions
- Prospective SMA Outcomes Study
- Patient Registry Transformation
- Community Organization Based NBS Follow-Up Program



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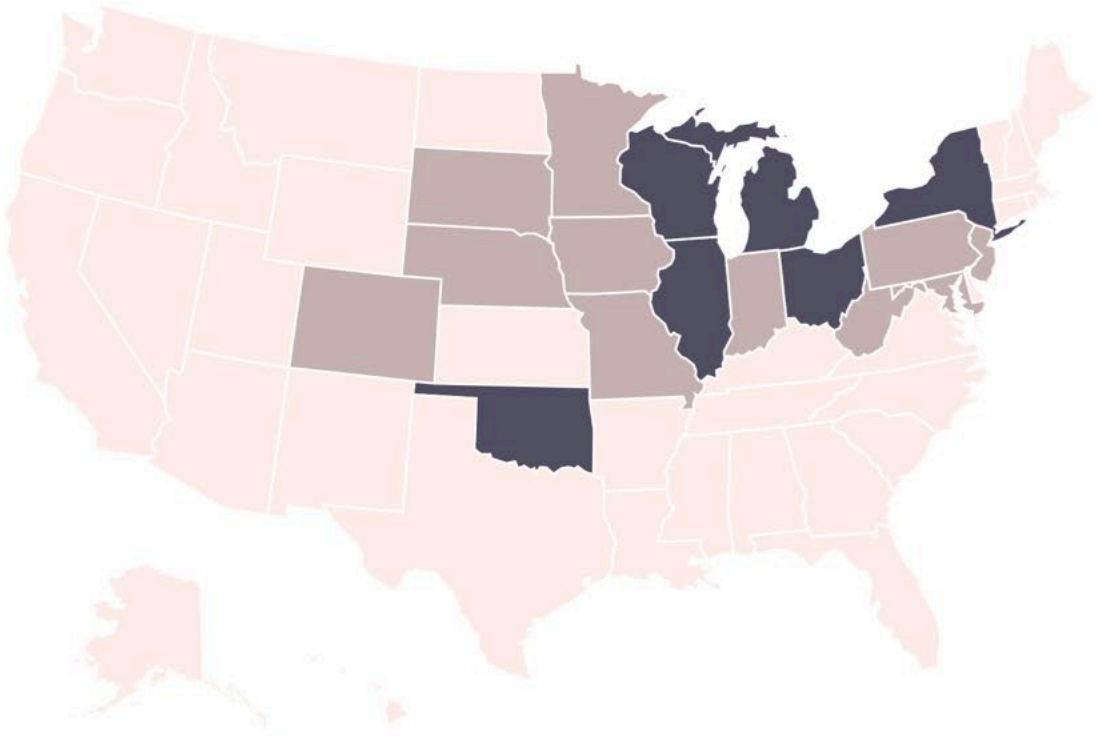
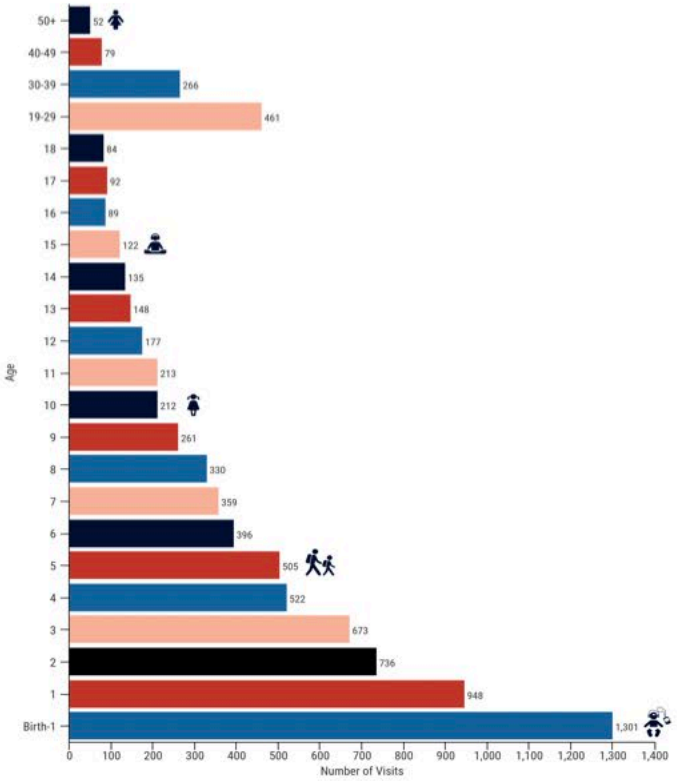
Case Study – IBEMC Natural History

724,474 Data Points

25 Clinics Across the United States



Ages Birth to 92 years



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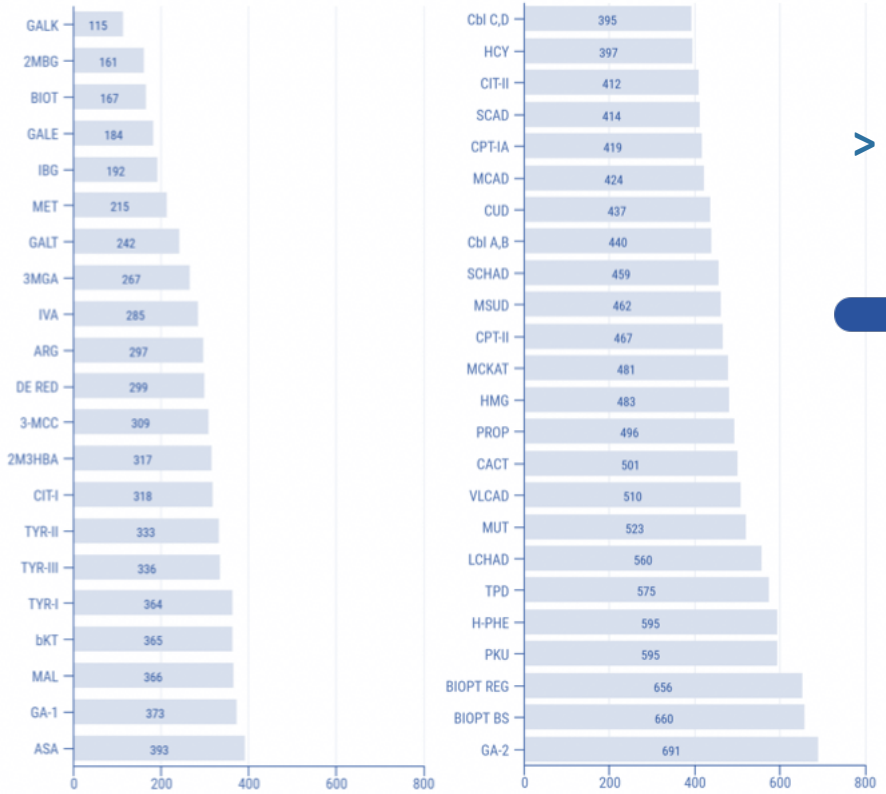
NBSTRN + NCC Follow-Up Effort

- Goal was to create a minimum set of questions and answers from the LPDR CDEs for use by State NBS Programs



> 2500 Questions

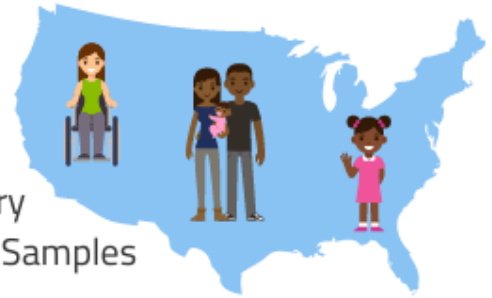
4 Questions

Condition	Age of Appropriate First Intervention
Alive?	Within the Last 12 Months Did They Receive Care and Treatment Specific to Their Condition?

LPDR
Longitudinal Pediatric Data Resource

NBS-CR
NBS Conditions Resource

NBS-VR
NBS Virtual Repository of States, Subjects & Samples



New Membership Site and Updated Tools

- De-identified Case Level Data Dashboards
- Expanded CDE Sets
- Disease Resource for RUSP, Pilot and Candidate Conditions
- State NBS Program Information for Investigators

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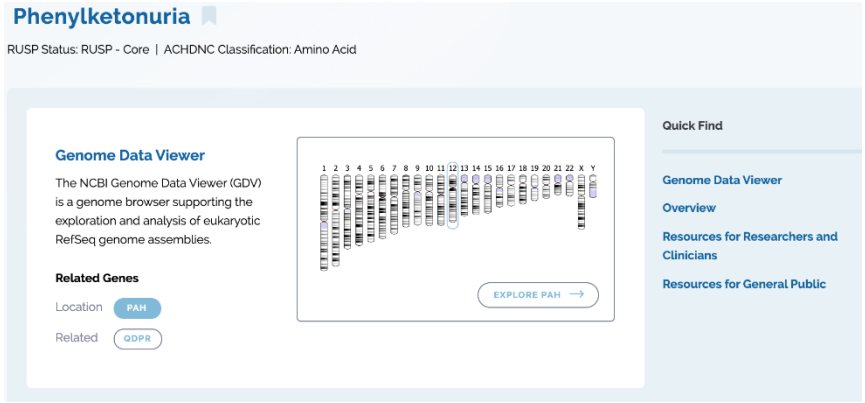
Key information on conditions that are part of, or candidates for newborn screening



MedGen

OMIM

Management

ACMG ACT Sheets

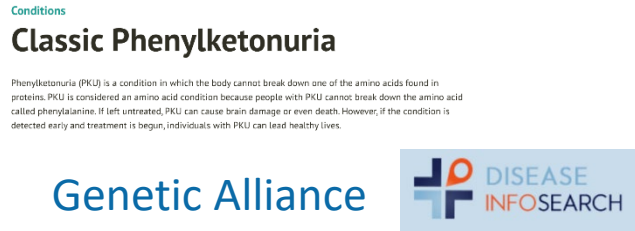
Diagnosis

Clinical Characteristics

NBS-CR

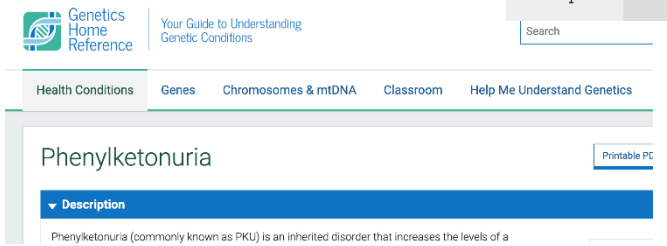
NBS Conditions Resource

Baby's First Test



Genetic Alliance

Genetics Home Reference



RUSP, Pilot and Candidate Disorders

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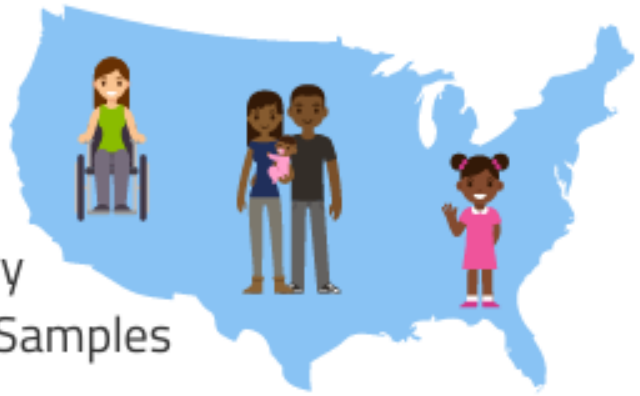
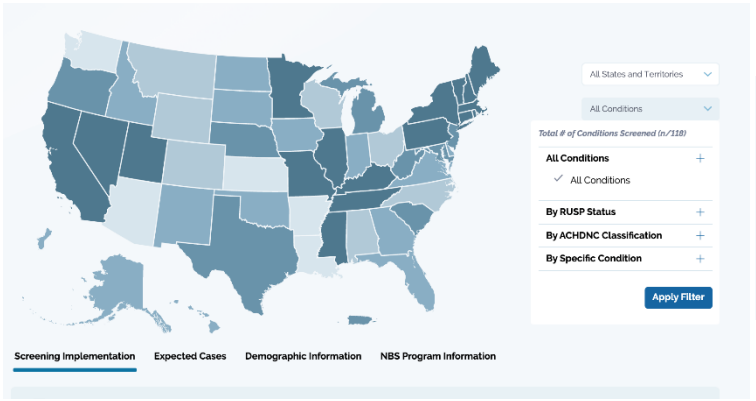
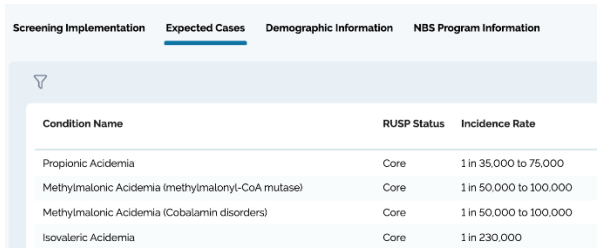
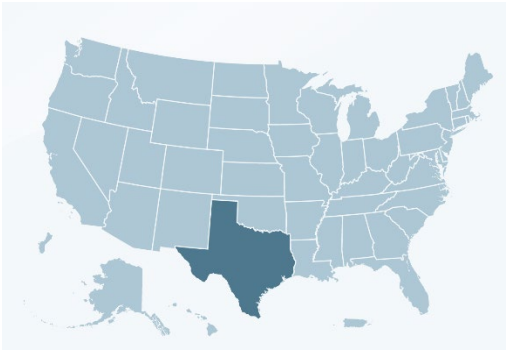
NBS-VR

NBS Virtual Repository
of States, Subjects & Samples









Texas State Profile

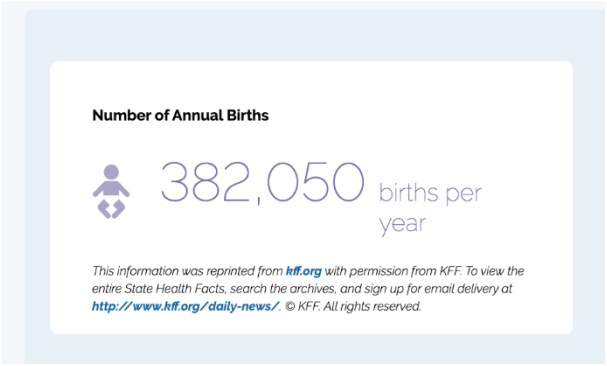
Mountain States Regional Genetics Network (MSRGN)

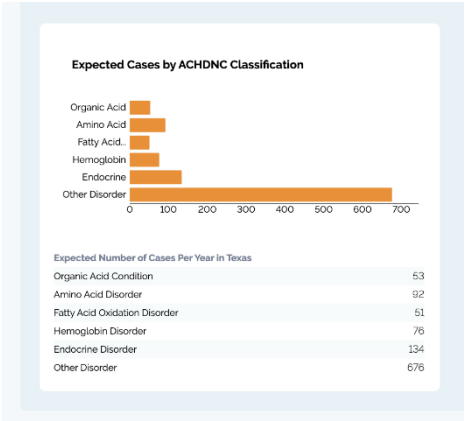
Annual Births: 382050

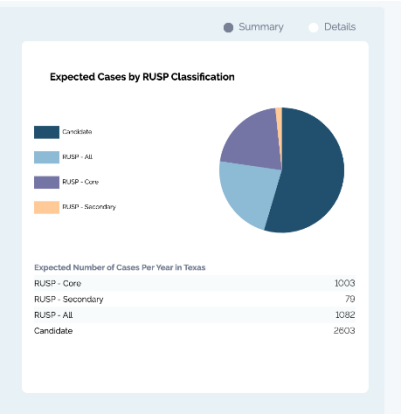
Total # RUSP Conditions Screened (n/61): 55

Total # Candidate Conditions Screened (n/57): 0

Last Updated April 8, 2020







Helping Investigators Navigate State NBS Programs

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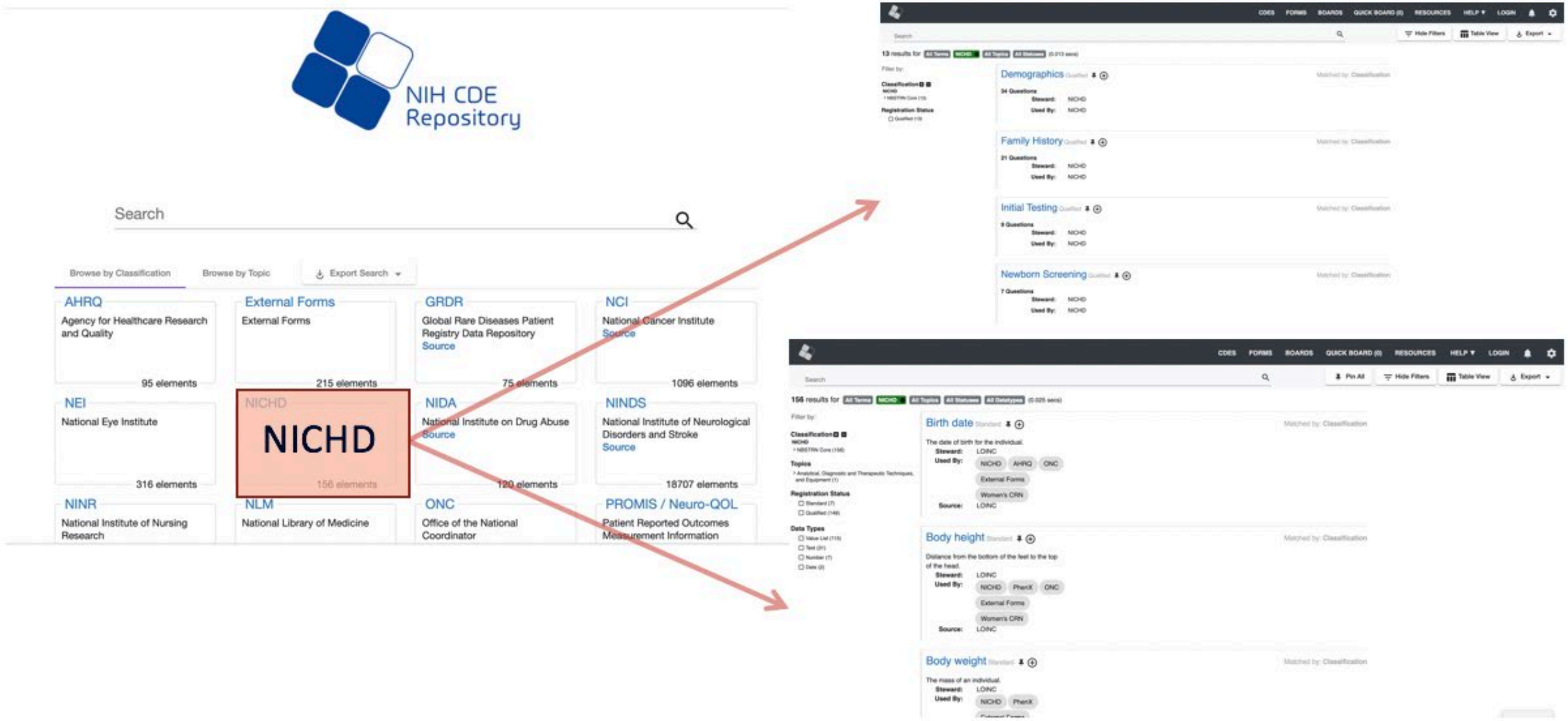
NICHD Module at NIH CDE Repository



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