

Innovations in Long-Term Follow-Up Panel

Data Tools and Resources from the Newborn Screening Translational Research Network

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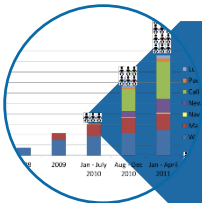
Eunice Kennedy Shriver National Institute of Child Health and Human Development Hunter Kelly Newborn Screening Research Program



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 | Newborn Screening Translational Research Network

Amy Brower ▾

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Accelerating Discoveries in Newborn Screening

Every day, babies born in the United States receive comprehensive screening for treatable diseases. Newborn screening saves lives, and discoveries by researchers make it possible!

[SIGN UP](#)



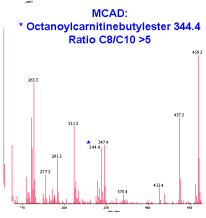
NBSTRN leverages the multi-component, multi-stakeholder system of NBS to facilitate research



Prenatal Engagement



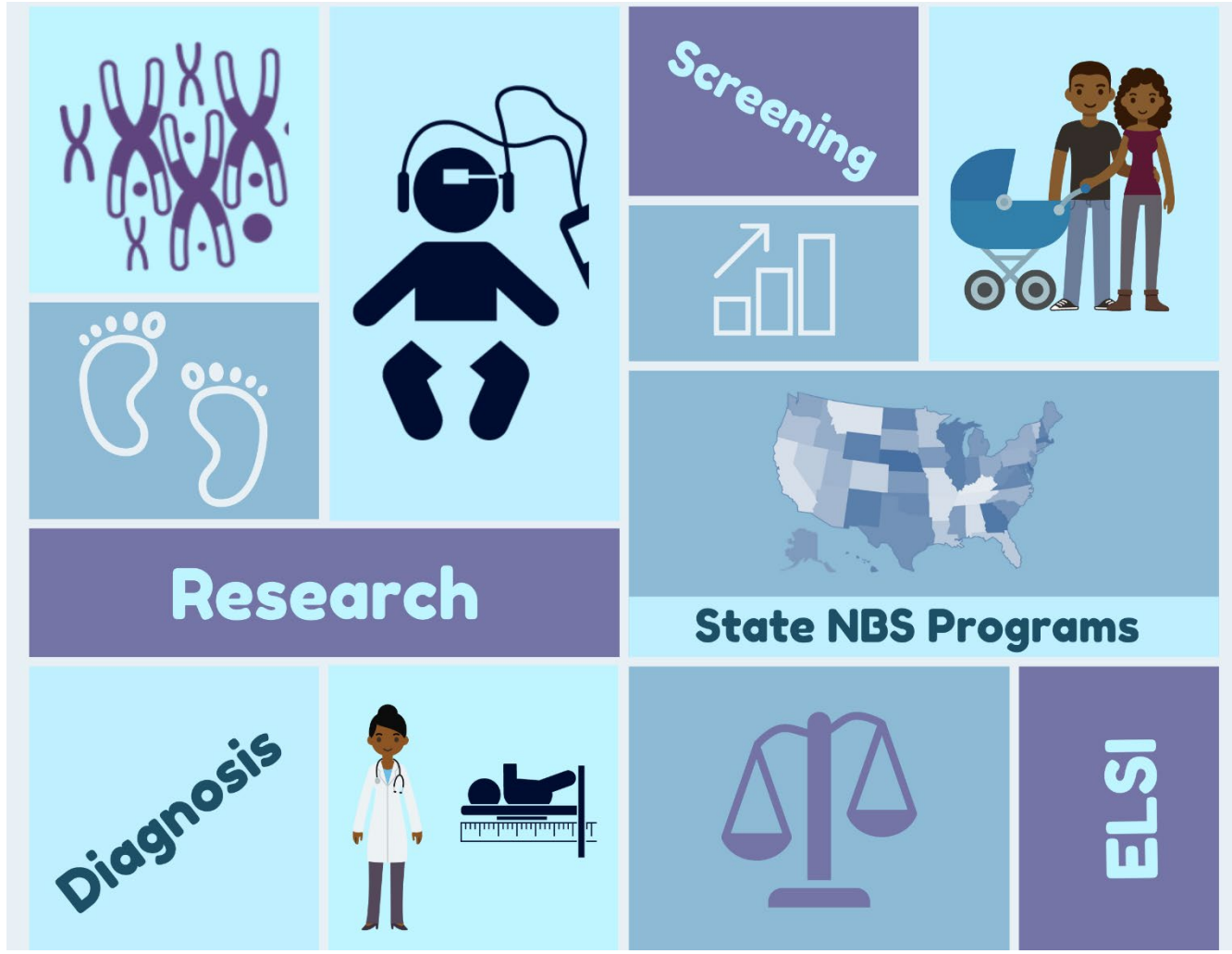
Neonatal Screening



Diagnosis and Clinical Assessment



Clinical Care and Longitudinal Follow-Up



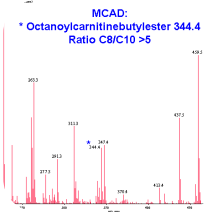
The NBS system offers a unique opportunity to conduct long-term follow-up



Prenatal Engagement



Neonatal Screening



Diagnosis and Clinical Assessment

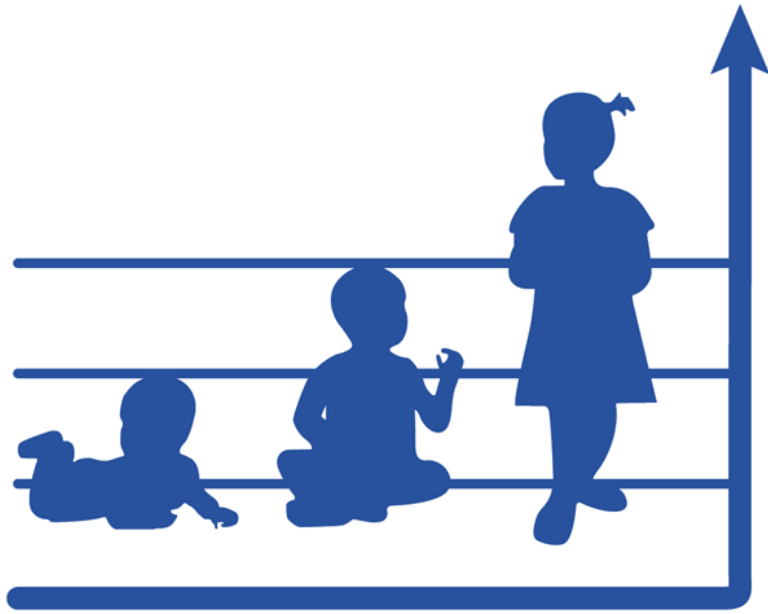


Clinical Care and Longitudinal Follow-Up



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

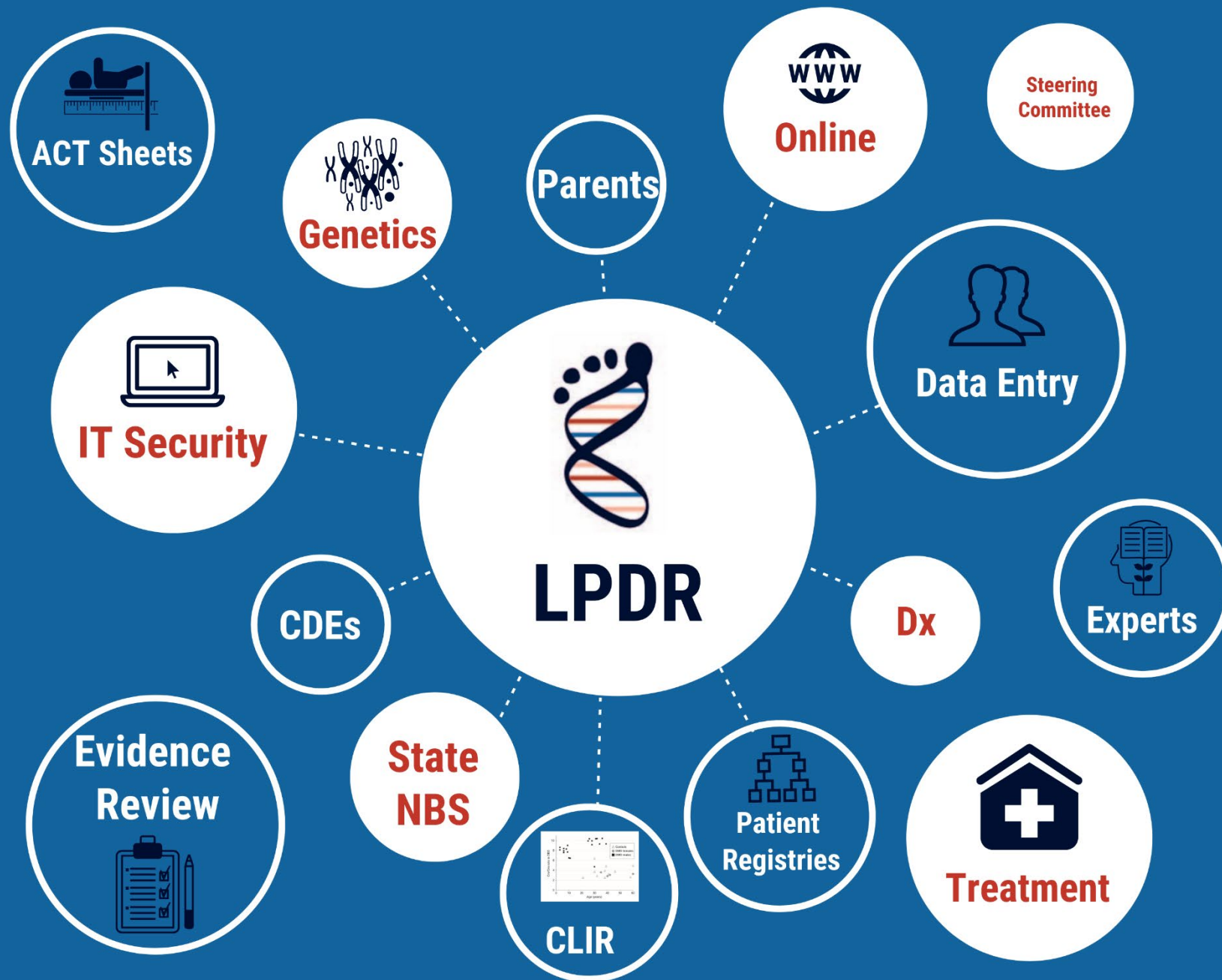
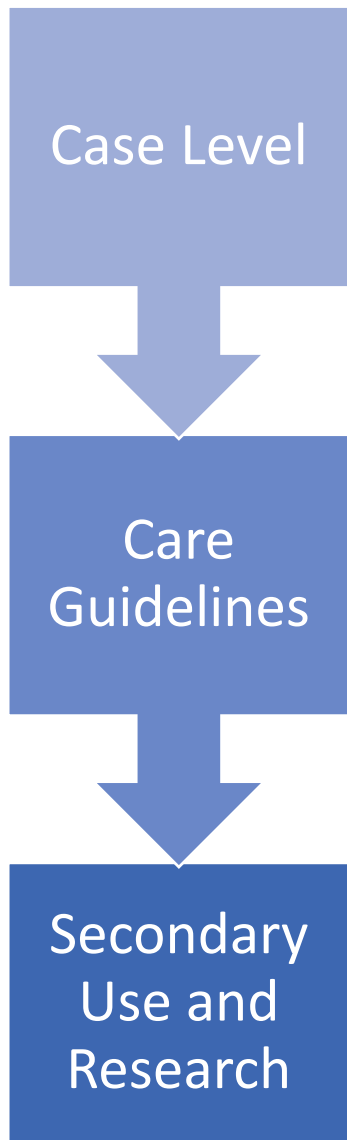
The ability to capture clinical information early in the clinical course of a disease, even before clinical symptoms appear, advances disease understanding, helps to establish the efficacy of new treatments and management approaches, informs the community at large about the value of early identification 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

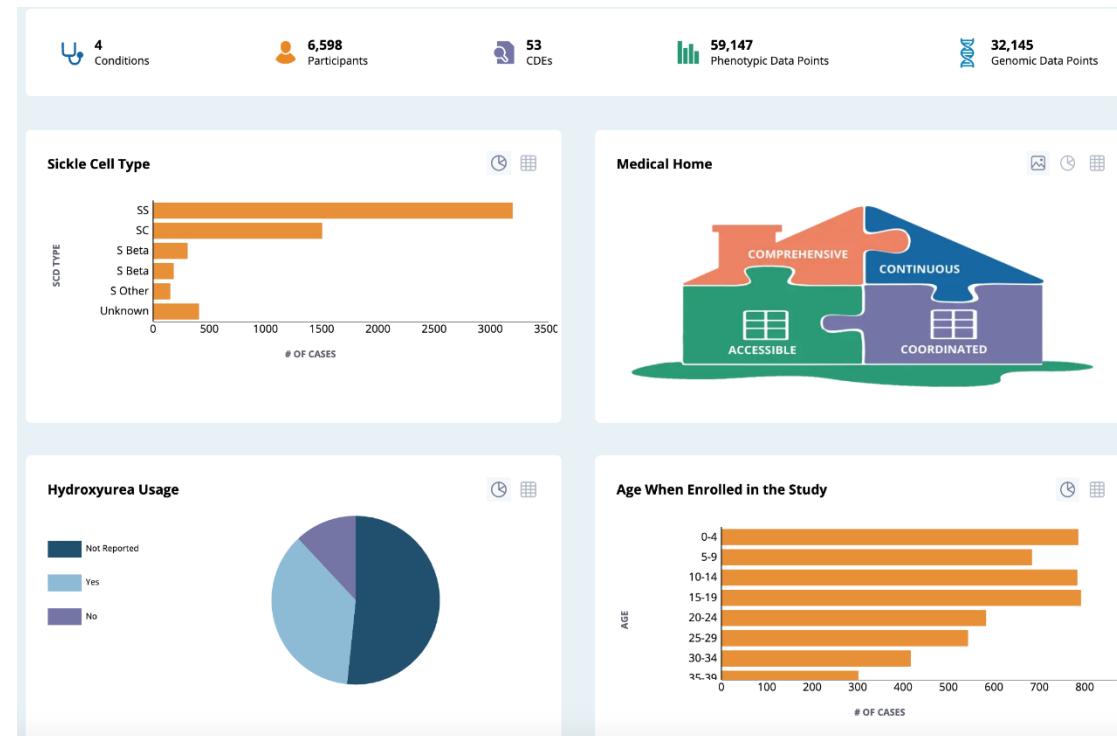


NBSTRN

Newborn Screening
Translational Research
Network



	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



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

Snapshots of Our Data

A central goal for NBSTRN is to foster collaboration between research teams, NBS programs, clinicians, advocacy groups, patients and families. NBSTRN houses millions of data points from ground-breaking studies available for data mining, visualization and secondary research.



33
STUDIES



68
COHORTS



1,830,493
PHENOTYPIC DATA POINTS



6 TB
OF DATA



8,833
PARTICIPANTS



2,370
GENOMIC DATA POINTS

[EXPLORE OUR DATA](#)

Case Study – IBEMC Natural History

724,474 Data Points

25 Clinics Across the United States

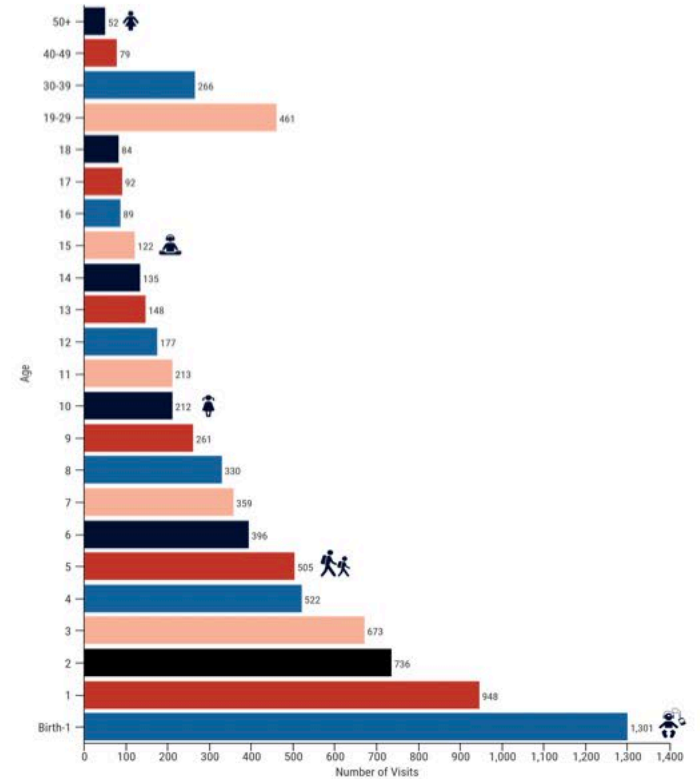
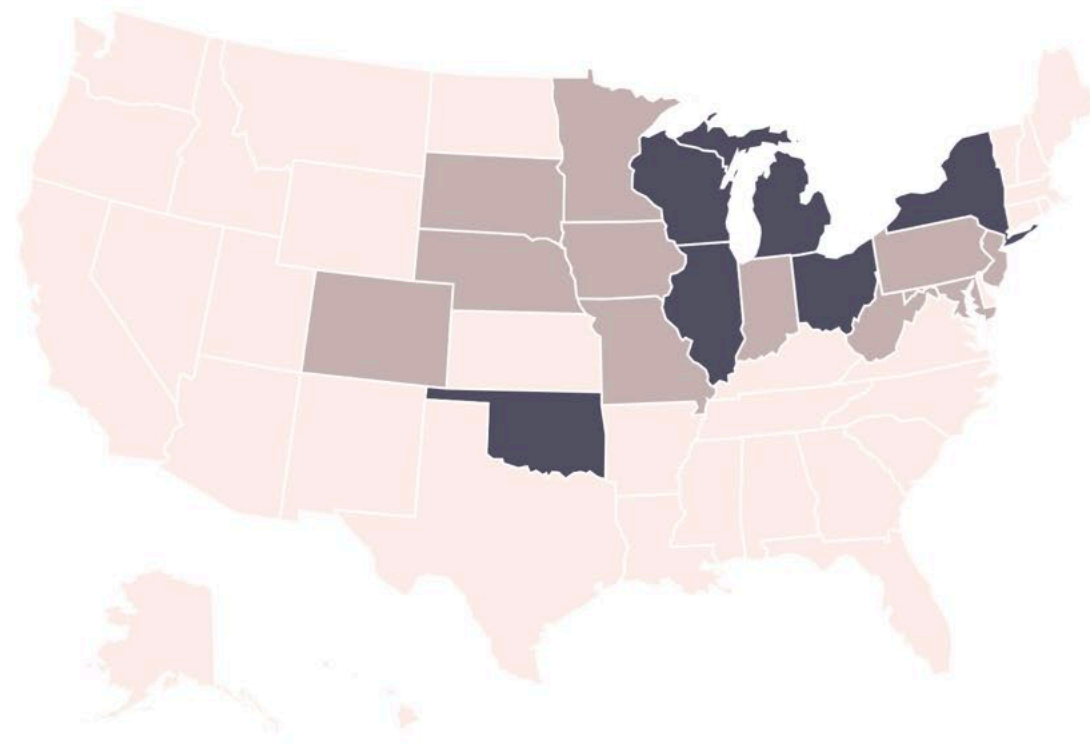
Ages Birth to 92 years



2136 subjects have data for an Initial Visit

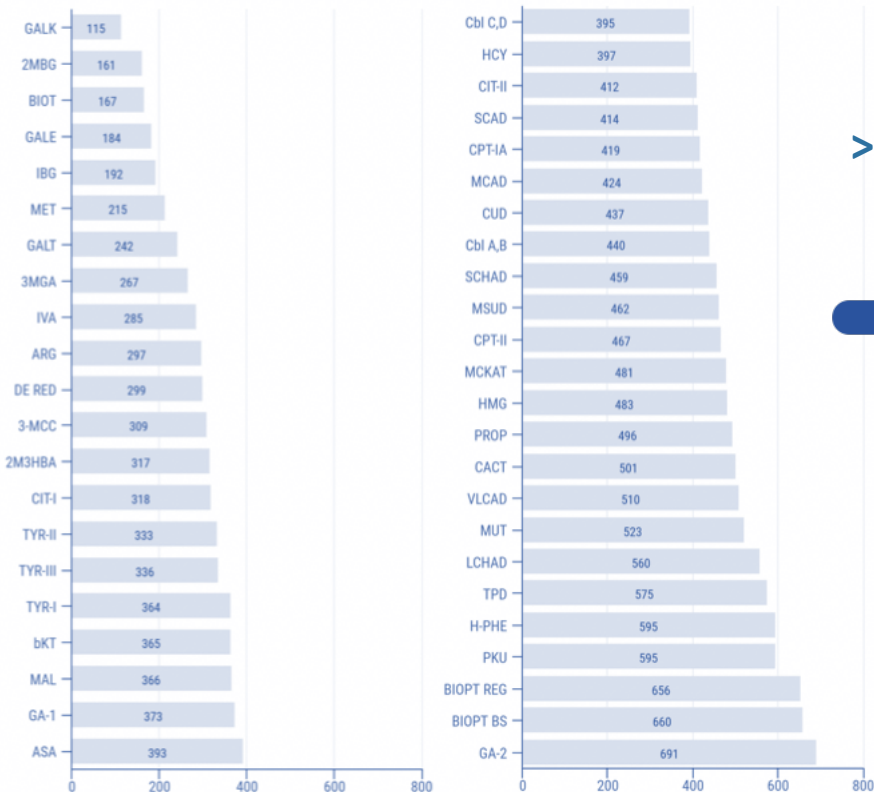


1621 subjects have longitudinal data



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?

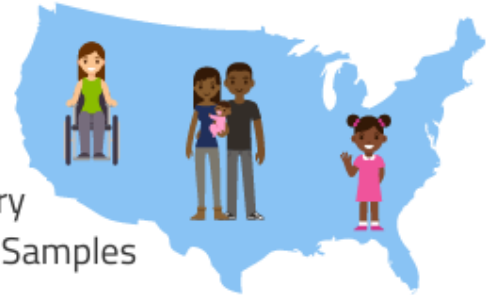
Within the Last 12 Months Did They Receive Care and Treatment Specific to Their Condition?



NBS-CR
NBS Conditions
Resource

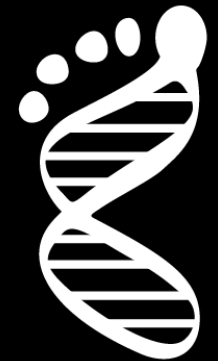


NBS-VR
NBS Virtual Repository
of States, Subjects & Samples



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



nbstrn.org

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

Phenylketonuria

RUSP Status: RUSP - Core | ACHDNC Classification: Amino Acid

Genome Data Viewer

The NCBI Genome Data Viewer (GDV) is a genome browser supporting the exploration and analysis of eukaryotic RefSeq genome assemblies.

Related Genes

Location: **PAH**

Related: **GDPR**

Quick Find

- Genome Data Viewer
- Overview
- Resources for Researchers and Clinicians
- Resources for General Public

EXPLORE PAH →

MedGen

ACMG ACT Sheets

OMIM

Diagnosis

Management

Clinical Characteristics

NBS-CR NBS Conditions Resource



Baby's First Test



Conditions

Classic Phenylketonuria

Phenylketonuria (PKU) is a condition in which the body cannot break down one of the amino acids found in proteins. PKU is considered an amino acid condition because people with PKU cannot break down the amino acid called phenylalanine. If left untreated, PKU can cause brain damage or even death. However, if the condition is detected early and treatment is begun, individuals with PKU can lead healthy lives.

Genetic Alliance



Genetics Home Reference



Your Guide to Understanding Genetic Conditions

Health Conditions Genes Chromosomes & mtDNA Classroom Help Me Understand Genetics

Phenylketonuria

Printable PDF

Description

Phenylketonuria (commonly known as PKU) is an inherited disorder that increases the levels of a

Find Support	Support Groups
News & Events	News Feeds Events
Clinical Trials	Open Studies
Publications	Editorial Articles Research Articles Review Articles
Participate	Share your experience

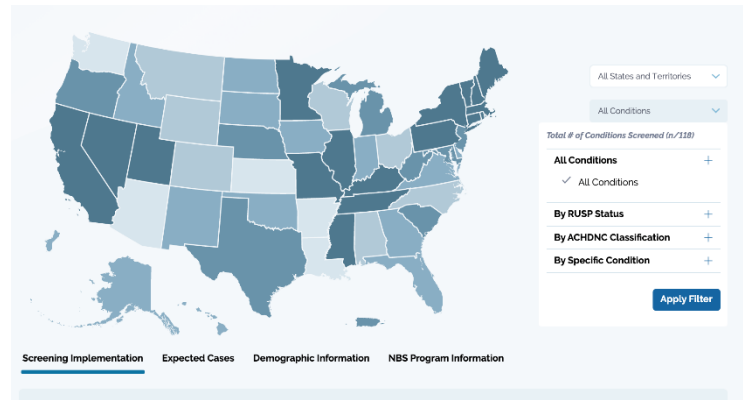
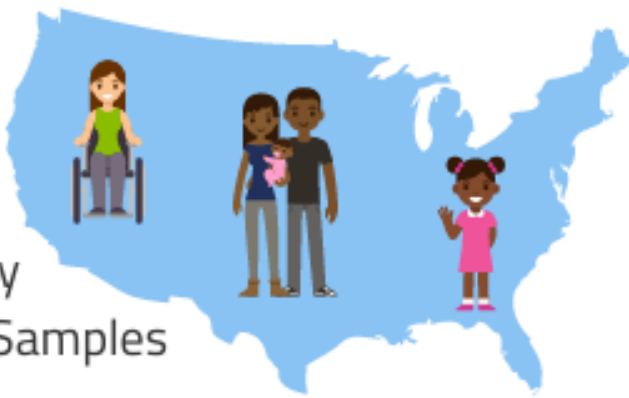
RUSP, Pilot and Candidate Disorders



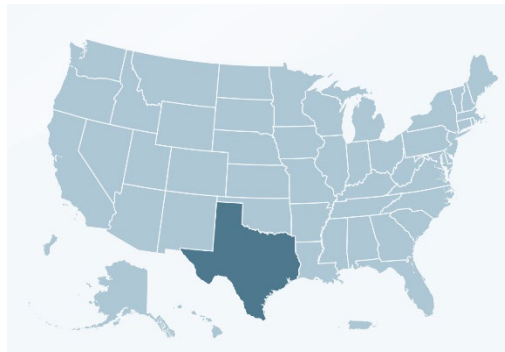
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NBS-VR

NBS Virtual Repository
of States, Subjects & Samples



Condition Name	RUSP Status	Incidence Rate
Propionic Acidemia	Core	1 in 35,000 to 75,000
Methylmalonic Acidemia (methylmalonyl-CoA mutase)	Core	1 in 50,000 to 100,000
Methylmalonic Acidemia (Cobalamin disorders)	Core	1 in 50,000 to 100,000
Isovaleric Acidemia	Core	1 in 230,000



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

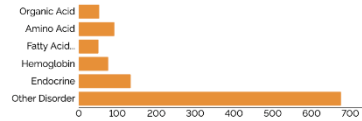
Number of Annual Births



382,050 births per year

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Expected Cases by ACHDNC Classification



Expected Number of Cases Per Year in Texas

Organic Acid Condition	53
Amino Acid Disorder	92
Fatty Acid Oxidation Disorder	51
Hemoglobin Disorder	76
Endocrine Disorder	134
Other Disorder	676

Expected Cases by RUSP Classification



Expected Number of Cases Per Year in Texas

RUSP - Core	1003
RUSP - Secondary	79
RUSP - All	1082
Candidate	2603

Helping Investigators Navigate State NBS Programs



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Newborn Screening
Translational Research
Network

NICHD Module at NIH CDE Repository

Search

Browse by Classification Browse by Topic Export Search

AHRQ Agency for Healthcare Research and Quality 95 elements	External Forms External Forms 215 elements	GRDR Global Rare Diseases Patient Registry Data Repository Source 75 elements	NCI National Cancer Institute Source 1096 elements
NEI National Eye Institute 316 elements	NICHD National Institute on Child Health and Human Development 156 elements	NIDA National Institute on Drug Abuse Source 120 elements	NINDS National Institute of Neurological Disorders and Stroke Source 18707 elements
NINR National Institute of Nursing Research	NLM National Library of Medicine	ONC Office of the National Coordinator	PROMIS / Neuro-QOL Patient Reported Outcomes Measurement Information

13 results for All Terms **NCHD** All Topics All Statuses All Data Types (0:210 sec)

Filter by:

- Classification: NCHD (140 items) (15)
- Registration Status: Qualified (15)

Demographics Qualified

34 Questions
Steward: NCHD
Used By: NCHD

14 Qualified

Matched by: Classification

Family History Qualified

21 Questions
Steward: NCHD
Used By: NCHD

Matched by: Classification

Initial Testing Qualified

9 Questions
Steward: NCHD
Used By: NCHD

Matched by: Classification

Newborn Screening Qualified

7 Questions
Steward: NCHD
Used By: NCHD

Matched by: Classification

156 results for All Terms **NCHD** All Topics All Statuses All Data Types (0:525 sec)

Filter by:

- Classification: NCHD (156 items) (156)
- Topics: Analytical, Diagnostic and Therapeutic Techniques, and Equipment (1)
- Registration Status: Member (7), Qualified (149)
- Data Types: Value List (115), Text (31), Number (7), Date (3)

Birth date Standard

The date of birth for the individual.
Steward: LONG
Used By: NCHD, AHRQ, ONC

Topics: External Forms, Women's ORN

Source: LONG

Matched by: Classification

Body height Standard

Distance from the bottom of the feet to the top of the head.
Steward: LONG
Used By: NCHD, PhenX, ONC

Topics: External Forms, Women's ORN

Source: LONG

Matched by: Classification

Body weight Standard

The mass of an individual.
Steward: LONG
Used By: NCHD, PhenX

Matched by: Classification

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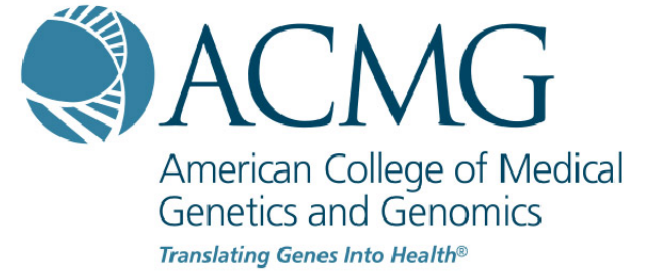


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