



# Long-Term Follow-Up Data Collection After Newborn Screening: Development of a Consensus Core Data Set

**Short-term and Long-term Follow-Up Workshop  
Data Harmonization**

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**for the Joint Committee of the NBSTRN Clinical Centers Workgroup and the NCC/RC Long-Term Follow-Up Workgroup**





- ◆ **Long-Term Follow-Up in Context**
  - ◆ **Joint Committee Effort**
  - ◆ **Development of Consensus Core Data Set**
  - ◆ **Development of Data Capture Tool**
  - ◆ **Implementation Considerations**
  - ◆ **Conclusions**
-



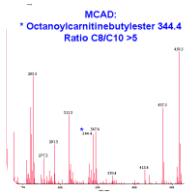
## Newborn Screening System



Prenatal Education



Screening



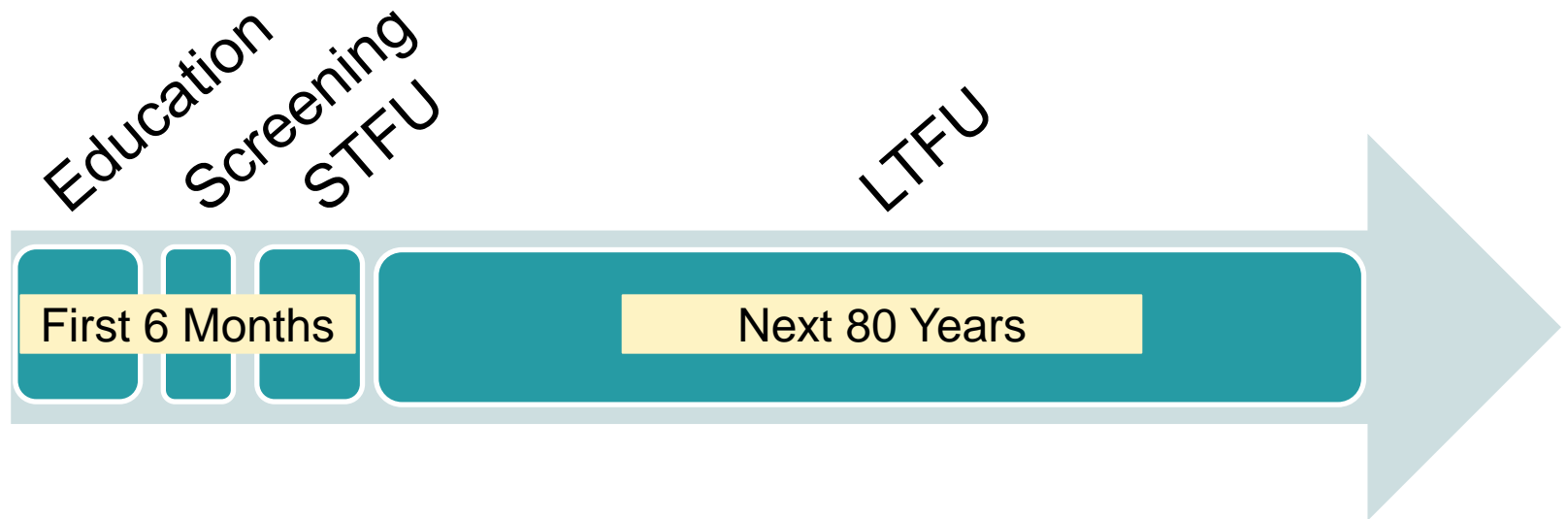
Diagnosis and Short-Term Follow-Up



Clinical Care and Long-Term Follow-Up



# Long-Term Follow-Up in Context

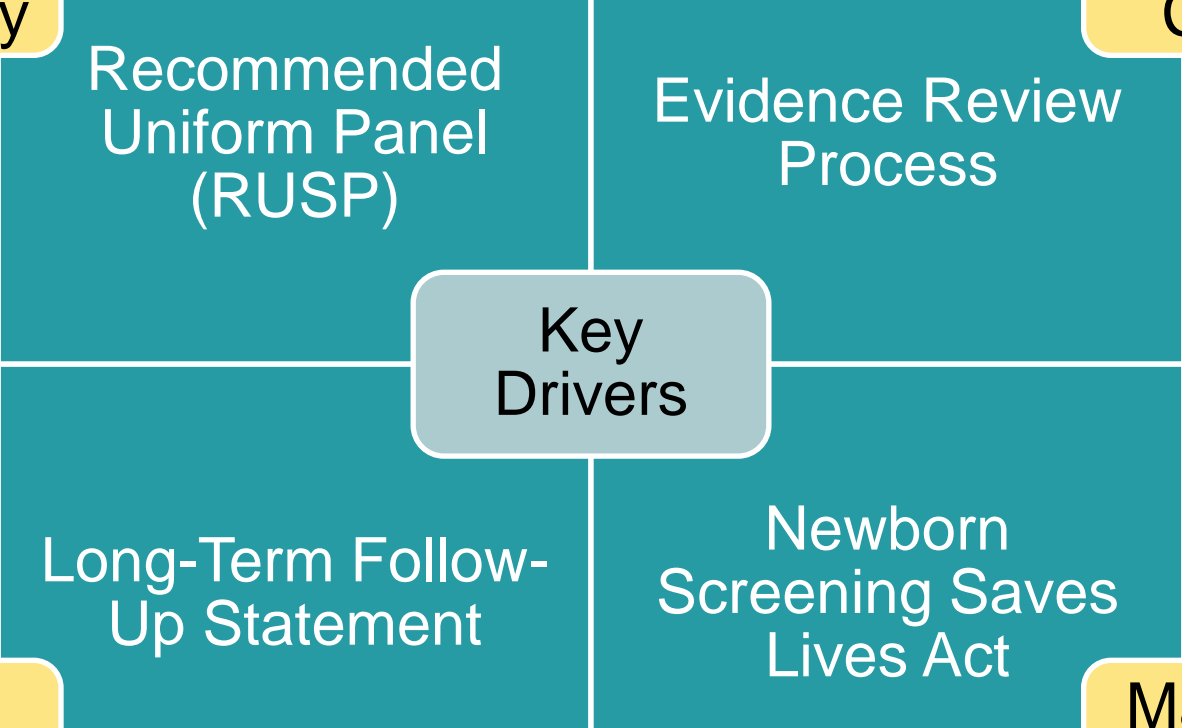




# Long-Term Follow-Up in Context

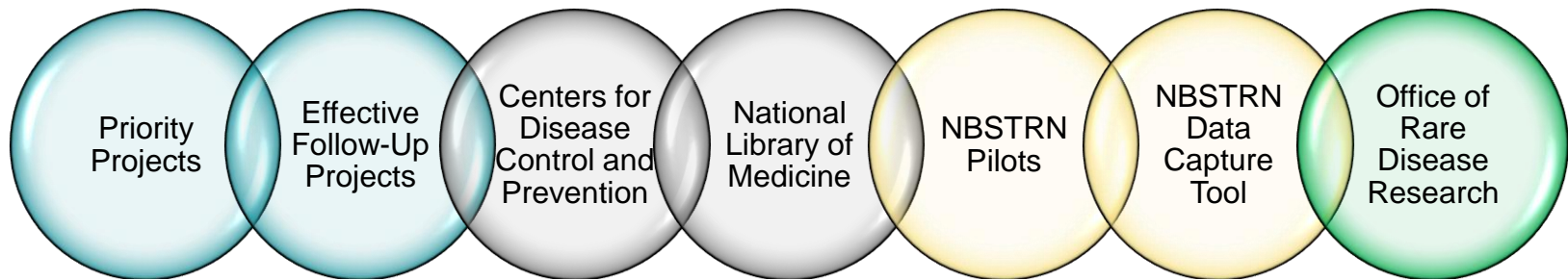
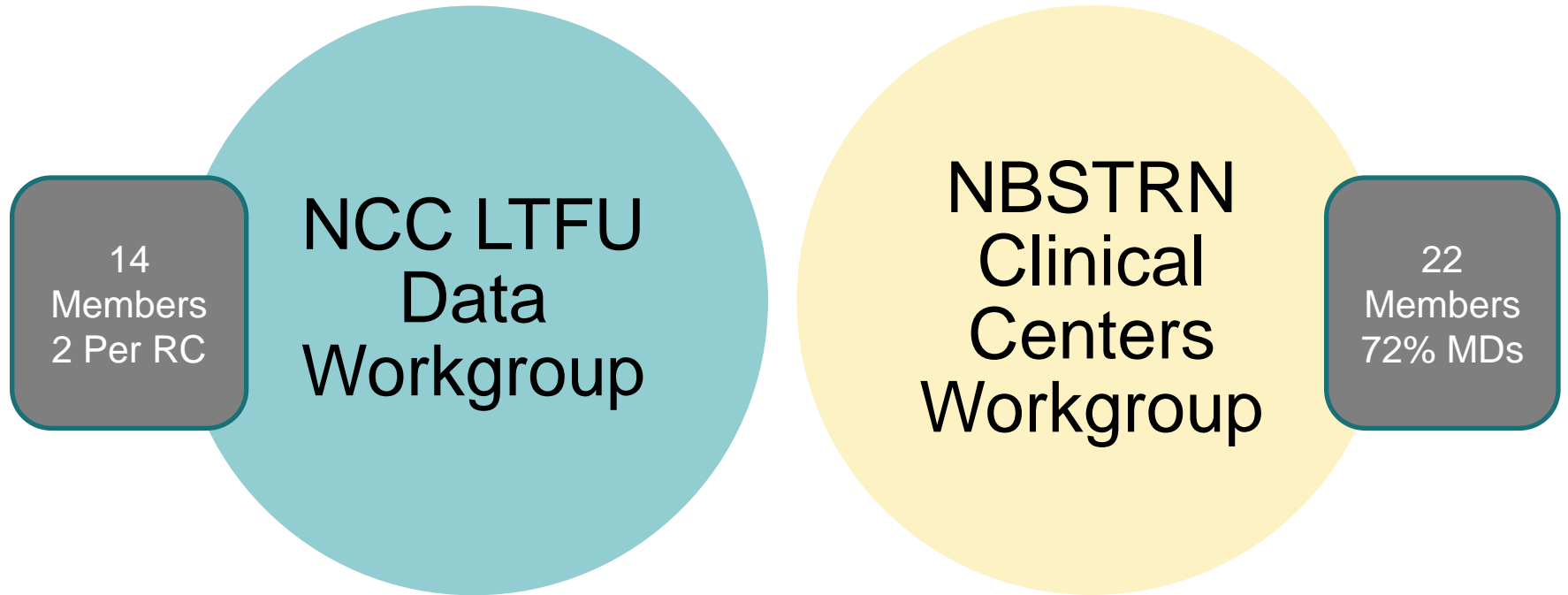
30 Core  
26 Secondary

New  
Conditions



4  
Components

Management  
& Outcomes





## ◆ Goal

- Uniform minimum data set
- Disease specific minimum data sets for the RUSP

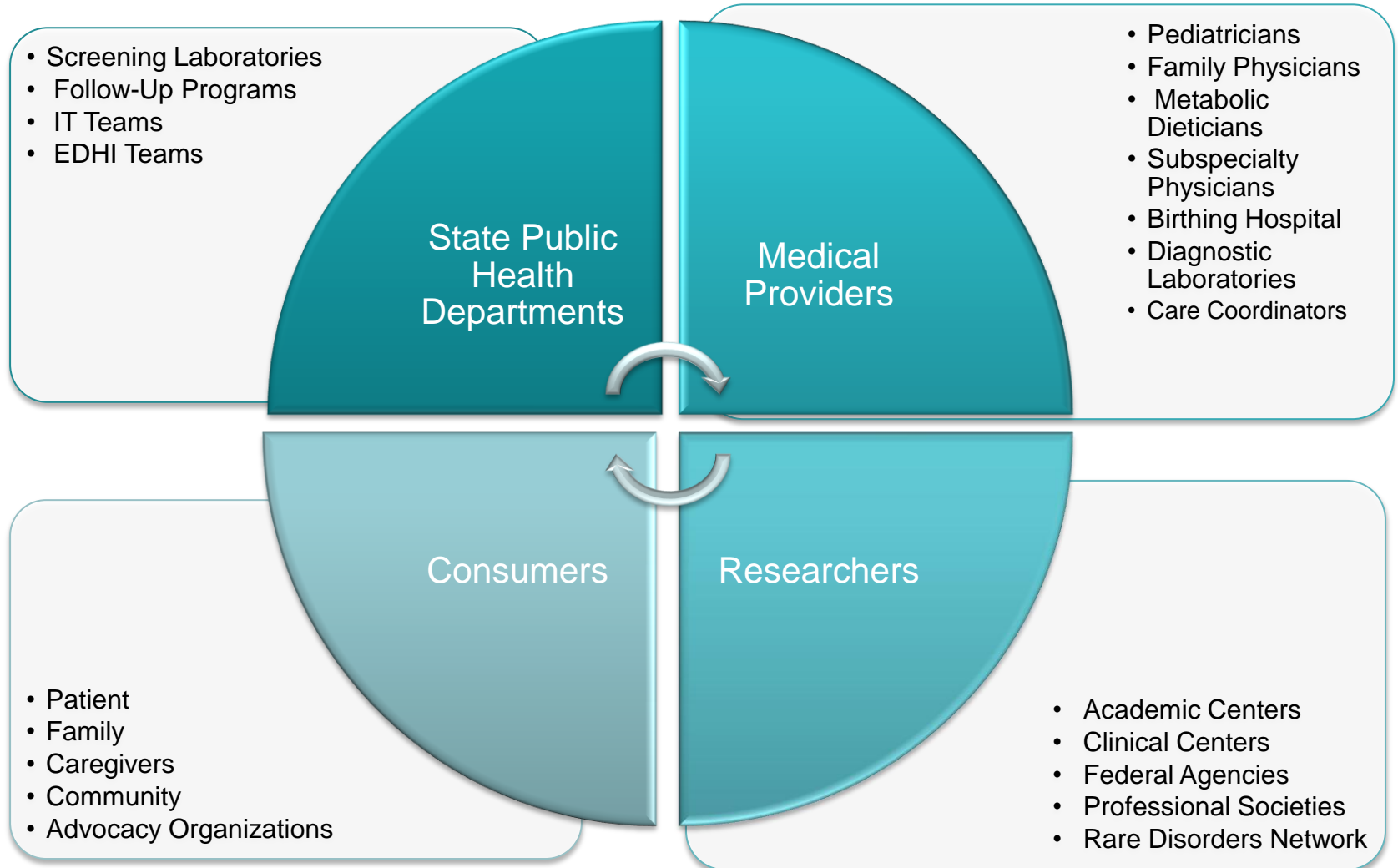
## ◆ Related Projects

- NBSTRN Pilots – LSDs, SMA
  - NBSTRN Data Capture Tools
  - NBSTRN Tools for Researchers – Model Consents
-



- ◆ **Literature and Key Effort Review** *(Repeated Periodically)*
  - HRSA Demonstration Projects
  - CDC Surveillance Project
  - ORDR Patient Registry Project
- ◆ **Stakeholder Engagement**
- ◆ **Establish Joint Workgroup**
- ◆ **Establish Disease Specific Workgroups**
- ◆ **Initiate Standardization and Coding with Partners**
- ◆ **Disseminate Data Sets**
- ◆ **Update, Disseminate, Repeat!**



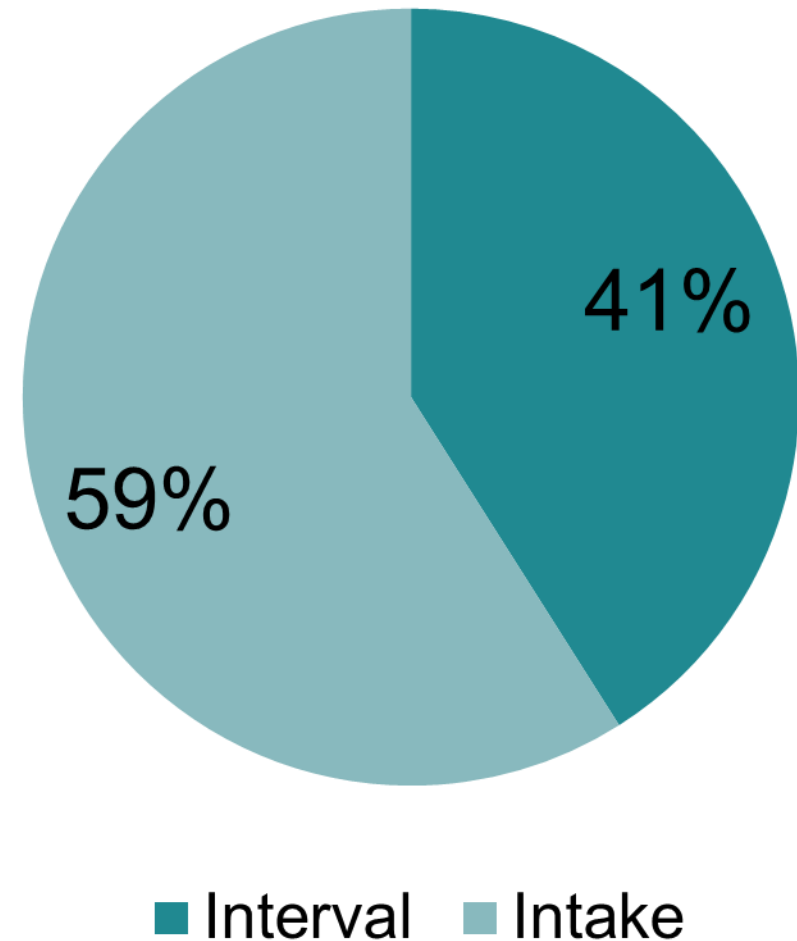




## Categories of Data

Category
Demographics/SES
Family History
Prenatal History
Newborn Screening
Neonatal History
Diagnostic Testing
Laboratory Studies
Past Health History
Pharmacotherapy
Developmental History
Education
Measurements
Nutrition
Imaging Studies
Care Coordination
Emergency Management

## Timing of Data Capture





# Data Capture Throughout the Lifespan



## Intake

- Demographics
- SES
- Family History
- Prenatal History
- Neonatal History
- Birth Measurements
- Newborn Screening
- Hearing Screening
- Diagnostic Testing



## Childhood

- Monitoring Labs
- Diet
- Therapies
- Emergency Management
- Developmental Screening
- Imaging Studies
- Intercurrent Complications

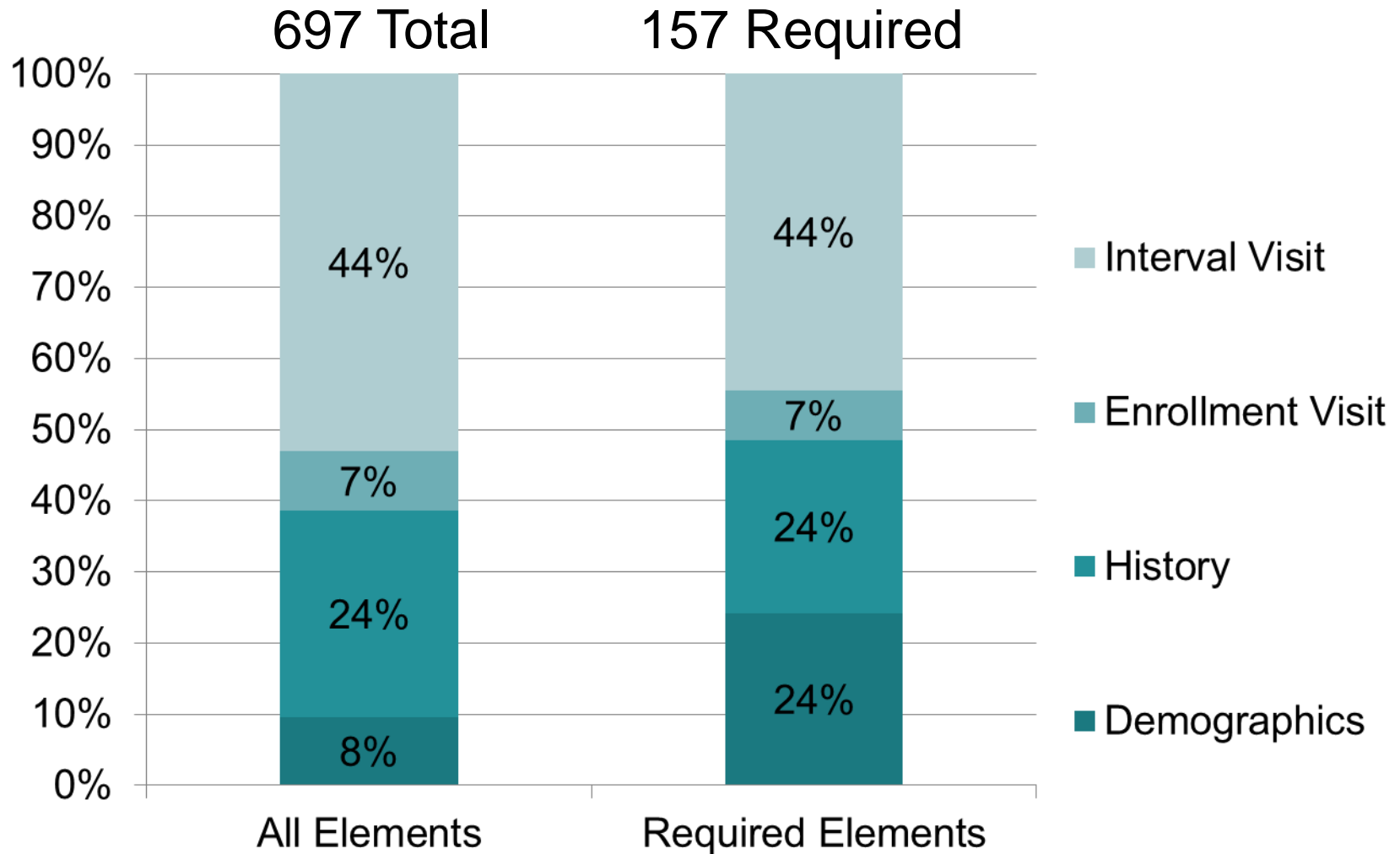


## Adulthood

- Monitoring Labs
- Diet
- Therapies
- Emergency Management
- Imaging Studies
- Intercurrent Complications

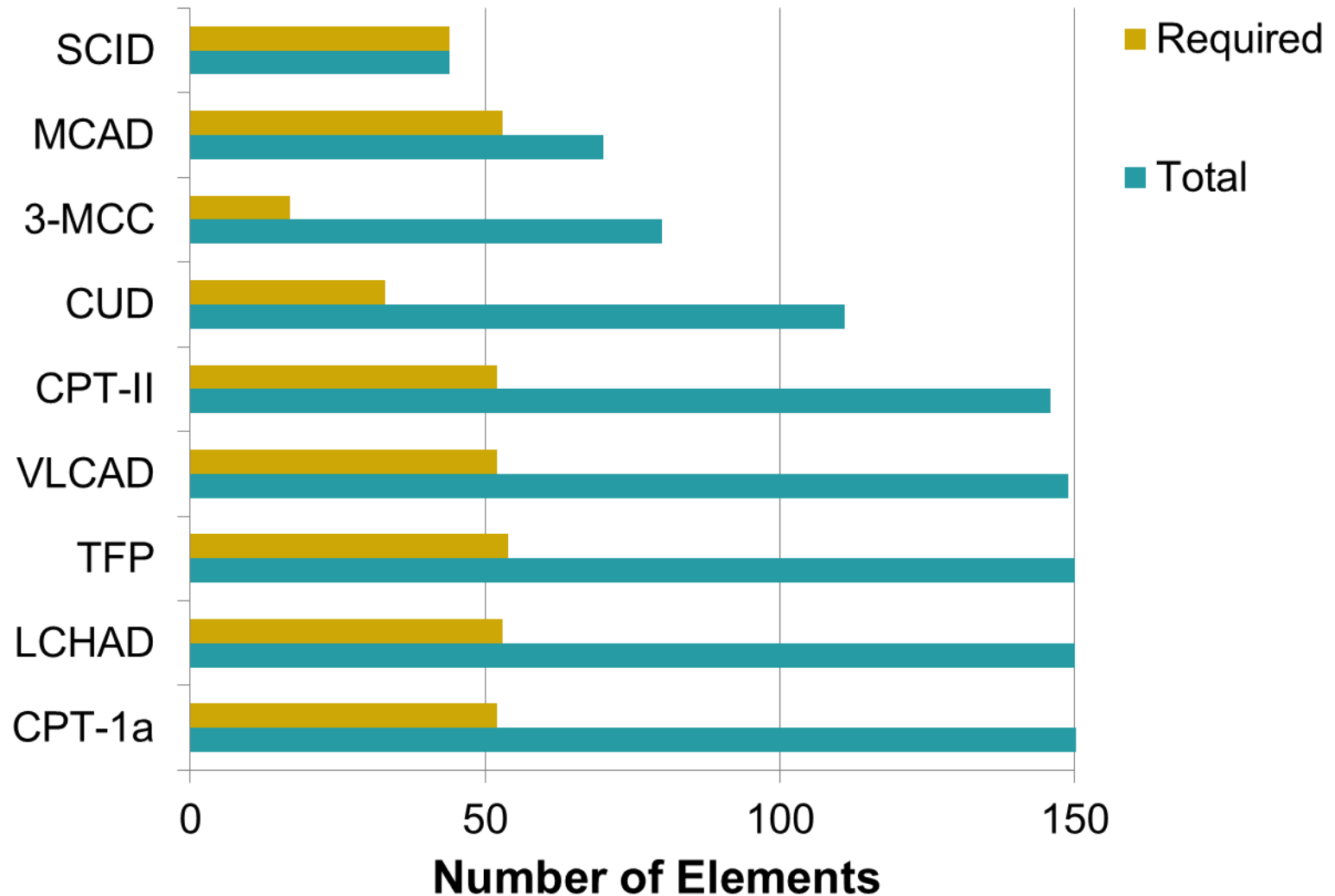


# Uniform Minimum Data Set





# Disease Specific Data Sets





## Investigator – New & Existing Technologies, Novel Treatment & Management Strategies

- Enable Novel Statistically Robust Proposals

Describe the clinical course of NBS identified conditions in which patients are asymptomatic.

## Grantee – New & Existing Technologies, Novel Treatment & Management Strategies

- Accelerate & Facilitate Research

What is the relationship between CFTR genotypes and lung function in adolescence for newborn screen identified cystic fibrosis patients?

## Public Health Partner – Service Delivery & Quality Assurance/Improvement

- Implement Technologies & Assess Health Outcomes for Novel Treatments

Describe the relationship between service delivery and treatment methods to define optimal follow-up care plans for children with MCAD.



## Relationship between C8 values and genotype, specifically A985G.

### C8

- NLM Coding Guide
  - Condition
  - Abbreviation
  - SNOMED CT Code
  - ICD-9-CM Code
  - ICD-10-CM Code
  - Enzyme Commission Number
  - UniProt Number
  - Analytes or Measurements
  - Other - OMIM

### A985G

- NCBI
  - HGNC Approved Gene Symbol
- OMIM
  - Gene Info
  - NCBI Gene
  - KEGG
  - PharmGKB
  - UCSC
- Clinical Resources
  - Animal Models
  - Cellular Pathways
- dbSNP
  - rs77931234
- PubMed

LOINC Long Common Name <sup>9</sup>	Analyte Short Name <sup>10</sup>	LOINC Number <sup>11</sup>	Units <sup>12</sup>
Decanoylcarnitine (C10) [Moles/volume] in Dried blood spot	C10	45197-1	umol/L
Decenoylcarnitine (C10:1) [Moles/volume] in Dried blood spot	C10:1	45198-9	umol/L
Hexanoylcarnitine (C6) [Moles/volume] in Dried blood spot	C6	45211-0	umol/L
Octanoylcarnitine (C8) [Moles/volume] in Dried blood spot	C8	53175-6	umol/L
Octanoylcarnitine (C8)/Acetylcarnitine (C2) [Molar ratio] in Dried blood spot	C8 / C2	53176-4	{Ratio}
Octanoylcarnitine (C8)/Decanoylcarnitine (C10) [Molar ratio] in Dried blood spot	C8 / C10	53177-2	{Ratio}

**ClinicalTrials.gov**  
A service of the U.S. National Institutes of Health

Found 1 study with search of: **ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN, DEFICIENCY OF**

Rank	Status	Study
1	Completed	Nutritional Therapy of the Deficits of Oxidation Mitochondrial of the Fatty Acids

Condition: Inborn Errors of Metabolism  
 Intervention: Drug: Oil special 107 and MYGLIOL 810

CTTATAGTTAGTTGTTAATACTATACCAAGTGTAACTGCTGTT  
 ATGGTTTGGTAATATGTTAACATTAGAGGAAGCTGGGTGTAACIT  
 TCGTTAATGAAAAGTTTAAAAAATAAAAAGGTCAGGCACAGTGGC  
 CAGCACTTTGAGAGGCCAAGTAGGAGGATTGCTTGAGGCCAGGA  
 GGGCACATAGCAGACCCCGTCACTATAAAAATGAAAAGCCCC  
 AGTTTCTCAATAAATACCTTAATTTTCTTTTAAATCTA  
 ATTATGCTGGCTGAATGGCAATGAAGTGTACTAGCTAGAT  
 ACCTTGGAGGTTGATCTGGTCTCGAATACTATTATGCTTC  
 TCTGGAGATATTGCAATCACTAGCTACTGATGCTGTGAGAT  
 ATTTAATACAGAAATCTCTAGAAAACCTAATAGGGATGCCAA  
 GTTAAGATGATTTTTTTGGTTTGCAGGAGAGAGAAATATTCA  
 TTCTGATTTATAGAAAATTTATGCTCCTAGTAGCAATAAGTAC  
 TGAGCCAGTTTTTGGRAATTAATAAGAAAAGAAATGTTATTT  
 TCTGAA



## ◆ **Goal**

- **Establish a network of centralized and institutionally-enabled infrastructure to support the capture and managed storage of longitudinal clinical data**
- **This data will then be used by public health, clinicians and researchers to develop treatment programs and further newborn screening research**

## ◆ **Subcontract to CHOP – Dr. Pete White PI**

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# Data Capture Screen Shot

Uniform Dataset | REDCap

https://bu.research.chop.edu/redcap\_nbstrn/redcap/redcap\_v4.3.2/data\_entry.php?pid=5&id=1&event\_id=22&page=demographics

**REDCap™**

Logged in as **loutrelj** | Log out

**My Projects**  
Project Home  
Project Setup  
Project status: **Development**

**Data Collection**

**Invite Participants**  
Get a public survey link or build a participant list for inviting respondents

**Data Entry**

**Participant ID 1 (Study ID PACH0001)**  
Event: **Intake**  
Data Collection Instruments:  
 Informed Consent (survey)  
 **Demographics**  
 History  
 Initial Testing  
 Lock all forms

**Applications**

- Calendar
- Data Export Tool
- Data Import Tool
- Data Comparison Tool
- Logging
- File Repository
- User Rights
- Record Locking Customization
- E-signature and Locking Mgmt
- Graphical Data View & Stats
- Report Builder

**Help & Information**

- Help & FAQ
- Video Tutorials
- Suggest a New Feature

If you are experiencing problems, please contact your [REDCap administrator](#).

**NBSTRN**  
Newborn Screening Translational Research Network

Newborn Screening Translational Research Network  
Long Term Follow-Up Database

**Uniform Dataset**

**Demographics**  Modify this instrument  Download PDF of - select PDF download option -

Editing existing Participant ID 1 (Study ID PACH0001)

Event Name: **Intake**

**Participant ID** 1

**Study ID** PACH0001 (Secondary unique field)  
NOTE: Modifying this value will also change the value any corresponding instances of this field in other Events where this form is used.

**Consent obtained**  Yes  No  IRB Exempt [reset value](#)

**Assent obtained**  Yes  No  Not needed [reset value](#)  
Not applicable unless 18 or older

**Permission to recontact**  Yes  No [reset value](#)

**Type of Visit**

**Demographics Information**

**Date of Birth**   M-D-Y

**Age**   
Enter age if Date of Birth is not available

**Current Age (calculated)**  [View equation](#) [Disclaimer](#)

**Gestational age**

**Sex**

**Biological mother's maiden name**   
Enter NA if not available

**Condition Follow-Up Status**  Active  Inactive [reset value](#)

**If Inactive, why?**

**Followed by more than one center**  Yes  No [reset value](#)

**Permission to share data?**  Yes  No  Unknown [reset value](#)

**Name of additional center**   
Select the name of the center from the dropdown list or specify a new center in the textbox below.



- ◆ **Disseminating Data Sets**
  - ◆ **Facilitating Adoption**
    - **Clinicians**
    - **Public Health Teams**
    - **Researchers**
  - ◆ **Preparing for Future Electronic Information Exchange**
-



- ◆ **A national community of specialty providers residing in public health, clinical centers and academic research centers can reach consensus regarding priorities for data collection for long-term follow-up.**
  - ◆ **Creates a foundation for a uniform minimum data set to ascertain the clinical history of screened disorders and for both public health and research-related activities.**
  - ◆ ***We can collect LTFU data to improve outcomes and facilitate basic and translational research for children identified by newborn screening***
-



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