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
Presentation Overview

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

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

- First 6 Months
- Next 80 Years
Joint Committee Effort

NCC LTFU Data Workgroup
- 14 Members
- 2 Per RC

NBSTRN Clinical Centers Workgroup
- 22 Members
- 72% MDs

Priority Projects
- Effective Follow-Up Projects
- Centers for Disease Control and Prevention
- National Library of Medicine
- NBSTRN Pilots
- NBSTRN Data Capture Tool
- Office of Rare Disease Research
Development of A Consensus Core Data Set

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
Methodology

- **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!**
NBS Stakeholders

- Academic Centers
- Clinical Centers
- Federal Agencies
- Professional Societies
- Rare Disorders Network

- Pediatricians
- Family Physicians
- Metabolic Dieticians
- Subspecialty Physicians
- Birthing Hospital
- Diagnostic Laboratories
- Care Coordinators

State Public Health Departments

Medical Providers

Consumers

Researchers

- Screening Laboratories
- Follow-Up Programs
- IT Teams
- EDHI Teams

- Patient
- Family
- Caregivers
- Community
- Advocacy Organizations
## Categories of Data

<table>
<thead>
<tr>
<th>Category</th>
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<tbody>
<tr>
<td>Demographics/SES</td>
</tr>
<tr>
<td>Family History</td>
</tr>
<tr>
<td>Prenatal History</td>
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<tr>
<td>Newborn Screening</td>
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<tr>
<td>Neonatal History</td>
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<td>Diagnostic Testing</td>
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<td>Past Health History</td>
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<td>Pharmacotherapy</td>
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<td>Developmental History</td>
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<td>Education</td>
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<td>Measurements</td>
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<td>Nutrition</td>
</tr>
<tr>
<td>Imaging Studies</td>
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<tr>
<td>Care Coordination</td>
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<tr>
<td>Emergency Management</td>
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### Timing of Data Capture

- **Interval:** 41%
- **Intake:** 59%
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

<table>
<thead>
<tr>
<th></th>
<th>All Elements</th>
<th>Required Elements</th>
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<tbody>
<tr>
<td>Interval Visit</td>
<td>44%</td>
<td>44%</td>
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<tr>
<td>Enrollment Visit</td>
<td>7%</td>
<td>7%</td>
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<tr>
<td>History</td>
<td>24%</td>
<td>24%</td>
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<tr>
<td>Demographics</td>
<td>8%</td>
<td>24%</td>
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</tbody>
</table>

697 Total
157 Required
Investigator – New & Existing Technologies, Novel Treatment & Management Strategies

- Enable Novel Statistically Robust Proposals

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

- Accelerate & Facilitate Research

Public Health Partner – Service Delivery & Quality Assurance/Improvement

- Implement Technologies & Assess Health Outcomes for Novel Treatments

Use Cases

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

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

Describe the relationship between service delivery and treatment methods to define optimal follow-up care plans for children with MCAD.
Use Case: Connecting Some Dots

Relationship between C8 values and genotype, specifically A985G.
Development of Data Capture Tool

♦ 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
Data Capture Screen Shot
Implementation Considerations

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

- 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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