

# Identification of A Data Set for Newborn Screening Program Assessment

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

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# Presentation Overview

- ◆ **Effective Follow-Up Initiative**
  - ◆ **Long-Term Follow-Up in the Public Health Context**
  - ◆ **Review of Consensus Core Data Set**
  - ◆ **Conclusions**
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# Effective Follow-Up Initiative

## ◆ Goal

- Utilization of electronic health information to improve the newborn screening system
- Focus on short-term and long-term follow-up

## ◆ Three-year effort in four states

- Indiana
  - New York
  - Utah
  - Colorado
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# SACHDNC Statement on LTFU

Assure the best possible outcome for individuals with disorders identified through newborn screening

## Key Features

Quality chronic disease management

Condition-specific treatment

Age-appropriate preventive care throughout the lifespan

## Central Components

Care coordination through a medical home

Evidence-based treatment

Continuous quality improvement

New knowledge discovery



# Effective Follow-Up Initiative

Allow secure and authorized exchange of health information relating to children

Create standards based data exchange interfaces

Interoperable System

Develop interoperable registries and clinical data repositories

Promote public health agencies' use of health information technology and health information exchange with clinicians



# Electronic Information Exchange

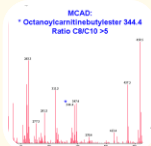
## Newborn Screening System



Prenatal Education



Screening



Diagnosis and Short-Term Follow-Up



Clinical Care and Long-Term Follow-Up

Birthing Facility	Public Health Follow-Up
Public Health Laboratory	Medical Home
Reference Laboratory	Specialty Care
Department of Health	National Databases



# Current and Emerging Standards

- ◆ **ACMG 2006 Report “Newborn Screening: Towards a Uniform Screening Panel and System”**
  - ◆ **CLSI’s “Newborn Screening Follow-Up: Approved Guideline”**
  - ◆ **HHS ONC’s Newborn Screening Use Case**
  - ◆ **NLM’s Newborn Screening Coding and Terminology Guide**
  - ◆ **Joint Committee’s LTFU Data Set**
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# Long-Term Follow-Up in the Public Health Context

## Selected Ongoing Efforts in Public Health

### CDC

- Surveillance Data Set
- Four States – NY, UT, IA, CA,

### California Department of Public Health

- STFU all positive screens
- LTFU all diagnosed cases

### New England

- Public Health Systems Approach
- Centralized Public Health Tracking

### South Dakota

- Legislated Effort
- Annual Questionnaire





# Long-Term Follow-Up in the Public Health Context

## PEAS

Long-Term Program Evaluation

Nine Assessments

## SACHDNC Statement

Overarching Questions

Families

Medical Home

State/Nation

## National Quality Forum

Newborn Screening Outcome Measure

Hearing Screening Outcome Measures

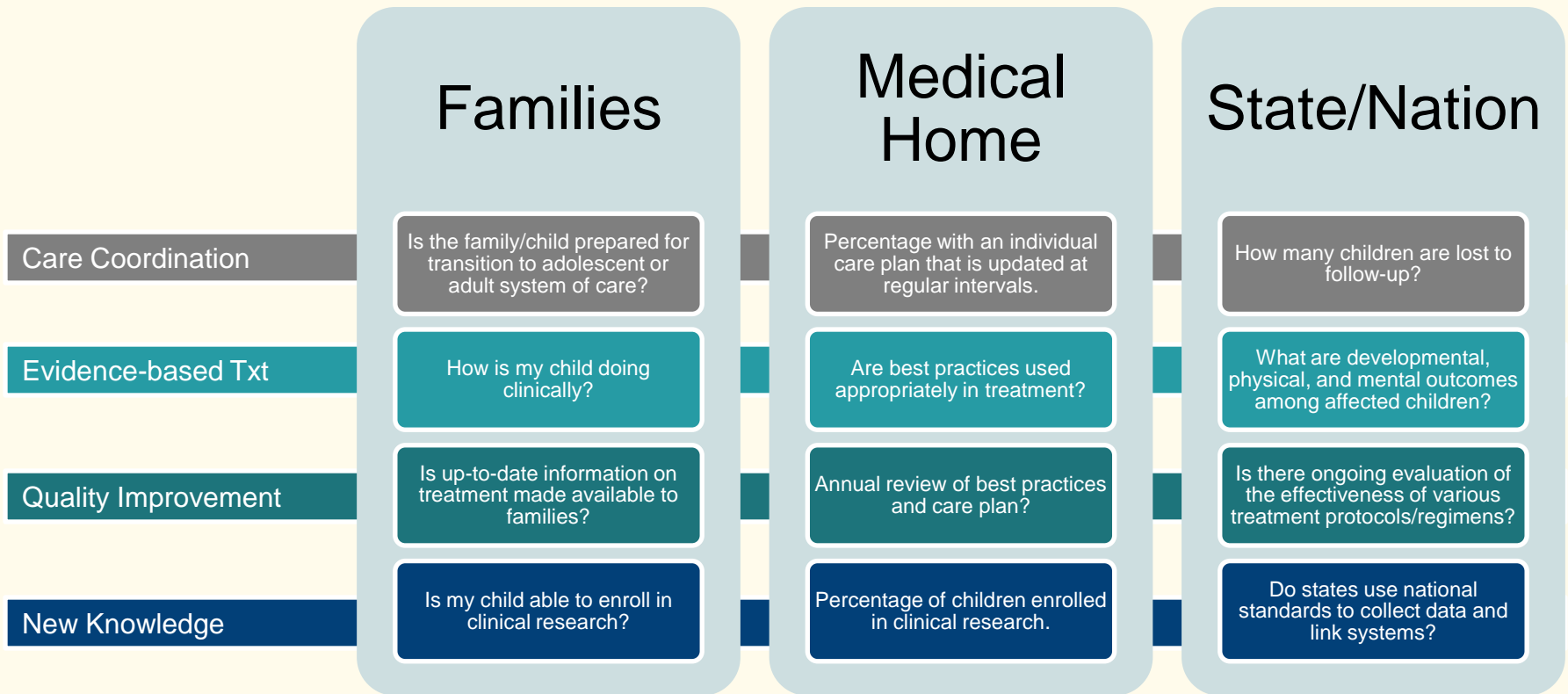


- a) A plan should exist for periodically evaluating patient progress through review of the defined outcome indicators.
  - b) Condition specific long-term outcome indicators should be developed in consultation with appropriate specialist consultation.
  - c) Long-term outcome indicators should be periodically reviewed for suitability by appropriate specialty consultants and updated as needed.
  - d) Long-term outcome data should be periodically solicited, compiled and evaluated, from families' primary care providers and/or specialists.
  - e) Long-term outcome data should be compared with expected state and national goals, and the information shared in an annual report.
  - f) Program improvements should be initiated on the basis of long-term program evaluation data.
  - g) Appropriate medical management indicators, developed by specialty consultants, should exist for each condition.
  - h) Medical management indicators should be periodically reviewed for suitability by specialty consultants and the advisory committee, and updated as appropriate.
  - i) Medical management outcome data should be periodically collected, evaluated, and reported to the NBS program's advisory committee.
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# SACHDNC Statement

## What questions should newborn screening long-term follow-up be able to answer?





# National Quality Forum

- ◆ **What percentage of infants had bloodspot newborn screening performed as mandated by state of birth?** *(NQF#1351)*
  - ◆ **Hearing Related**
    - **The proportion of births that have been screened for hearing loss before hospital discharge.** *(NQF#1354)*
    - **Percentage of children who turned 6 months old during the measurement year who had documentation in the medical record of a review of their newborn hearing screening results by their 3-month birthday.** *(NQF#1402)*
    - **Percentage of newborns who did not pass hearing screening and have an audiological evaluation no later than 3 months of age.** *(NQF#1360)*
    - **Proportion of all newborns who did not complete a hearing screening prior to discharge, went on to receive outpatient screen before the child was 31 days of age.** *(NQF#1357)*
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# Review of Consensus Core Data Set

## ◆ Primary Goal

- Identify data elements within Joint Committee LTFU data set of interest to Public Health Programs

## ◆ Secondary Goal

- Develop measures for the overall assessment of the newborn screening system through follow-up to the medical home and care coordination to submit for the National Quality Forum
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# Review of Consensus Core Data Set

## ◆ March 2011 Meeting

- **Effective Follow-Up Grantees (NY, CO, IN, UT)**
  - **Six Partner States (GA, KY, NM, MA, AR, IA)**
  - **Seven Invited States (MN, CA, NE, OK, KS, MO, MI)**
  - **NNSGRC**
  - **NLM**
  - **Joint Committee Chair and Representatives**
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# Review of Consensus Core Data Set

## ◆ Meeting Result

- Twelve Data Categories
- Eighteen Key Questions

## ◆ Proposed Next Steps

- Survey meeting attendees to match public health data categories with LTFU data set elements
  - Generate public health data set
  - Disseminate public health data set
  - Finalize public health data set
  - Pilot
  - Develop assessment measures
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# Data Categories Identified

- ◆ **Subjective Summary – Well or Not**
  - ◆ **Continuity of Care**
  - ◆ **Patient Tracking**
  - ◆ **Physical/Growth Parameters**
  - ◆ **Access/Barriers to Care**
  - ◆ **Services**
  - ◆ **Health Status**
  - ◆ **Review of Systems**
  - ◆ **Disorder Related Interventions**
  - ◆ **Developmental Assessment**
  - ◆ **Patient Self Assessment**
  - ◆ **Data Quality**
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# Identified Questions

- ◆ Are we preventing or reducing morbidity/mortality without additional harm?
  - ◆ Is there universal access to the program?
  - ◆ Are we doing this in the most effective way?
  - ◆ Are we doing this in the most cost effective way?
  - ◆ Is the disorder on newborn panel?
  - ◆ What percent of children with disorders remain in care between the ages of one and five years old?
  - ◆ What percent become lost to follow-up?
  - ◆ What percent of parents refuse treatment?
  - ◆ What percent died due to problems associated with the disorder?
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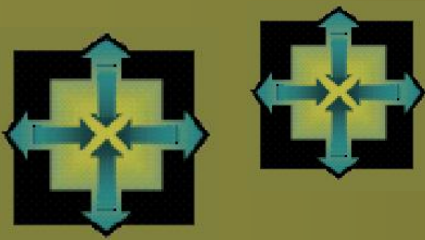
# Identified Questions

- ◆ What percent were determined not to need ongoing treatment?
  - ◆ What percent of children (combined or by specific type of disease) had age appropriate developmental status with respect to speech, physical development, mental/cognitive development, gross motor and fine motor development?
  - ◆ What percent of children were severely delayed with respect to any of the developmental measures and what year of life did the delays become apparent?
  - ◆ What percent of patients experienced symptoms associated with their disorder and at what age did the symptoms become apparent?
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# Identified Questions

- ◆ In any given year, what percent of children experienced the loss of skills they had previously acquired?
  - ◆ What percent of children had no hospitalizations or emergency room visits in the previous year of life?
  - ◆ What disorders are associated with the greatest number of hospitalizations and emergency room visits due to disorder-related complications?
  - ◆ What disorders are associated with the highest utilization of metabolic center visits?
  - ◆ What percent of children are receiving a multidisciplinary team of services, including nutritional counseling, health education and social services counseling?
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# Use Cases

## 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.



## Conclusion

- ◆ **Data categories and key questions that are of interest to Public Health Programs were identified**
  - ◆ **The Joint Committee LTFU Data Set is a useful source for the information to answer the key questions**
  - ◆ **Effort lays the foundation for development of measures for overall assessment**
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