

Long-Term Follow-up After Newborn Screening: Environmental Scan



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August 2, 2018

Defining LTFU

- All care and special services that individuals receive after being diagnosed with a condition through NBS
- Program evaluation and research

Four Components of Long-Term Follow-up

- Care coordination through a medical home
- Evidence-based treatment
- Continuous quality improvement
- New knowledge discovery

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A lot has been
accomplished



Key Aspects to Consider

- Wide variety of stakeholders
 - Patients and families
 - Public health
 - Specialty and primary care
 - Payers
 - Drug and device manufacturers
 - The wide variety of others that patients and families intersect with over the lifespan
- Variation in conditions
 - Epidemiology, treatment, timing
 - Accessibility of experts
 - Cost
 - Knowledge
- Funding for LTFU
- Authorization

Central Themes

- It can be complicated to know how to get to where you want to be
- Often the route is indirect and requires multiple lines



Objective

- To conduct a landscape review to help inform the ACHDNC about opportunities for improving LTFU

ACHDNC LTFU Activities

- What questions should newborn screening long-term follow-up be able to answer? *Genet Med.* 2011;861-865
 - Patient-family level
 - Medical care/medical research level
 - State/national level
- Some examples from the LTFU subcommittee
 - Standardized terminology
 - Quality metrics
 - Models, including information exchange

As you listen to this overview

- Focus on activities relevant to newborn screening
- Not condition specific

Methods

- Technical Expert Panel and Stakeholder Input

Technical Expert Panel	Stakeholder Group
Amy Brower, PhD	
American College of Medical Genetics	Researcher
Christine S. Brown, MS	
National PKU Alliance	Consumer
Susan A. Berry, MD	
Div of Genetics & Metabolism, Univ of MN	Researcher
Marci Sontag, PhD	
Director, NewSTEPS 360	Researcher
Sylvia Mann, MS, CGC	
Genomics Section, Hawaii Dept of Health	State Public Health
Jennifer M. Kwon, MD	
Univ of Rochester – Neurology and Pediatrics	Specialty Care Provider
Tony Steyermark, PhD	
Newborn Child Follow-Up Program Minnesota Department of Health (MDH)	State Public Health

Findings from the TEP

- Most NBS are involved with LTFU, but the extent and nature is variable
 - Sometimes LTFU is imbedded and indirect (e.g., referral contracts with specialists)
 - The absence of responsibility or authority limits what NBS can do
- The time horizon is challenging
- Engaging families is critical

Horizon Scan: Categories

1. Recommendations about how to conduct LTFU
2. Prospective studies of data collected by NBS programs for specific purpose of LTFU
3. Prospective studies of data collected outside of NBS programs for reasons other than LTFU
4. Retrospective studies of existing data collected for research or non-research purposes

Conduct of Long-Term Follow-up

- How it should function
 - Questions LTFU should answer
 - Business Process Analysis
 - Data requirements for Registries
- Specific recommendations for making LTFU more feasible
 - Linking screening results from point-of-care testing
 - Linking bloodspot collection device serial numbers to birth certificates
- Regional collaborative partnerships, describing approaches to LTFU
- Newborn screening program surveys
 - Data collection
 - Follow-up activities
 - Barriers

Conduct of Long-Term Follow-up: Lessons

- LTFU is well defined
- Models for conducting LTFU, including illustrating data exchange
- Focus of the Regional Collaboratives
- Concerns about funding and sustainability often raised
- Quality metrics can promote meaningful LTFU. There is a tension between condition-specific vs. more general measures

Prospective Studies in NBS

- Wide range of conditions conducted in many different countries
- Study design
 - Most large cohort studies following individuals identified through newborn screening, but some included a comparison population
- Most go up to about 6 years of age
- All focused on predictors of outcomes

Prospective Studies in NBS: Lessons

- Data collected by newborn screening programs for LTFU can be used to evaluate specific hypotheses
- More information about how these projects were developed and maintained would be helpful

Prospective Studies of Data Collected Outside of Newborn Screening

- One study of hearing screening in the UK was captured in the search
- However, there are disease-specific registries that collect long-term follow-up data that can be linked back to newborn screening, for example:
 - Pompe disease registry (Sanofi Genzyme)
 - Cystic Fibrosis Registry (CF Foundation)

Prospective Studies of Data Collected Outside of Newborn Screening: Lessons

- Prospective studies are uncommon
- Disease-specific registries might be under-utilized for prospective research

Retrospective Studies

- Two general types
 - Chart audits in treatment centers
 - Data linkage studies
 - Newborn screening data
 - Vital records (e.g., birth and death certificates)
 - Healthcare claims data
 - Etc.

Retrospective Studies: Lessons

- Efficient, especially for rare disorders, but
 - Time delay
 - Linkage can be difficult
 - Claims data can be incomplete or inaccurate
 - Better knowledge of the association between intermediate measures and long-term outcomes would be helpful

Examples of Long-Term Follow-up Activities

- Not meant to be complete but illustrative

State Activities

- California: Web-based Screening Information System, which among other functions than facilitate referral tracking and coordination with specialty-care follow-up centers. These centers provide follow-up through 5-years of age using an Annual Patient Summary
- Colorado (CRCSN): Legislative requirement mandating reporting of birth defects and other newborn disorders, facilitating linkage of data sets (vital statistics, hospital data)

State Activities

- Illinois: Annual NBS report based on data collected for children through 15 years of age
- Minnesota: Dedicated long-term follow-up supervisor; engaged in tracking a wide arrange of outcomes, including parent-reported developmental status

Regional and National LTFU Activities

- Focus of the National Coordinating Center and the Regional Genetics Networks (e.g., data collection, evaluation and quality improvement)
- NBS Connect – patient registry/portal, focusing on inherited metabolic disorders
- Newborn Screening and Translational Research Network – facilitates research
- Longitudinal Pediatric Data Resource, developed by the NBSTRN, data repository for specific disorders (e.g., SMA) built on REDCap)
- NewSTEPS – data reporting infrastructure and specific short-term quality indicators

Summary

- Two distinct but related aims
 - Assuring care
 - Program evaluation and research
- Opportunities
 - Standardizing LTFU measures, focus on quality metrics
 - Patient
 - Population
 - System
 - Expanded use of registries
 - Expanded support for retrospective research
 - Datasets, linkages, risk of bias assessment

Questions?

