

The Role of Quality Measures to Promote Long-Term Follow-up of Children Identified by Newborn Screening Programs

Presented by the Follow-up and Treatment Workgroup to the Advisory Committee on Heritable Disorders in Newborns and Children on February 8, 2018.

Executive Summary

Newborn screening (NBS) is a public health success, saving lives and improving health and developmental outcomes of children across the United States. NBS has been defined as including the components of long-term follow-up (LTFU): care coordination, evidence-based treatment, continuous quality improvement, and new knowledge discovery. Quality assessment (QA) and quality improvement (QI) are essential to maintain and improve outcomes through NBS LTFU. This intent of this report is to focus on the use of quality measures to assess and to help drive the success of NBS using NBS LTFU.

Quality measures are specific, well-defined indicators of quality that help support accountability and improvement of health care implementation and outcomes. Measures are defined by a numerator, denominator, inclusion criteria, and exclusion criteria that are developed, tested, and maintained by a designated measure steward. Quality measures may take a wide range of forms to address different quality goals, including improvement, regulation, accreditation, performance reporting, and surveillance. The ACHDNC Follow-up and Treatment Workgroup supports the concept that the use of quality measures is central to improving quality in LTFU of conditions identified through NBS. This document summarizes the findings and recommendations of the Follow-up and Treatment Workgroup, focusing on the disorders that are identified through NBS.

Development of quality improvement measures for LTFU of individuals who have been identified through NBS programs ought to be applied at NBS state program, care provider, practice, and system levels. These measures could drive QI in care and provide a tool for consistent data collection documenting current care practices and identifying gaps. Implementation of QI activities using these measures at each of these levels is anticipated to engage those caring for children in collecting data. Data on the quality measures for each condition may be aggregated through advances in health information technology (e.g. electronic medical record exchanges and interoperability standards).

Case studies collected from across the U.S. illustrate that well-defined quality measures applied to specific disease conditions may be implemented at a variety of levels for data collection, including national, regional, state, and institutional. Moreover, data may be obtained through means that are public, private (e.g. payer-guided), or a mix. Quality improvement efforts have been applied to selected rare diseases with benefit from large centrally maintained databases that accept and retrieve data for interested parties to use. Security and steady funding for these databases need to be assured. Collaboration between specialists, primary care providers and emergency departments and between institutions can lead to robust data collection and quality measure implementation. Incentives to participate include recognition as a center of excellence and direct financial benefit. In contrast, incorporation of quality activities into routine medical care and electronic health records through standardization across platforms can be challenging.

Several examples of the use of quality measures through provider implementation and database tracking are described in the accompanying report. Examples include government and privately-funded programs for tracking one disease (e.g. cystic fibrosis) or multiple diseases simultaneously (e.g. metabolic conditions through multi-state data sharing). Accomplishments, challenges, and lessons learned are described for each example.

Despite these examples of success, quality measures remain underused. No national standard exists for the use of quality measures in LTFU by state NBS programs or other parties. Relatively few disease-specific quality measures for NBS conditions are used because gaps in the evidence must be filled to develop specific measures. Even when quality measures are available, most are not routinely applied, in large part because data collection is cost- and resource-intensive and their value is not sufficiently interwoven into the elements of health care services where the impact of measures would be appreciated. Furthermore, communication of information among specialists, primary care providers, and public health agencies remains a critical challenge. Finally, there is a need to move beyond disease-specific measures and to include the patient's and family's perspectives in measuring quality.

Existing resources and standards could accelerate the use of quality measures for NBS. The Office of the National Coordinator for Health Information Technology (ONC) works with CMS and AHRQ to maintain an Electronic Clinical Quality Improvement Resource Center (<http://ECQI.HealthIt.gov>). Fast Health Interoperability Format (FHIR) is facilitating development of interfaces with EHR. The APHL NewSTEPS program (HRSA U22MC24078) has developed case definitions and a national reporting repository that can help define the denominator of affected infants for NBS quality measures. The NBSTRN's LPDR is available as a REDCap

database with definitions of data fields and including core, disease specific, and public health variables. Clinical decision support tools which prompt the clinician to enter appropriate information for LTFU of their specific patient's disorder in the EHR during a clinical encounter could be developed to facilitate data collection. Gaps in funding for all aspects of LTFU including treatment, monitoring, development of measures and networks to share tools and data, remain a concern that limits the use of quality measures.

Potential next steps include:

1. Make the case for the importance of prioritizing development and use of quality measures at multiple levels and systems of care for LTFU of NBS as a strategy for engaging a broad range of stakeholders including Federal, State, Provider, and Consumer groups to participate in LTFU of NBS.
2. Identify a core set of long term follow-up quality measures and associated data resources for conditions identified by newborn screening that will maximize existing collaborative efforts by groups such as The Association of Public Health Laboratories Newborn Screening Technical assistance and Evaluation Program (APHL/NewSTEPS), The Newborn Screening Translational Research Network Longitudinal Pediatric Data Resource (NBSTRN/LPDR), and The National Coordinating Center for Regional Genetics Networks (NCC) to gather uniform LTFU data from more states and other organizations.
3. Encourage the use of large data collection activities such as the National Survey of Children's Health (NSCH) and quality improvement activities such as Medicaid quality reporting and HEDIS to provide data on LTFU of NBS by identifying cohorts of children with disorders identified by NBS.
4. Work with key stakeholders, such as consumer advocates and professional associations to leverage research networks that collect data from patients and families to participate in quality measure development and quality improvement activities targeted to LTFU of NBS.
5. Assist the use of new Health Information Technology (HIT) standards for implementing and sharing quality measures as a strategy for integrating quality measures into routine care and using Clinical Decision Support (CDS) in the EHR to capture data and guide care.

We recognize that the availability of resources will promote (or limit) the pace of taking each of these next steps to disseminate the lessons we have learned.

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I. Overview

The goal of newborn screening (NBS) is to reduce morbidity and mortality in newborns and children by early identification and treatment. Since the 1960s, when state-based NBS programs were created to ensure that NBS is universally available in the U.S., thousands of individual children and their families have benefited. More than a decade ago, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recognized that long-term follow-up (LTFU), including treatment, is essential to the success of NBS. Through a series of publications, the ACHDNC has promoted a framework for assessing outcomes. In the spring of 2016, the Follow-up and Treatment Workgroup formed a sub-workgroup to investigate the role of quality measures for assuring quality in long-term follow-up of NBS conditions. This report defines quality measures and their relevance to LTFU, describes case studies, summarizes key findings, and provides recommendations for next steps.

II. What are Quality Measures?

Quality measures are specific, well-defined indicators of quality defined by a numerator, denominator, inclusion criteria, and exclusion criteria that are developed, tested, and maintained by a designated measure steward. The primary purpose of quality measures in medicine is to assess structure, processes, or outcomes relating to care. Measurement serves as an important step in identifying areas for improvement, interventions, and need for subsequent cycles of monitoring. It is also useful for those who design, implement, and evaluate proactive quality improvement (QI) strategies such as clinical decision support and real-time reminder systems to adhere to quality standards. Quality measures may take a wide range of forms to address different quality goals, which can include improvement, regulation, accreditation, performance reporting, and surveillance.

A number of national organizations develop quality measures for both adults and children, including the National Committee for Quality Assurance (NCQA) and the American Medical Association's Physician Consortium for Performance Improvement (AMA-PCPI). The AHRQ Pediatric Quality Measures Program (PQMP) is the one of several national programs focused on pediatric-specific measure development and implementation; others include the National Institute for Children's Health Quality (NICHQ) and the American Academy of Pediatrics (AAP). The National Quality Forum (NQF) is a non-partisan organization that serves as a primary endorsement body for quality measures which have been defined and tested for a specific use or level of aggregation. The NQF evaluates measures to ensure that they are evidence-based and scientifically sound.

Given the growing focus on the value and costs of care for the Centers for Medicare and Medicaid Services (CMS) and other public and private health care entities, quality measurement and quality improvement are quickly becoming integral parts of health care delivery. Quality measurement is a tool for advancing high-quality and safe health care for children and, more recently, for value-based payments and incentives. Quality measures have also become a part of maintenance of certification and licensure requirements and the Meaningful Use incentive program for electronic health records (EHR). Health Information Technology (HIT) will continue to be a critical component of quality measures and quality improvement as it has the promise of reducing data collection burden on providers and systems (Appendix 1). The changing climate of health care and public health is driving not only the uptake of measures, but also the use of measures to evaluate performance and improve care at multiple levels of care, including providers, health plans, hospitals, states and nationally.

For the purposes of this report, we will use the term “quality measures” as a broad umbrella term. As such, we consider other terms such as “performance measures” and “accountability measures” also fall under the broader category of “quality measures.” Defined this way, quality measures can be used to demonstrate a variety of activities/processes and health care outcomes for particular populations. The use of quality measurement helps strengthen accountability and support performance improvement initiatives at numerous levels.

III. Prior Work of the ACHDNC Follow-up and Treatment Workgroup

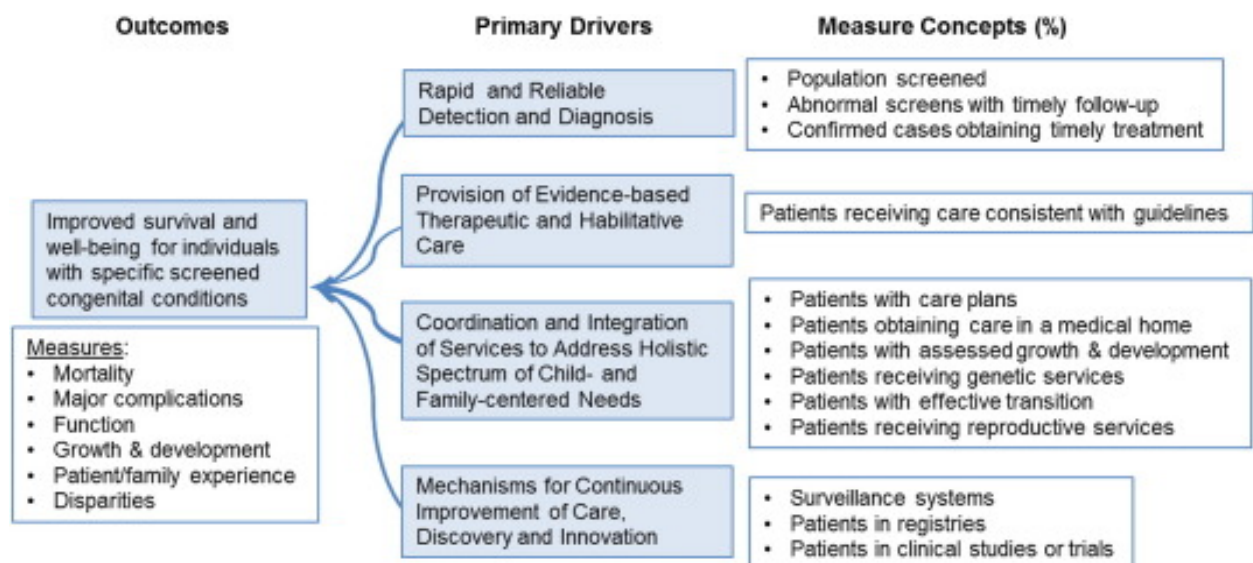
Writing for the ACHDNC, in 2008 Kemper et al¹ defined LTFU (Figure 1) and described its three key components: the assurance and provision of quality chronic disease management, condition-specific treatment, and age-appropriate preventive care throughout the lifespan of individuals. To achieve these goals, they recommended attention to care coordination through a medical home, evidence-based treatment, new knowledge discovery, and continuous quality improvement. In 2011 Hinton et al² built on this work to investigate what questions should NBS long-term follow-up data collection and analysis be able to answer (Appendix 2). They added that it is essential to include the different perspectives of families, state and nation, and primary and specialty health care providers. Most recently, Hinton et al³ created a framework for assessing outcomes (Figure 2); the authors described how to apply the framework to specific examples of NBS conditions, phenylketonuria and sickle cell disease.

Figure 1. Definition of Long Term Follow-up for Newborn Screening

Fundamentally, long-term follow-up comprises the assurance and provision of quality chronic disease management, condition-specific treatment, and age-appropriate preventive care throughout the lifespan of individuals identified with a condition included in newborn screening. Integral to assuring appropriate long-term follow-up are activities related to improving care delivery, including engagement of affected individuals and their families as effective partners in care management, continuous quality improvement through the medical home, research into pathophysiology and treatment options, and active surveillance and evaluation of data related to care and outcomes.

Kemper et al. 2008¹

Figure 2. The driver diagram establishes the elements and primary goals needed to attain optimal outcomes for children diagnosed through public health newborn screening.³



Per the ACHDNC, in 2016 members of the Follow up and Treatment Workgroup came together to work on a new charge from the ACHDNC with the goal of investigating the role of quality measures in improving long-term follow-up of NBS. Several objectives emerged in the workgroup's deliberations:

- Draft a background document to describe what is known about quality measures and newborn screening and identify areas of need and possible opportunities to use clinical quality measures in long-term follow-up.
- Develop case studies that provide examples of how quality measures are used in newborn screening – describe success or barriers.
- Describe other key findings (e.g. use of quality measures vs performance measures, and different approaches for disease-specific measures vs public health services vs patient/child specific measures).

To organize the workgroup's findings, we have identified four levels at which quality measures and improvement efforts might focus to improve outcomes (Figure 3). Each of the four levels offers opportunities for measuring and improving outcomes, and many quality measurement and improvement efforts already happening across the U.S. Relevant case studies are summarized below with a brief summary for each of important takeaways or lessons learned that can be useful for considering next steps for the workgroup.

(1) **Specific condition.** A child born with a NBS condition will require medical and educational interventions related to that specific condition (e.g. daily penicillin for a child with sickle cell disease). Specific medical conditions often have formal performance measures and organized research networks, sometime with the participation of patient/family advocacy groups. The Newborn Screening Translational Research Network (NBSTRN) worked with stakeholders to develop the Longitudinal Pediatric Data Resource (LPDR), a tool to help collect data on NBS conditions in particular.

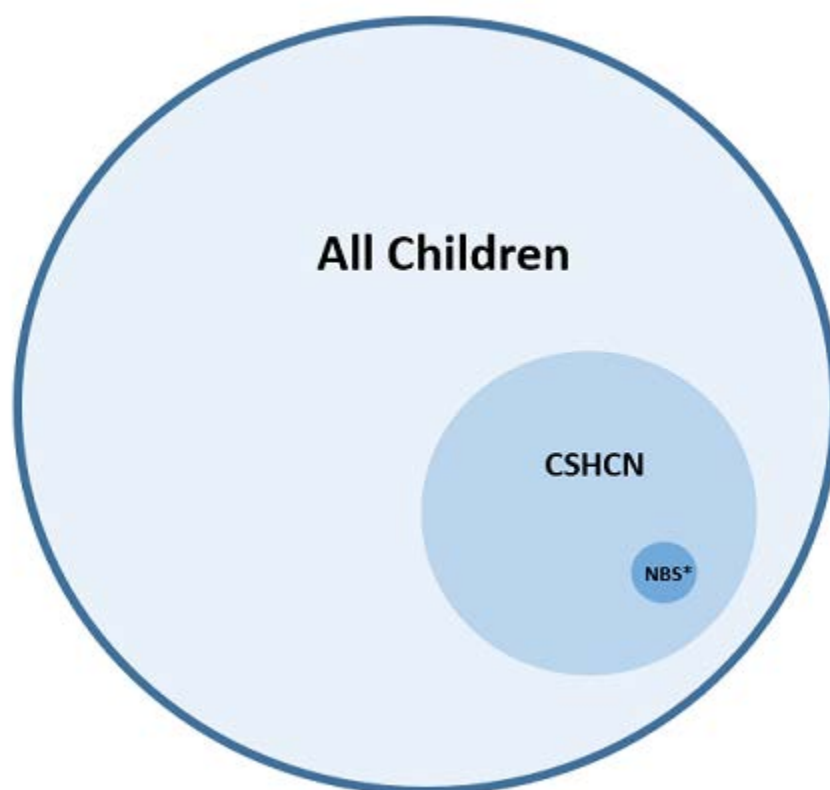
(2) **All Conditions Identified by NBS.** A variety of state and national organizations, such as state NBS screening programs, monitor the short-term and selected long-term outcomes of children identified with a NBS condition. Collaborative efforts include LPDR's dataset of public health questions, the Association of Public Health Laboratories (APHL)/NewSTEPS program, and the National Coordinating Center (NCC) that supports the Regional Genetics Networks.

(3) **Children with Special Health Care Needs (CSHCN).** Children identified through NBS are part of a larger population of children who require more medical, behavioral, or educational interventions than a typical child. As such, they may experience challenges in accessing services and community participation, and their outcomes may be monitored through mechanisms such as the National Survey of Children's Health (NSCH).

(4) **All Children.** Many quality measures that are developed for all children (e.g. vaccination according to national standards), such as HEDIS (Healthcare Effectiveness

Data and Information Set), can be aggregated at multiple levels of care delivery such as providers, hospitals, health plans and state or nationally. These quality measures can also apply to children identified with NBS conditions.

Figure 3. Organization of Quality Measures for Newborn Screening.



*e.g. sickle cell disease, cystic fibrosis, congenital hypothyroidism, medium chain acyl-CoA dehydrogenase deficiency (MCADD). For a list of all conditions on the Recommended Uniform Screening Panel, see Appendix 3

IV. Case Studies

Case studies illustrating the use of quality measures for LTFU, including successes and challenges, are divided into sections according to the four levels at which quality measures and improvement efforts might focus to improve outcomes: (1) specific conditions, (2) all conditions identified by NBS, (3) children with special health care needs, and (4) all children. Some case studies could fit in more than one of these sections. While few of these case studies have formally developed and validated quality measures, case studies were included if the study, program, or project reported on measurable performance or outcomes measures. Some of these case studies are time limited projects and some are ongoing programs. Some collect data from single programs or single states, and some are national. Strategies for collection of data vary, including wide variation in ascertainment of cases. This variability is recognized in the description of lessons learned and in considering implications for future efforts in development and use of quality measures for improving LTFU. The case studies are summarized in Table 1 for specific conditions and in Table 2 for quality measures that apply to population beyond those with specific newborn screening disorders. The tables are at the end of the case study section.

1. Specific Conditions

A. Sickle Cell Disease Measures as an Example of Tracking Proven Therapies in Use

Primary national/systematic efforts to utilize quality improvement methods in sickle cell disease have been coordinated through Health Resources and Services Administration's (HRSA) Sickle Cell Disease Treatment Demonstration Program (SCDTDP), coordinated through the National Institute for Children's Health Quality. Through learning collaboratives, grantee teams including primary and specialty care providers learn improvement methodology that was applied to grant-funded activities. Per the 2014 Report to Congress⁴, this resulted in decreases in emergency department wait-times for treatment, increase in the number of primary and specialty care visits at funded sites, increased newborn screening follow-up activities for sickle cell trait, and development and testing of tools for youth transition.⁵ Program activities shifted in the subsequent funding cycle to seek administrative data from state Medicaid and other programs with a focus on use of hydroxyurea (HU), the only Food and Drug Administration (FDA)-approved disease-modifying therapy. Given several barriers to access these data, emphasis has returned to local practice-based application of QI methodology.

Some sites involved in the Pacific Sickle Cell Regional Collaborative are also participating in the AHRQ IMPLementing Measures NeTwork for Child Health Network (IMPLEMENT)⁶. This grant program seeks to evaluate and refine measures for utilization of Transcranial Doppler and hydroxyurea as a part of the PQMP.

Of note, QI activities are conducted by collaborative participants supported by a coordinating center with expertise in quality improvement strategies in settings with a sufficient number of affected individuals to motivate local health care systems to consider small cycles of change. It is anticipated that providers and systems have new/expanded skills to implement change, but the extent to which activities continue without grant funding and applicability to non-SCDTDP sites, especially those with fewer affected individuals, requires further evaluation.

Lessons Learned: The use of quality measures has revealed that: (1) therapies such as immunizations and prophylactic antibiotics that have strong evidence showing effectiveness have not been used for all children with sickle cell disease at the optimal time; (2) programs for quality improvement can create higher compliance rates; (3) sickle cell disease is cared for in a variety of settings; and (4) it is important to encourage cooperation and engagement of primary care, specialists, and emergency physicians to optimize care for this condition. This example

also demonstrates how long-term funding can support data collection, analysis, and quality improvement activities.

B. Registry and Surveillance System for Hemoglobinopathies (RuSH)

Since 2010, the Centers for Disease Control and Prevention's Division of Blood Disorders has been coordinating activities to develop state-wide surveillance systems for sickle cell disease (SCD). Initially, seven states were involved in the project. The current iteration, the Sickle Cell Data Collection program is comprised of two states, California and Georgia. The data for these surveillance systems is collected from a variety of sources, including newborn screening records, hospital discharge data, emergency room records, death records, clinical records, and state Medicaid claims. Information from a number of clinical centers who provide specialty care for patients with SCD is also included. Patients with SCD are identified in each of these data sources based on laboratory results and/or International Classification of Diseases, Clinical Modification, Ninth Revision and Tenth Revision (ICD-9-CM and ICD-10-CM, respectively) codes. At this time, surveillance data from 2004 through 2015 has been collected by the two participating states.

The purpose of surveillance for SCD is to document health information over time, so as to identify gaps in diagnosis, treatment, and healthcare access, which will hopefully lead to improved health outcomes and longer lives for patient with SCD. These improvements will be brought about by increased understanding of the disease, policy changes, and improved healthcare practices.

Lessons Learned: (1) this method of longitudinal data collection, especially for a patient population that often receives healthcare outside of specialty care centers, provides a comprehensive understanding of the entire spectrum of patients; (2) this surveillance system has provided helpful information to strengthen other projects, such as HRSA's SCDTDP program and NHLBI's Implementation Research project; and (3) maintaining consistent and long-term staffing, data sharing agreements, and funding are paramount to the success of this program.

C. Cystic Fibrosis Foundation Comparative Outcomes

One of the best examples of new knowledge discovery through quality measures has been the work of the Cystic Fibrosis Foundation (CFF) to compare clinical outcomes, treatment modalities, and morbidity between accredited care centers nationally. Over 48,000 unique patients have been reported to the Cystic Fibrosis Foundation Patient Registry (CFFPR) since

its inception in 1986⁷. Data collection within the CFFPR has evolved, beginning with annual reporting on demographics and basic patient outcomes to its current online format collecting encounter based data on clinical measurements, treatments, and complications related to cystic fibrosis. Development, maintenance, and user support of the CFFPR is provided by the CFF. Accredited CF Centers are supported financially by the CFF to provide quality care through a team approach, complete data entry into the CFFPR, and utilize the data for local quality improvement initiatives⁸. Patient participation in the CFFPR is through consent and is overseen by local institutional review boards.

The utility of the CFF Registry has been demonstrated through epidemiologic reports, comparative studies, and quality improvement initiatives. Aggregate reports on the data have been published since 1988, and center specific reports were first produced in 1999. Quality metrics from the CFF were made public in 2006, in parallel with quality improvement initiatives designed to share successful strategies between care centers. Clinicians are able to download patient specific reports to track longitudinal outcomes for both clinical care planning and patient education⁸. It is estimated that 80% of persons with CF in the U.S. were reported to the CFFPR in 2012. Loss to follow-up, as measured through the CFFPR, is low in the CF Center model, at less than 10% of patients in 2012-2013. In aggregate, data from the CFFPR has also been useful in studying the impact of newborn screening using long-term follow-up data⁹.

Lessons Learned: Longitudinal and transparent application of quality measures has demonstrated that: (1) CFF accredited centers were initially reluctant to allow transparency of their data, but accepted the change and have shared care strategies between centers for quality improvement purposes; (2) families who are engaged in clinical care at a CFF accredited center will partner with personnel at that center in order to improve care and the resulting quality measures; (3) understanding the course of the disorder over the lifespan requires well-defined and consistent measures collected across centers.

D. California Newborn Screening Follow-Up of Cystic Fibrosis:

The California Department of Public Health, (CDPH) Genetic Disease Screening Program (GDSP) successfully initiated newborn screening for Cystic Fibrosis in 2007 utilizing a tiered IRT-DNA-DNA screening methodology. Legislatively supported fees are collected for every child born and screened in California. These funds are used to support follow-up services provided at 15 state-contracted CF specialty care centers that order confirmatory testing and other diagnostic services for screen-positive cases. Short term follow-up data, as well as long-term follow-up data collected on children with confirmed CF or cystic fibrosis related metabolic

syndrome (CRMS) is entered by the state-contracted centers through a secure web-based computer system, called the Screening Information System (SIS). Long term follow-up data is collected yearly through age five using SIS computer screens (see Appendix 4). GDSP's passive, but robust follow-up data collection about children with CF has allowed the publication of papers (listed in Appendix 4) spanning diagnosis and genetic analysis of children as well as longer-term identification of CF among CRMS cases that were followed by the program within the 5-year time frame.

Lessons Learned: (1) Short- and long-term follow-up data collection about children with CF along with a burgeoning genetic database has helped inform appropriate diagnosis and interventions for CF through information sharing, scientific collaboration and publication; (2) California has leveraged its relationships with the CF Foundation-approved centers and utilized these state-contracted centers to provide patient access to centers of excellence for lifelong CF care; and, (3) follow-up data collection and analysis has been successfully used to provide feedback to local practitioners to improve timeliness of diagnosis and information on other outcomes of interest.

Collection of detailed and validated data with little ascertainment bias in this program benefits from the collaboration with nationwide private support from the CF Foundation for centers of excellence, so that replication of this model for other conditions would be challenging. In addition, California provided funding for specific data collection and analysis. While this California model has been extremely effective and generated considerable useful data, other states may not be able to replicate this approach due to lack of funding and there is a need to explore more limited strategies that can be replicated elsewhere. Another limit of this model is collection of data only to age 5 years, before many of the long-term effects of CF (and of other conditions identified by NBS) become apparent.

E. Mountain States Genetics Collaborative medium chain acyl-CoA dehydrogenase deficiency (MCADD) QI Study

This was a provider-initiated, quality improvement study at the Inherited Metabolic Diseases (IMD) clinic at the Children's Hospital Colorado (CHCO) that successfully integrated an MCADD checklist into the Epic EHR to ensure that important management information was given to families. Prevention of fasting and emergency management during illness are the mainstays of therapy for individuals with MCADD. Currently recognized measures of care in this patient population include: (1) provision of an emergency medical letter that outlines initiation of care during illness; (2) discussion of appropriate lengths of fasting times; (3) provision of home

management recommendations typically via a home Polycose protocol or alternative; and, (4) recommendations for Medic Alert bracelet or tag and, when appropriate, provision of car seat stickers. With multiple physicians involved in the care of patients, it was unclear if all were consistent in providing these recommendations.

A chart review of the electronic medical records of a subset of patients with MCADD followed in the IMD clinic (N=11) occurred. Charts were reviewed by two IMD physicians to assess the information noted in Table 1 Appendix 5, with the table also demonstrating the results of the review.

The goal of the project was to ensure all individuals with MCADD had emergency management procedures in place. The chart review identified which measures were consistently in place. To address the deficiencies identified, charts were updated with a patient highlight notification (Pop Up) so the diagnosis and the location of the emergency medical letter were easily evident, and an Epic smart phrase was created to be utilized in patient documentation that provides a check list of important safety measures to be discussed and documented (see Appendix 5). This project also identified the need for a standardized anesthesia letter which has consequently been implemented. Overall, this project greatly helped to ensure all providers are giving consistent information to their shared patients to help ensure patient safety¹⁰.

Lessons Learned: (1) Integrating quality activities into routine care is feasible and can overcome the need for separate funding and duplicate data entry; (2) display of important newborn screening condition related information at the time of an emergency room visit or when seeing a new provider is feasible if appropriate care plans and data collection forms are integrated into an EHR (and assuming the new provider has access to that same EHR); and, (3) such quality activities for one screened disorder can be modified to become applicable to other disorders. For example, the MCADD checklist and smart phrase can be modified for very long chain acyl-CoA dehydrogenase deficiency, glutaric acidemia, type I, or 3-methylcrotonyl-CoA carboxylase deficiency.

F. The Centers for Disease Control and Prevention (CDC) Early Hearing and Detection Intervention (EHDI) Measures Approved by NQF

Based on established international standards, reporting of clinical quality measures of newborn hearing screening has been developed and coordinated by the CDC EHDI program.¹¹ Progress towards the national EHDI benchmarks (screening no later than age 1 month, audiologic testing no later than age 3 months and enrollment in early intervention no later than

age 6 months) is measured by Healthy People 2020 Objective ENT-VSL-1 and three child health quality measures that have been endorsed by the NQF since 2011.

An assessment of more than 1,000,000 newborns conducted for CDC's EHDI program revealed that State capability to capture screening results before hospital discharge is high with 98% percent of reported births being screened. However, in an effort to provide an audit of screening, diagnosis, and intervention, it became clear to CDC that health information technology standard based electronic messaging would reduce manual entry error and missed babies and provide a clear provenance of the data. This would allow states to track babies and results at each step of the process beginning with screening. To this end, Integrating the Healthcare Enterprise's (IHE), Quality, Research and Public Health Committee produced a Technical Profile entitled Newborn Admission Notification Information (NANI). IHE NANI is an implementation guide comprised of specific Admission, Discharge, Transfer messages, modified to apply to newborns with data elements critical to Newborn Screening Programs. NANI was initially written for EHDI programs but is broadly applicable to newborn screening. NANI provides states with an accurate denominator of hospital births in real time and thus is the baseline to capture other quality indicators. NANI has been implemented in approximately 240 hospitals directly to the State's EHDI Information Systems (IS); indirectly via a state Health Information Exchange (HIE) to the EHDI IS and even from the EHDI IS back to the HIE and to other newborn programs needing similar information.

By providing the denominator, and then assessing how many newborns had documented screening outcomes, the next stage of the quality indicator could be evaluated more accurately since each newborn's data could be accounted for and tracked. Such auditable data revealed that while many newborns were moving on to diagnosis, a large population needing diagnosis appeared to be lost to documentation. Similarly, the numbers identified as needing intervention was higher than the number known to receive it. The NANI denominator is especially helpful in determining babies that did not receive a mandated screen. These infants would otherwise not be known to public health in a time that meets the timeliness goals for newborn screening and before the onset of symptoms. Building the initial denominator of births to identify infants to screen is the first step in tracking follow-up data to diagnosis.

Lessons Learned: (1) Custodianship and maintenance of measures is important; and, (2) more organizations need to be encouraged and facilitated in navigating the process of creating standardized, validated, measures. 3) Ascertainment bias or loss to follow up for data collection is a challenge to the quality improvement system.

G. Long-term Follow-up of Newborn Screening for Primary Congenital Hypothyroidism: A Pilot Study in California

The California Department of Public Health, Genetic Disease Screening Program tracked outcomes for the most common newborn disease, Primary Congenital Hypothyroidism (PCH), as part of a 3-year pilot study that was funded by HRSA. On a routine basis, the GDSP long-term follow-up data collection model relies upon data collected by state-contracted endocrine specialty care centers that enter data via the GDSP Screening Information System (SIS); however, the program has contracts with only ~50% of the endocrine centers in the state and thus, data collection is incomplete. As part of the pilot study, GDSP identified and recruited primary care physicians and enlisted them to collect clinical and utilization data outside of SIS, using instead a web-based REDCap data collection system that was developed in collaboration with the NBSTRN. During the pilot study, two-years of encounter-based treatment, drug dosage and health outcome data for 24 newborns and children with PCH were collected by 17 primary care providers. Physicians were paid \$200 for each PCH patient the pediatrician consented through the child's guardian. The pilot study was successful for this small group of pediatricians: they found data entry to be facile and some would be willing to do data entry without payment.

During the pilot, GDSP staff finalized primary care provider treatment guidelines for dissemination to pediatricians who are not experts, but who occasionally manage children with PCH (Appendix 6). The pilot also stimulated a successful endocrine CME course developed by Stanford University entitled, [*Congenital Hypothyroidism: What Every Primary Care Provider Needs to Know*](#)¹².

Lessons learned: (1) This approach using primary care providers, may be appropriate for states with smaller populations than California as a method to collect long term follow-up data for children with PCH and perhaps, other newborn disorders; (2) the model requires a minimal infrastructure investment, project management, recruitment staffing and part-time epidemiological consulting resources that might be already available within state health departments; (3) the model relies on identification of and payment to the data holders (primary care providers); (4) the software is agile and the database itself is maintained on the cloud. This use case also illustrates the challenges of acquiring more complete follow up data.

H. Organic Acidemia Association Collects Data Directly from Families

Several disease advocacy organizations have begun collecting data from member families that can provide key consumer insights into the natural history of the disease and availability of

services and support. Part of the Organic Acidemia Association's (OAA) mission is to empower families and health care professionals with knowledge in organic acidemia metabolic disorders. In an effort to increase and support research toward improved knowledge, treatment and eventual cures in the areas of Organic Acid Disorders, the OAA was one of twenty rare disease patient organizations awarded a grant from the National Organization of Rare Disorders to develop a Natural History Patient Registry in 2016. With input from the National Institutes of Health and the FDA, a tool was developed to collect data directly from the families and patients to include data such as diet, exercise, environmental factors and other variables that may affect disease progression.

While this is a self-selected population that may not be a representative sample, the high motivation of families to participate may be a useful strategy to overcome funding limitations and time constraints that limit such activities in health departments and provider settings. The goal is to assist medical researchers in gaining a better understanding of how these organic acidemias develop and progress over time so that more promising therapies can be developed. Data collected can also reveal any variations or gaps in treatment and coverage from state to state that could be quantified.

Lessons Learned: (1) Patients and families are the only source for accurate input of certain kinds of data and outcomes. Limitations of this approach are (2) ascertainment bias (study participants are self-selected and may not be representative of the full spectrum of the disorder) and (3) patients and families will not always be aware of or agree with "data dictionaries" for elements of reporting.

2. All Conditions Identified by NBS

A. The National Coordinating Center for the Regional Genetics Networks

The mission of the National Coordinating Center for the Regional Genetics Networks, a cooperative agreement between HRSA and the American College of Medical Genetics and Genomics (ACMG), is to increase access to high-quality genetic services to medically underserved individuals and their families. The NCC and the seven Regional Genetics Networks provide training, resources, and technical assistance to genetics and non-genetic providers on telemedicine. One resource, the NCC Telegenetics Workgroup, will work to develop, in collaboration with the American Telemedicine Association, are telegenic guidelines. The guidelines will consult published data, literature, expert consensus, and expert opinion to address areas such technical standards, administrative standards, business processes, clinical

delivery standards, quality metrics, operational considerations for forms of virtual services, and types of encounters. Though the establishment of these telegenetic guidelines, NCC aims to establish quality measures that can be used to assess telegenetics.

Lessons Learned: (1) Regional collaboratives can improve access to high-quality treatment of NBS conditions; (2) Telemedicine can help overcome issues related to access to specialists and can be used for education of providers.

B. California Department of Health Long-Term Follow-up

The California Newborn Screening (NBS) Program is administered by the Genetic Disease Screening Program (GDSP) of the California Department of Public Health. The GDSP has been collecting long term follow-up data from state-contracted specialty care centers for newborns since 2005. A legislated mandatory screening fee is paid by the birth hospital into the Genetic Disease Testing Fund. The Fund pays for screening and confirmatory testing of genetic diseases targeted by the NBS program, including short- and long-term follow-up of newborns diagnosed with a screened disorder. State legislation has recently expanded screening and follow-up for all diseases adopted by the federal Recommended Uniform Screening Panel. The Fund supports the infrastructure for data collection and analysis, including the *California Newborn Screening Follow-Up of Cystic Fibrosis* case study (see above for details). As part of the routine long-term follow-up data collection system, California requests an “Annual Patient Summary” up to age 5 years for children with metabolic and endocrine disorders and hemoglobinopathies. Severe Combined Immune Deficiency patients are followed for 2 years, and children with adrenoleukodystrophy can be followed through age 21.

Lessons Learned: (1) With appropriate resources and infrastructure, State NBS programs can provide LTFU of specific conditions.

C. University of Maryland Study of LTFU of Newborn Screening in Primary Care

A study at the University of Maryland explored long-term follow-up for newborn screening in the primary care setting. Three primary care practices were recruited and the focus was on sickle cell disease and hearing loss since the numbers are larger than other conditions and the health department follows these conditions beyond newborn screening. Use of the primary care setting allowed inclusion of children not identified by newborn screening. All 8 cases of sickle cell disease were identified by newborn screening and 4 of the 6 hearing loss cases were identified by newborn screening. NCQA levels were used to measure Patient Centered Medical Home status including organizational capacity for children with special health care needs and

their families. Other measures included use of individual care plans, clinical outcomes, and electronic data sharing. The study confirmed that children identified by newborn screening are successful in getting integrated into care but that primary care providers have incomplete information regarding long-term follow-up care.

Lessons Learned: (1) It is feasible to carry out quality measurement in the primary care setting; (2) important future targets should focus on communication of information; (3) practices can assess their medical home capabilities which is an important component of newborn screening follow-up. and (4) data collection was labor-intensive and costly, requiring a two-year grant to study three practices and 14 patients.

D. Newborn Screening Translational Research Network (NBSTRN)

The American College of Medical Genetics and Genomics coordinates the Newborn Screening Translational Research Network (NBSTRN), a part of the National Institute of Health's *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD) Hunter Kelly Newborn Screening (NBS) Research Center. In accordance with the Newborn Screening Saves Lives Act of 2014 (P.L. 113-240), the Hunter Kelly NBS Research Center develops systematic methods to identify additional conditions appropriate for NBS; develops interventions and treatments to improve outcomes; and sponsors pilots of conditions recently recommended for nationwide screening. Although focused on research, some tools developed by NBSTRN could be developed and used as quality measures.

Over the past decade, advances in technology and improved understanding of genetic disease provided new opportunities to improve the scope and quality of NBS services.¹³

¹⁴NBSTRN is developing a suite of tools to help researchers translate these research and technology discoveries to the state-based NBS programs. One of the tools NBSTRN has developed, the Longitudinal Pediatric Data Resource (LPDR), enables collection of health information across the lifespan for individuals with NBS conditions. Data collection, aggregation and sharing using the LPDR is critical because the majority of the conditions that are part of, or candidates for, newborn screening in the United States are rare. The LPDR supports basic researchers working to understand the disease process, translational research programs developing technologies to screen and therapies to treat, and NBS programs implementing screening for new conditions. As of 2016, longitudinal data from 7412 participants with one of 46 NBS conditions have been deposited in the LPDR by six research projects.¹⁵ The accumulated data is available to qualified researchers for secondary analysis, including data

mining for quality improvement (QI) efforts. The LPDR includes a set of public health data fields (Appendix 7) that can be used for any NBS condition.

The creation of tools, like the LPDR, to facilitate data sharing, aggregation and analysis that are critical to advancing NBS. Researchers, public health team members, and clinicians report that the use of standardized vocabularies, interactive computer systems, and robust security measures have been key components in the success of the LPDR. Identifying and supporting incentives to sustain robust data collection across rare diseases is a challenge, but will ensure the continued creation of this valuable resource and ultimately improved health outcomes.

Lessons Learned: (1) Providers will participate without financial incentive. (2) Data aggregation and sharing is critical for rare diseases; standardized vocabularies are helpful. Data security is important.

E. NewSTEPs

NewSTEPs, a HRSA funded cooperative agreement through the Association of Public Health Laboratories, has partnered with newborn screening stakeholders to develop several foundational resources for quality measures. Case definitions for public health surveillance of newborn screening conditions developed will help to consistently define affected individuals with a given condition across the nation. NewSTEPs has developed a national reporting infrastructure for state newborn screening programs that has been applied to short term follow-up and could be extended to handle foundational measures from long term follow up in the public health context. The newborn screening process is inherently a state-level activity, but complete long-term follow-up requires tracking patients over time as they move to different states from the one in which they were originally screened. There may be a role for NewSTEPs to facilitate connecting patients who have moved to another state to their original NBS program with appropriate consent.

Lessons Learned: Data that is locally identifiable but de-identified and combined at a national level can improve access to data at multiple levels without redundant mechanisms. Such a national database could aid in LTFU as patients move state to state

F. NBS Connect: A Web - Based Self - Report Registry for Patients with Disorders Identified by Newborn Screening

As part of Emory University's comprehensive clinical and research program on metabolic genetics and nutrition, we have developed the web-based patient registry, NBS Connect (www.nbsconnect.org).¹ The purpose of NBS Connect is to collect and analyze primary data on

diagnosis, treatment, symptoms, outcomes, and barriers to care in inherited metabolic disorders (IMDs) included in newborn screening (NBS) programs. NBS Connect also provides professional support resources for patients and families. Features of NBS Connect include educational materials, low protein recipes analyzed by registered dietitians, interactive health tracking tools for data visualization, information about the latest research and clinical trials, opportunities to connect with experts, and a forum for patients and their parents to connect with each other. NBS Connect was built and refined with feedback from all stakeholders, including individuals with IMDs.

Many of the IMDs identified by NBS are rare and therefore do not have the infrastructure of support groups or independent registries to connect patients with each other, clinicians, and researchers. NBS Connect aims to fill this tremendous gap, and to provide a reliable source of publically available data to enhance the knowledge base and improve research.

Patients with any IMD are welcome to register and complete a profile survey, including demographic questions such as family history, development and social history, insurance, and research interests. Disease-specific surveys, including questions on diagnosis, genetic testing, clinical symptoms, treatment, and diet management, are currently available for phenylketonuria (PKU), tyrosinemia (TYR) and maple syrup urine disease (MSUD), with a goal of expanding to include all of the IMDs in the American College of Medical Genetics and Genomics recommended uniform screening panel. As of October 2017, 541 people have registered with NBS Connect and 284 registrants with PKU, MSUD, or TYR have completed a disease-specific survey. We are actively working with advocacy organizations to increase the number of registrants.

Participants are encouraged to update their profile once per year, with de-identified patient data offering participants, clinicians and researchers a temporal view of patient health over time. This valuable tool allows patients and families to see how they compare to other patients in the registry, and for clinicians to obtain information on patient experience, including management techniques, clinical signs and symptoms, and barriers to care. In addition, the data visualization feature allows registry users to see aggregate data in charts and graphs for each of the survey questions. Researchers can export the data in MS Excel, CSV, or HTML formats for more detailed analysis.

The use of NBS Connect registry data has been described in two recent reports. First, we published an overview of 217 participants with PKU and provided examples of the data visualization capacity of NBS Connect.¹⁶ Second, data from 39 patients with MSUD were analyzed to describe management techniques, clinical signs and symptoms, factors potentially

associated with metabolic control of plasma leucine levels, and the impact of NBS on outcomes.¹⁷

Lessons Learned: (1) The results presented in these two papers highlight the value of patient self-report registry data. (2) Patients are willing to register and use a website for data entry and a single website can serve multiple conditions identified by newborn screening.

3. Children with Special Health Care Needs (CSHCN)

A. National Survey of Children's Health (NSCH)

The NSCH is sponsored by the U.S. Department of Health Services, Health Resources and Services Administration, Maternal and Child Health Bureau and conducted by the U.S. Census Bureau to provide national and state level estimates of key measures of child health and well-being. Prior to 2016, two surveys were conducted, the National Survey of Children's Health (NSCH) designed to produce estimates of the health and well-being of children overall and a second, the National Survey of Children with Special Health Care Needs (NS-CSHCN) designed to produce estimates of the prevalence and impact of special health care needs. In 2016, these two surveys were merged, though data can be analyzed separately for CSHCN. Information is collected on factors related to the health and well-being of children, including access to and utilization of health care, receipt of care in a medical home, insurance status, type and adequacy, health care transition planning family interactions, parental health, school and after-school experiences, and neighborhood characteristics. In 2012-2015, the surveys underwent a significant redesign, shifting from a telephone- to an address-based sampling frame and changing mode of administration from an interviewer-assisted survey to a self-administered web or paper-based survey. The 2016 NSCH utilized a sample of 364,153 household addresses drawn from the Census Master Address File resulting in completed topical questionnaires for 50,212 children and an overall weighted response rate of 40.7%. A screener questionnaire was sent to determine if children resided in the household. Follow-up surveys were targeted by age of children in the household; parents and caregivers were the respondents. Households with children with special health needs were oversampled. Micro-data files are now available on the survey website.

Many of the survey questions align with the LFTU goals and questions defined in the publications of the FUTR subcommittee. This annual survey may have potential to be analyzed for a subset of children identified through newborn screening if appropriate questions are added and the use of multiple years of data was deemed acceptable in order to ensure adequate

samples for rare conditions or sub-populations. NSCH data are not longitudinal, rather these data are cross-sectional, even with an annual administration. Focusing attention on these questions and gaps in services and support may be useful to increase provider attention to many of these issues. However, the limitation is that the sampled population may be too small to collect enough data to analyze or indicate any trends.

Lessons Learned: (1) National surveys may be an important tool to gather data on the entire population and not just patients served by a specific provider or health department. (2) There may be an important opportunity to modify the survey to generate data specifically on newborn screening.

B. Lessons for NBS from Quality Measures Developed for Other Childhood Chronic Diseases

These are described in Appendix 8.

4. All Children

A. HEDIS

The National Committee for Quality Assurance (NCQA) is a private organization devoted to measuring quality and improving health care. They are best known for their HEDIS quality measurement program and for their Physician Recognition program for Patient Centered Medical Home (PCMH).

Annual sets of HEDIS measures are used by Medicare, Medicaid, and Private Insurance companies to score practices on quality using data capture and reporting tools that may involve sampling of records. They can be applied to paper as well as electronic records and may involve no charge billing codes for quality assurance tasks such as asking about smoking. Most measures are oriented toward adult chronic diseases, but there are some child-oriented measures such as appropriate strep testing and follow-up of ADHD medications. Some pediatricians and family medicine practitioners have raised concerns that the PCMH certification is also too adult specific and not be oriented towards the needs of children with special healthcare needs.

Lessons Learned: (1) The requirements of insurance programs are strong motivators for participation in quality measurement programs, and appropriate data collection and reporting tools are in place to make these audits feasible; (2) available measures focus on common problems such as adult chronic diseases or throat infections in children; measures that are more

relevant to children with NBS conditions are not included (e.g. transition to adult systems of care); (3) children identified by newborn screening cannot easily be studied using this approach; (4) NCQA tools to assess medical home status may be helpful, but credibility of this approach for newborn screening depends on considering the needs of children with special healthcare needs.

B. Pediatric Quality Measures Program

The PQMP was initially established in 2011 under the Children's Health Insurance Program Reauthorization Act (CHIPRA) with the aim of increasing the portfolio of evidence-based, consensus pediatric quality measures. These are used by state Medicaid and Children's Health Insurance Programs (CHIP) and other public and private programs, providers, plans, patients, and their families to measure and improve the quality of children's health care. The initial phase of the PQMP funded seven Centers of Excellence (COEs) to develop new and innovative pediatric measures.

In October 2016, through the Medicare Access and CHIP Reauthorization Act (MACRA), AHRQ and CMS awarded funding to six grantees to continue support for the PQMP. This current phase of work focuses on implementing and testing the newly developed pediatric measures in real-world settings, as well as the development of quality improvement projects working with a broad group of stakeholders which includes states. The projects will allow for learning on how measures are used, and also barriers and challenges related to measurement that is encountered at multiple levels of care. The PQMP measures span diverse areas such as perinatal care, child clinical preventive services, and management of chronic and acute conditions, including measures focused on screening, follow-up, and sickle cell anemia, which may be useful for better understanding the potential role of quality measures to promote long-term follow-up of NBS.

Lessons learned. (1) Several of the PQMP measures and also knowledge gained from their uptake and implementation can be leveraged to develop long-term follow up measures of NBS. (2) Emphasis should not be placed solely on developing measures that are scientifically sound (reliable/valid) but measures must also be useable and feasible—this is vital to actually improving quality of care processes and outcomes. Having stakeholder input from those implementing the measures and also from families and others key users of the data/information that comes from measures is key to ensuring measure usability and feasibility.

C. Health Resources and Services Administration, Maternal and Child Health Bureau (MCHB) - Title V Program

The Title V Maternal and Child Health Block Grant Program is the nation's oldest federal-state partnership. It is funded with a block grant appropriation from Congress and with a 4 to 3 dollar federal/state match of funds. Fifty-nine states and jurisdictions receive Title V funding. The Title V program aims to improve the health and well-being of women (particularly mothers) and children and in 2014, Title V programs reached over 50 million pregnant women, infants, and children.

The transformation of Title V included a new performance measure framework to demonstrate how Title V programs improve health outcomes. Each measure is linked to a national data source. The program uses:

- National Outcome Measures (NOMs) – intended to represent the desired result of Title V program activities and interventions. These measures for improved health are longer-term than National Performance Measures (NPM).
- National Performance Measures – intended to drive improved outcomes relative to one or more indicators of health status (i.e., NOMs) for the maternal and child health population.
- Evidence-based Strategy Measures (ESMs) – intended to hold states accountable for improving quality and performance related to the NPMs and related public health issues. ESMs will assist state efforts to more directly measure the impact of specific strategies on the NPMs.

The data from states is available on the HRSA Title V Information System which is an on-line system which allows users to look up state by state measures, data, and progress towards improvement of health outcomes. The sharing of data allows states to learn best practices from other states and find partners to work collaboratively on activities.

Lessons learned. (1) This type of long term and continuous measurement of outcomes can be used as a model for newborn screening quality improvement activities.

E. National Patient-Centered Clinical Research Network (PCORnet)

The adoption of the Nuremburg Code, which required research subjects to give informed consent, seventy years ago ushered in a new age of patient participation in research.¹⁸ Organizations like the March of Dimes empowered patients to join together to advocate for research that addressed patients' needs. The Patient-Centered Research Institute (PCORI) is

one of the key forces advocating for a change in the culture of research from being researcher-driven to becoming patient-driven, and promotes studying questions that matter most to patients.¹⁹ PCORI encourages engaging patients and other stakeholders throughout the research process and facilitates the interaction of PCORI efforts through the National Patient-Centered Clinical Research Network (PCORnet). The goal of PCORnet is increased collaboration, efficiency, and people-centeredness in clinical research. PCORnet is designed to enable patients, their families and clinicians to make informed healthcare decisions by supporting an innovative approach to clinical research.

PCORnet is made up of 13 Clinical Data Research Networks (CDRN), 20 Patient-Powered Research Networks (PPRN) and 2 Health Plan Research Networks (HPRN). The PCORnet Research Networks use a distributed research system to securely collect and store data within their own institution. Data collected across the networks is then aggregated and made available to qualified researchers. Through a shared infrastructure that supports clinical research, PCORnet is designed to significantly reduce the time and effort required to conduct important studies. The generation of data that empowers individual patients and clinicians to make informed choices for care based on their individual circumstances may provide an assessment of QI across a variety of clinical settings and conditions.²⁰

Lessons learned. (1) Local and regional data collection can be combined across research networks to improve research efficiency and ease

F. National Survey of Children's Health (NSCH)

The NSCH conducted by HRSA is described above in section 3A collects important data on all children that provides an essential comparison group to evaluate how children with special healthcare (CSHCN) needs are doing. If we are able to identify a cohort of CSHCN identified through NBS among the survey participants, we can also gain insights into the potential unmet needs and health status of this population.

Use Case Summary Tables

Table 1. Summary of programs that employ quality measures for specific conditions

Condition	Program	Purpose	Participants	Funding source	Level of care	Barriers	Lessons learned
Sickle cell disease	Sickle Cell	Promote use of	Collaborative	Federal	Primary and	Challenges with	Evidence based
	Disease	quality measures in	teams	competitive grant	specialty	data collection	therapies are not
	Treatment	sickle cell care	proposing	(HRSA)	clinics	across institutions	universally applied. QI
	Demonstration Program	across practice settings	specific quality measures			limited scope of influence	programs can increase use of proven therapies. Collaboration across practice settings (primary, specialty, ED) can improve care. Funding supports QI goals
	AHRQ IMPLEMENT	Assess feasibility of implementing quality measures to be used by PQMP	Sites in the Pacific Sickle Cell Regional Collaborative applying PQMP measures	Federal grant (AHRQ)	Multiple	Ongoing project	
	CDC RuSH	Surveillance of Hemoglobinopathies	7 states now reduced to 2 states	Federal grant (CDC) transitioned to private foundation	Multiple	Sustainable funding	

Condition	Program	Purpose	Participants	Funding source	Level of care	Barriers	Lessons learned
Cystic fibrosis	Cystic Fibrosis Foundation care center accreditation	Promote transparency, accountability and quality improvement among centers through data sharing	Individual centers contribute to and can access a centralized registry	Foundation	Foundation accredited specialty care centers	Local IRB oversight is required	Foundation support can catalyze widespread data collection and sharing and implementation of quality measures. The incentive of accredited center status is a sufficient motivator.
	California genetic disease screening program	Improve care and follow up of patients with CF identified through newborn screening	Centers contracted with the state health department provide data annually for patients through 5 years of age	State funds collected from birth hospitals given to contracted sites	Foundation accredited specialty care centers	5 year time frame is too short for some conditions. California's model for funding newborn screening efforts is not universal.	Data sharing through a centrally maintained database allows large scale knowledge discovery and feedback to individual sites for quality improvement. State and foundation collaboration improves access to centers of excellence

Condition	Program	Purpose	Participants	Funding source	Level of care	Barriers	Lessons learned
MCADD	Mountain States Genetic Collaborative	Standardize information sharing and emergency procedures for patients with MCADD	Providers in a single center (CHCO)	Provider initiated/unfunded	Chart review of standard of care safety measures. Standardized chart popup	Technical support for popups is not standard across platforms so interventions are not generalizable to different systems	Quality activities can be integrated into routine care, chart tags can quickly communicate patient or disease specific information. Such activities can be applied to multiple disorders. MCADD is a proof of principle for EMR use
Hearing loss	CDC, NQF	Implement a data collection system with in states to provide accurate counts of babies born and screened (NANI)	CDC provided data collection tool to states	Federally initiated, state adopted. Federal/state funding	Data collection system in birth hospitals uploads data to state NBS programs for real time analysis	Complete documentation of patients receiving follow up after nursery discharge	Identification of standardized validated measures is the first step in widespread adoption of such measures. Centralized data collection allows for clearer understanding of the scope of a given problem related to quality measures applied to patient care. Ascertainment bias is a challenge in QI data collection.

Condition	Program	Purpose	Participants	Funding source	Level of care	Barriers	Lessons learned
Primary congenital hypothyroidism	California genetic disease screening program	Assess health outcome data of patients through medical home	Primary care providers contracted with the state health department	State funds collected from birth hospitals given to contracted physicians	Data collection system uploaded to state	If care is provided at a noncontract center, it is not tracked by the program; payment of participants can be expensive	Upon program implementation, guidelines and education were provided which can benefit everyone, not just the participating providers. Minimal infrastructure investment and staff in state health department allows robust data collection when primary care physicians participate in the data entry.
Organic acidemia	Organic Acidemia Association	Develop natural history registry Improve understanding of natural history and practice variations	Families contacted by association	Association initiated with grant from NORD	Data voluntarily provided by families via questionnaire	Self-selected population of respondents; family perspective somewhat anecdotal (did not involve standardized data or provider input)	Data can be collected directly from families; consider ascertainment bias

AHQR - Agency for Healthcare Research and Quality; IMPLEMENT - IMPLementing MEasures NeTwork for Child Health (IMPLEMENT for Child Health); PQMP - Pediatric Quality Measures Program; HRSA -Health Research and Services Administration; NQF -National Quality Forum; NANI - Newborn Admission Notification Information; CHCO, Children's Hospital of Colorado; CDC - Centers for Disease Control and Prevention; NBS - Newborn Screening

Table 2. Case studies illustrating the use of quality measures in pediatric populations beyond those with specific newborn screening disorders.

Program	Purpose	Lessons learned
All conditions identified by newborn screening		
National Coordinating Center for Regional Genetics Networks	Provide training, resources, and technical assistance to genetics and non-genetic providers on telemedicine	Regional collaboratives can help improve quality of care for NBS conditions. Telemedicine can overcome issues related to access to specialists and can be used for education of providers
California Department of Public Health	Provide long term follow-up data for newborn screening through funds collected as part of a legislated fee to birth hospitals for specific pilot projects	With appropriate resources, State NBS programs can provide LTFU of specific conditions
University of Maryland	Study long-term follow up of newborn screening conditions in primary care settings using medical home measurements	Patients with NBS conditions are getting identified and into medical homes, but there are gaps information transferred to PCPs by specialists. Data collection was labor-intensive and expensive
Newborn screening translational research network	Develop tools to help researchers translate research and technology discoveries to state-based NBS programs. i.e. the Longitudinal Pediatric Data Resource (LPDR) for data collection across the lifespan	Providers will participate without financial incentive. Data aggregation and sharing is critical for rare diseases; standardized vocabularies are helpful. Data security is important
NewSTEPS	Provide a national resource for data collection, technical assistance and training to newborn screening programs and to assist states with quality improvement initiatives	Data that is locally identifiable but de-identified and combined at a national level can improve access to data at multiple levels without redundant mechanisms. Such a national database could aid in LTFU as patients move state to state
NBS Connect		

Program	Purpose	Lessons learned
All children with special healthcare needs		
National survey of children's health	Provide national and state level estimates of key measures of child health and well-being	Specific questions could be added and analyzed for children with NBS conditions. Attention to care coordination and availability in services through longitudinal data collection can benefit all children.
All children		
Healthcare Effectiveness Data and Information Set (HEDIS) from NCQA	Measures used by payers to score practices on quality	Incentives to increase reimbursement are strong motivators for quality improvement. Measures specific to NBS conditions need to be developed
Pediatric quality measures program	Develop quality measures at designated and funded sites	Piloting measures at several designated sites allows development of reliable measures for broader implementation
Maternal and Child Health Bureau	Allocate programmatic funding to states that participate in long-term reporting of outcome measures	This type of long term and continuous measurement of outcomes can be used as a model for newborn screening quality improvement activities

V. Key Findings

As demonstrated in the examples above, quality measures applied to specific disease conditions can be successfully implemented at a variety of levels including nationally through federal, foundation and payer guided efforts as well as at the state, institution and individual clinic/provider levels. Quality improvement applied to rare diseases benefits from large centrally maintained databases when they are designed to accept and retrieve data for interested parties to use, although security and funding are areas of concern. Collaboration between specialists, primary care providers and emergency department and between institutions can lead to robust data collection and quality measure implementation. Incentives to participate in such efforts including recognition as a center of excellence, improved reimbursement or direct financial benefit to individual providers can motivate participation. Quality activities can be incorporated into routine care and the power of the EMR can be harnessed to facilitate these activities although standardization across platforms is needed.

Despite these many examples, overall there are relatively few disease-specific quality measures for NBS conditions, and there is no national standard for the use of quality measures in LTFU by state NBS programs. Additional gaps and barriers in applying quality measures to newborn screening include gaps in evidence, gaps in developing measures, gaps in adopting and using measures, and gaps in funding.

Gaps in Evidence

One evidence gap that continues to be a concern is that many conditions detected by newborn screening are individually rare, may have sub-types, and variable age of onset so that the best treatment is not always clear. There are many non-condition specific measures that can be applied to any newborn screening condition. The Cystic Fibrosis Foundation has demonstrated that quality measures can actually serve as a pathway to gather evidence and fill gaps.

Gaps in Developing Measures

Developing measures is a challenge for rare disorders. In fact, developing measures for children was a challenge; the lack of pediatric quality measures led to the CMS/AHRQ PQMP, mandated by the CHIPRA legislation. These pediatric measures, in development since 2011 include several relevant to newborn screening, including those focused on sickle cell anemia and also measures focused on long-term follow up in other conditions. Development of measures can be costly as testing of measures to ensure appropriate reliability and validity can be time and resource intensive.

Gaps in Adopting and Using Measures

The cost of data collection and small numbers of patients with specific conditions in a single practice are challenges. Measures for sickle cell disease are now available and expected to increase in use. Some

models used by health departments are hard to replicate elsewhere because of the need for funding or lack of a mandate to engage in long-term follow-up, including data collection, after newborn screening. Moving forward, efforts focused on integrating quality measures into routine care so that it is an ongoing activity may be an important consideration as it reduces burden for providers and systems of care, but can provide valuable real-time data for action.

Gaps in Funding

Gaps in funding have been important barriers to the use of quality measures for LTFU of NBS. Sustainability of data collection depends on continuation of funding which is why a shift to limited data collection integrated into routine care may be an important strategy to supplement funded research activities. Development and validation of measures is a time limited activity that expands when funding is available. The proven value of networks to capture and share data as well as best practices for LTFU justifies sustainable funding or the ability to leverage existing resources for this purpose.

There is a Need to Move Beyond “Disease Specific Measures”

Disease specific measures is an ambiguous term that has two different, but related meanings. It is used to refer to measures used for a single condition such as those that apply only to sickle cell disease. It is also used to refer to physiologic or other proxy outcomes such as laboratory results that are easier to measure and assumed to be correlated with true patient specific outcomes such as mortality and morbidity. Traditional approaches to quality measurement may fall short for newborn screening. We need to focus on the development and use of measures that can be used within a Public Health or Newborn Screening System to evaluate long term follow up including access to and receipt of services, and transition to adult care. We also need child-specific measures that focus more broadly on access to medical homes, child well-being, and family satisfaction with the care process. To meet these needs, data sources may need to move beyond the clinical context.

Patient/Family Perspective on Quality

Patients and families have their own definition of quality. They have an important role and are vital for identifying needs and gaps that providers and the system may be missing, including perspectives on care, interest in and opportunity ability to participate in research studies, access to specialists, and insurance coverage for their conditions. Patient and family experiences are an important data sources and can serve to complement other measures of quality. Several advocacy organizations have successfully collected important disease specific data collected directly from patients and families using surveys and patient natural history registries which can serve as exemplars.

Available Resources and Standards Could Accelerate the Use of Quality Measures for Newborn Screening

Quality measurement is cost and resource intensive, but promising new tools may make it easier in the future and reduce these barriers. The Office of the National Coordinator for Health Information Technology (ONC) works with CMS, and AHRQ to maintain an Electronic Clinical Quality Improvement Resource Center (<http://ECQI.HealthIt.gov>) The website provides access to Health IT standards for quality measure definition and reporting and access to available quality measures and incentive programs. The ACHDNC has worked with the ONC and the National Library of Medicine to develop a Newborn Screening Use Case, standard coding for newborn screening conditions and tests, and to encourage development of certified eCQM. A few eCQM have been developed and validated for EHDI, sickle cell disease, and Cystic Fibrosis. The adoption of eCQM has been slow and the integration of quality assurance and long-term follow-up data elements related to newborn screening conditions into EHR has been limited. Fast Health Interoperability Format (FHIR) is facilitating development of interfaces with EHR.

Communication of information between specialists, primary care, and public health remains an important challenge despite available HIT standards that could be used for this purpose. The APHL NewSTEPs program has developed case definitions and a national reporting repository that can help define the denominator of affected infants for NBS quality measures. The NBSTRN's LPDR is available as a REDCap database with definitions of data fields and including core, disease specific, and public health variables (Appendix 7).

The Approaches to Clinical Quality Measures for Newborn Screening are Very Diverse

Data sources and specifications for quality measures for newborn screening vary greatly. Measures are developed and intended for use across many sectors or levels of care including specialty or primary care providers, health departments, or directly collected from consumers including patients and families. Some measures are directed at the process of care, some at outcomes, and some at the care experience and there are varying specifications for measures making it difficult to compare performance.

VI. Next Steps

The field of quality measurement and improvement is wide-ranging and used for a variety of purposes. Efforts across many sectors and levels of care delivery are detailed in case studies. These can provide important lessons learned to guide next steps in considering the use of quality measurement and improvement to enhance long-term follow of newborn screening. The Follow Up and Treatment Workgroup's report to the ACHDNC provides a synthesis of the available information on the development and use of quality measures as a means to improve long-term follow-up and outcomes for children diagnosed through public health newborn screening.

The report outlines for the ACHDNC potential targets for new knowledge discovery, and highlights possible deficiencies in the current processes of care that could be addressed by quality measurement and improvement efforts as well as existing work that can be leveraged.

Given that many newborn screening conditions are rare and funding for measure development is limited, it is unlikely that formal NQF-endorsed quality measures will be the primary focus of ongoing public or private efforts. Nonetheless, quality measures can play an important role in improving care processes, family experience, and health outcomes, and, therefore, progress can be made in the following areas:

1. Make the case for the importance of prioritizing development and use of quality measures at multiple levels and systems of care for LTFU of NBS as a strategy for engaging a broad range of stakeholders including Federal, State, Provider, and Consumer groups to participate in LTFU of NBS.
2. Identify a core set of long term follow-up quality measures and associated data resources for conditions identified by newborn screening that will maximize existing collaborative efforts by groups such as APHL/NewSTEPs, NBSTRN/LPDR), and NCC to gather uniform LTFU data from more states and other organizations.
3. Encourage the use of large data collection activities such as the National Survey of Children's Health (NSCH) and quality improvement activities such as Medicaid quality reporting and HEDIS to provide data on LTFU of NBS by identifying cohorts of children with disorders identified by NBS.
4. Work with key stakeholders, such as consumer advocates and professional associations to leverage research networks that collect data from patients and families to participate in quality measure development and quality improvement activities targeted to LTFU of NBS.
5. Assist the use of new Health Information Technology (HIT) standards for implementing and sharing quality measures as a strategy for integrating quality measures into routine care and using Clinical Decision Support (CDS) in the EHR to capture data and guide care.

We recognize that the availability of resources will promote (or limit) the pace of taking each of these next steps to disseminate the lessons we have learned.

VII. Conclusion

The growing interest in the use of quality measures for improving care makes this an opportune time for the ACHDNC to identify a role for itself in encouraging stakeholders, identifying targets, and facilitating the use of quality reporting, quality improvement, and clinical decision support to improve long-term follow-up of conditions identified through newborn screening.

Appendices

Appendix 1: Health Information Technology^{xxi,xxii}

The SACHDNC participated in the Decade of Health Information Technology that began in 2004 with the creation of the Office of the National Coordinator for Health IT and the publication of a Framework for Strategic Action called *The Decade of Health Information Technology* that set a goal of providing electronic medical records for most Americans by 2014. The goal was achieved through the ARRA HITECH Act that created the Meaningful Use EHR Incentive Program and drove adoption, but also created disappointment regarding ease of use and achieved value of the technology.

We are now in the second Decade of HIT that began in 2015 with the publication of *Connecting Health and Care for the Nation: A Shared Nationwide Interoperability Roadmap* with a goal to achieve nationwide interoperability to enable a learning health system by 2024. Quality measures will be a key component of the learning health system that will use data captured in the EHR to monitor outcomes of care, leading to new knowledge discovery and appropriate guidelines for care, that are integrated into the EHR through clinical decision support that will change what is done during the clinical encounter. The EHR of the future will no longer focus on documenting the encounter for audits and future care of the same patient, but will be expected to change what takes place during an encounter and influence the care of other patients in the future.

The SACHDNC did participate in, monitor, endorse or encourage several activities to promote the application of Health Information Technology in Newborn Screening including:

- Development of LOINC codes for newborn screening laboratory tests and SNOMED CT codes for newborn screening conditions
- Development of a Newborn Screening Use Case for interoperability
- Development of implementation guides for reporting newborn screening results using the HL7 electronic laboratory results reporting standards specific for Meaningful Use
- Development of National Quality Foundation (NQF) certified quality reporting measures as required for Meaningful Use as well as other CMS/AHRQ Child Health Quality Measures relevant to newborn screening
- Development standard fields and datasets for research databases at NBSTRN LPDR should be spelled out here or previously
- Development of standard case definitions and case reporting tools at NewSTEPS

All of these activities lay the foundation for the current work to promote the use of quality measures as a tool for long-term follow-up of newborn screening.

HIT Standards for Defining and Reporting Quality Measures

New HIT standards are playing an important role in integrating quality measures into routine care, reducing the incremental cost of data collection, and providing a mechanism for replicating activities between organizations in a standard way with limited cost. The Office of the National Coordinator for Health IT (ONC) is playing a central role in disseminating and demonstrating these new technologies.

The best central reference for the current state of standards for Electronic Clinical Quality Measures is the eCQI Resource Center maintained by the Office of the National Coordinator for Health IT (ONC) at <http://ecqi.healthit.gov>. This website maintains current links to relevant documents maintained by various organizations and links to approved measures. Standards and available measures change regularly and the website clarifies current versions and relevant history.

QDM Quality Data Model: The Quality Data Model (QDM) is a solution to the “curly braces” problem (where to find data in a specific EHR) of the older Arden Syntax for clinical decision support. The QDM defines a clear general-purpose data model for storing information in any EHR and extracting it for use in quality assessment.

CQL Clinical Quality Language: Clinical Quality Language (CQL) is an HL7 standard that is part of the effort to harmonize standards between electronic clinical quality measures (eCQMs) and clinical decision support (CDS). CQL provides the ability to express logic that is human readable yet structured enough for processing a query electronically. In the future, CQL is to be used in all of the clinical quality measure HQMF electronic specifications.

HQMF Health Quality Measures Format: The NQF Health Quality Measure Format (HQMF) is a format for specifying the detailed definition and implementation instructions for a eCQM. Because it is implemented in XML, the document is both human readable and machine readable.

QRDA Quality Reporting Data Architecture: The HL7 Quality Reporting Data Architecture (QRDA) is an XML clinical document architecture (CDA) document for reporting the results of quality assessment to a quality assurance agency.

FHIR Fast Health Interoperability Resources: The HL7 Fast Health Interoperability Resources (FHIR) is an Application Programming Interface (API) approach to adding applications to EHRs from multiple vendors that exposes the data required for tasks such as quality assessment or clinical decision support. It is an important strategy for the ten-year Nationwide Interoperability Roadmap of the ONC and may play an important role in implementing eCQM with EHR produced by different vendors.

CDS Clinical Decision Support: HL7 also supports various standards for providing CDS to various EHRs by sending data to a standardized web interface and getting decision support returned to the EHR for display to the user. Data sent to the CDS server can usually be de-identified so that decision logic is shared between different health care organizations, but specific patient data is never shared.

CDS Hooks: Is a standard for triggering CDS in an EHR when standard events occur as defined in a CDS Hooks Card file. Triggering CDS specific to NBS conditions could be accomplished using CDS Hooks if

standard SNOMED CT coded diagnosis terms are included in the problem list of all infants identified by NBS and specific encounter types, ages, or events such as an ER visit, are defined when LTFU data collection should occur.

Notes on Using eCQM for NBS

All clinical quality measures must clearly define the population that they should be applied to that is typically represented in the denominator of the measure. These should include specific supporting evidence for a diagnosis as well as specific inclusion and exclusion criteria. The case definitions and documentation tools developed by APHL NewSTEPS provide an important resource to facilitate enrollment of patients in quality measures.

A clinical quality measures must clearly define the measurement parameters that form the numerator of the measure. The Longitudinal Pediatric Data Resource (LPDR) developed by ACMG NBSTRN can provide a framework for implementing eCQM in newborn screening and serve as a tool for testing new measures with existing datasets.

All clinical quality measures must have a custodial organization that will attest to the clinical validity of the measure and that will maintain and revise the measure on a regular basis. Several organizations such as the NQF, AHRQ/CMS, and professional societies such as the American College of Medical Genetics (ACMG) endorse specific measures that meet NQF or CMS selection criteria. A subset of endorsed measures may be selected for use in specific incentive programs such as the Medicare Incentive Reimbursement Program (MIPS).

The National Coordinating Center (NCC) and Regional Genetics Collaboratives have already begun to engage in quality measures and provide a framework to engaging a range of stakeholders. Use of standards for eCQM will help integrate quality measures and LTFU into routine care and reduce the cost of implementation.

Lessons Learned: (1) Most newborn screening conditions are rare and developing and validating eCQM is difficult and expensive. It is unlikely that more eCQM will be developed for conditions other than EHDI, sickle cell disease, and Cystic Fibrosis; (2) the promise of integrating portable Quality Assessment, QI, and Clinical Decision Support into any EHR has been demonstrated, but is not widely available. Doing this for newborn screening conditions is a low priority for most institutions; and (3) developing portable applications that end users could add to their EHR is a promising strategy even if the data collected might not be stored in the EHR and access to data in the EHR is limited.

Appendix 2: Previous ACHDNC FUTR Committee Work

These four key components are presented with key questions identified by the prior FUTR Committee that that data collection about long-term follow-up of NBS should be able to answer. Each key question should be

addressed from three perspectives: Families, Medical home/primary care provider/specialists/clinical investigators, and State/nation:

- Component 1: Care coordination through a medical home
 - Is my child receiving coordinated care through a medical home?
 - Are children/adolescents receiving coordinated care through a medical home?
 - Do children/adolescents receive coordinated care through a medical home?
- Component 2: Evidence-based treatment
 - How is my child doing clinically?
 - How are the children/adolescents doing clinically? Are children identified through NBS and enrolled in care doing better than those identified clinically?
 - How are the children/adolescents doing clinically?
- Component 3: Continuous Quality Improvement
 - Is my child getting the best care and treatment? How can I improve my child's outcome?
 - Am I doing the best for my patients?
 - How do we assure ongoing QI?
- Component 4: New knowledge discovery
 - Is my child able to enroll in clinical research related to his/her disorder?
 - Do children in a provider's practice have the opportunity to enroll in clinical research?
 - What clinical and observational long-term follow-up research efforts are being performed at the state and national levels?

The most recent paper included a framework and strategy for long-term follow-up of NBS:

- Rapid and reliable detection and diagnosis
 - Condition detected by NBS
 - Condition confirmed and diagnosed
- Provision of evidence-based therapeutic and habilitative care
 - Prevention of major disease-related mortality and morbidities
 - Growth and development
- Coordination and integration of services to address holistic spectrum of child and family centered needs
 - Patient-centered engagement and satisfaction
 - Primary care provider
 - Specialty care provider
 - Genetic services
 - Other community resources

- Mechanisms for continuous improvement of care, discovery and innovation
 - Patients enrolled in registries
 - Patients enrolled in clinical studies or trials
 - Demonstrated improvements in care
 - Demonstrated improvements in outcomes

Appendix 3: Recommended Uniform Screening Panel

Recommended Uniform Screening Panel
Core Conditions
 (As of November 2016)

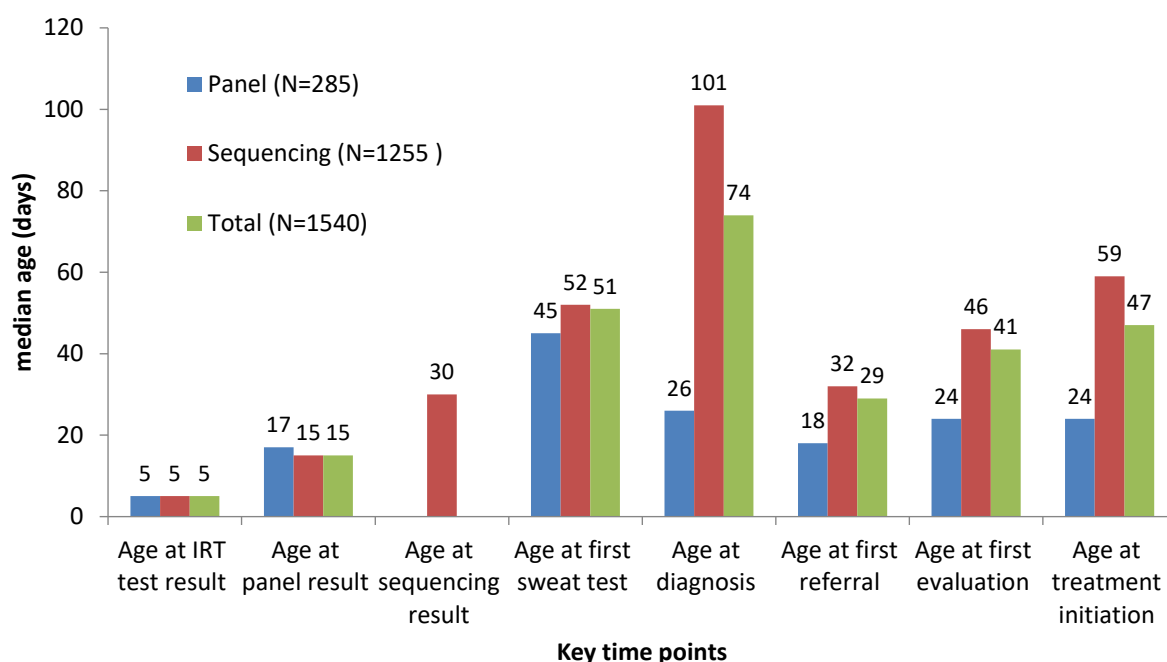
Core Condition	Metabolic Disorder			Endocrine Disorder	Hemoglobin Disorder	Other Disorder
	Organic acid condition	Fatty acid oxidation disorder	Amino acid disorder			
Propionic Acidemia	X					
Methylmalonic Acidemia (methylmalonyl-CoA mutase)	X					
Methylmalonic Acidemia (Cobalamin disorders)	X					
Isovaleric Acidemia	X					
3-Methylcrotonyl-CoA Carboxylase Deficiency	X					
3-Hydroxy-3-Methylglutaric Aciduria	X					
Holocarboxylase Synthase Deficiency	X					
β-Ketothiolase Deficiency	X					
Glutaric Acidemia Type I	X					
Carnitine Uptake Defect/Carnitine Transport Defect		X				
Medium-chain Acyl-CoA Dehydrogenase Deficiency		X				
Very Long-chain Acyl-CoA Dehydrogenase Deficiency		X				
Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency		X				
Trifunctional Protein Deficiency		X				
Argininosuccinic Aciduria			X			
Citrullinemia, Type I			X			
Maple Syrup Urine Disease			X			
Homocystinuria			X			
Classic Phenylketonuria			X			
Tyrosinemia, Type I			X			
Primary Congenital Hypothyroidism				X		
Congenital adrenal hyperplasia				X		
S,S Disease (Sickle Cell Anemia)					X	
S, β-Thalassemia					X	

Core Condition	Metabolic Disorder			Endocrine Disorder	Hemoglobin Disorder	Other Disorder
	Organic acid condition	Fatty acid oxidation disorder	Amino acid disorder			
S,C Disease					X	
Biotinidase Deficiency						X
Critical Congenital Heart Disease						X
Cystic Fibrosis						X
Classic Galactosemia						X
Glycogen Storage Disease Type II (Pompe)						X
Hearing Loss						X
Severe Combined Immunodeficiencies						X
Mucopolysaccharidosis Type 1						X
X-linked Adrenoleukodystrophy						X

Appendix 4: California Newborn Screening Follow- Up of Cystic Fibrosis

Figure 1.

Median Age at Key Screening Points for Screen Positive Cases: July 16, 2007–June 30, 2015



Bibliography of papers on Cystic Fibrosis and Cystic Fibrosis Related Metabolic Syndrome cases identified by the Genetic Disease Screening Program:

Currier RJ, Sciortino S, Liu R, Bishop T, Alikhani Koupaei R, Feuchtbaum L. Genomic sequencing in cystic fibrosis newborn screening: what works best, two-tier predefined CFTR mutation panels or second-tier CFTR panel followed by third-tier sequencing? Genetics in medicine. 2017.

Keiles, S., et al. (2012). "Impact of IVS8-(TG)m(T)n on IRT and sweat chloride levels in newborns identified by California CF newborn screening." J Cyst Fibros 11(3): 257-260.

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Feuchtbaum, L., et al. (2012). "Birth prevalence of disorders detectable through newborn screening by race/ethnicity." *Genet Med* 14(11): 937-945.

Lefterova, M. I., et al. (2016). "Next-Generation Molecular Testing of Newborn Dried Blood Spots for Cystic Fibrosis." *J Mol Diagn* 18(2): 267-282.

Salinas, D. B., et al. (2016). "Benign and Deleterious Cystic Fibrosis Transmembrane Conductance Regulator Mutations Identified by Sequencing in Positive Cystic Fibrosis Newborn Screen Children from California." *PLoS One* 11(5): e0155624.

Schrijver, I., et al. (2016). "The Spectrum of CFTR Variants in Nonwhite Cystic Fibrosis Patients: Implications for Molecular Diagnostic Testing." *J Mol Diagn* 18(1): 39-50.

Schrijver, I., et al. (2008). "Multiplex ligation-dependent probe amplification identification of whole exon and single nucleotide deletions in the CFTR gene of Hispanic individuals with cystic fibrosis." *J Mol Diagn* 10(4): 368-375.

Screen Shots of CF Data Forms

GDSP-SIS

Biobank Data Intake Entity Monitor Follow Up Center CCC Utilities

Client/Case Search
Id Search
View Client Profile
1st T TRF Data Entry (PNS)
2nd T TRF Data Entry (PNS)
Case Summary (PNS)
TRF Data Entry (NBS)
Case Summary (NBS)
Lab CofC
View Edit Log
Resolve Duplicates
Maint Client Relation
Upload CCR
Add Test Results
Audit Data Entry
Biotransfer Expected List
Expected Recall (NBS)
View List of CCR
Case Resolution
HPKV Registry Search
Hait-Bank Center Services
Reassign Follow-up Center
Cystic Fibrosis Service Report
MS/MS Ref Lab Expctd
Enter Other Mutation Test Results
View Holding Tank Records
Expected Phe Monitoring
MC Annual Patient Summary
ALD Annual Patient Summary
CFC Annual Patient Summary
View CFF File Records
Search for 1st T Specimen
Endocrine Service Report
Hemoglobinopathy Service Report
HD Annual Patient Summary
EC Annual Patient Summary
SCPD Confirmatory Results List
SCPD Service Report
SCPD Annual Patient Summary

Data Intake - CFC Annual Patient Summary

Client Name: _____ Date of Birth: _____
Accession Number: _____

Cystic Fibrosis Center Annual Patient Summary

Select	Date	UserID	Case Status	Submitted
<input type="radio"/>	2016		Active - patient currently being seen at this center	<input checked="" type="checkbox"/>
<input checked="" type="radio"/>	2015		Active - patient currently being seen at this center	<input checked="" type="checkbox"/>
<input type="radio"/>	2014		Active - patient currently being seen at this center	<input checked="" type="checkbox"/>
<input type="radio"/>	2013		Active - patient currently being seen at this center	<input checked="" type="checkbox"/>

New View Update

Date: _____
User ID: _____
Cystic Fibrosis Center Code: _____
Cystic Fibrosis Center Name: CHILDREN'S HOSPITAL

Patient Information:

Complete the Patient Information section before completing the rest of the APS

Age of patient today: 4
The current diagnosis on record for the patient is: _____
Additional information: _____
Patient Cystic Fibrosis Foundation Number: _____
Last known patient address and telephone number on record: _____

*As far as you know, is this still the patient's address? No
If no, please provide patient's new address:
Address (Number, Street): _____
Address (Apt / Suite #): _____
City: _____
State: Select _____
Zip: _____
☐ Do not validate address
☐ Unknown

*As far as you know, is this still the patient's telephone number? No
If no, please provide patient's new home telephone number: _____ X _____
☐ Phone Number Unknown

*Follow-up Status: Active - patient currently being seen at this center
Date (MM/DD/YYYY): _____
If transferred, indicate new center: SELECT _____
If patient died, indicate date of death: _____
Was death a result of a complication of cystic fibrosis? Select _____
Cause of Death: _____
APS Start Date (MM/DD/YYYY): 2014
APS End Date (MM/DD/YYYY): 2015

Last Visit Information

*Date of last visit/interaction with patient that occurred in the previous year: 1/4/2014 through 1/3/2015 ☐ Patient not seen during this year

Last anthropometric measures and pulmonary function tests taken in the previous year 1/4/2014 through 1/3/2015

Measure/Test	Value	Unit	Date	
Height		Centimeters	2014	<input type="checkbox"/> Unknown (N/A)
Weight		Kilograms	2014	<input type="checkbox"/> Unknown (N/A)
FVC		Liters		<input checked="" type="checkbox"/> Unknown (N/A)
FEV 1.0		Liters		<input checked="" type="checkbox"/> Unknown (N/A)
FEV 0.5		Liters		<input checked="" type="checkbox"/> Unknown (N/A)

The Role of Quality Measures Report, 2/8/2018

☐ Not Done or ☒ Unable to Perform Reliable Test
 Total number of FEV1 measurements taken in the previous year 1/4/2014 through 1/3/2015
 If more than 0, average FEV1 value (L):
 If 0, select:
☐ Not Done or ☒ Unable to Perform Reliable Test

Did patient have a transplantation (check all that apply)?
☐ Lung
☐ Heart
☐ Liver
☐ Other (specify):

Type of insurance at last visit of year (check all that apply)?
☒ Medicaid/CCS/Public Program ☐ Self-Pay/Uninsured
☐ Military/Government/CHAMPUS ☐ Unknown (N/A)
☐ Private Insurance/Employer Sponsored ☐ Other

Services Provided

*Services provided by your cystic fibrosis center in the previous year 1/4/2014 through 1/3/2015
☐ Consultation with Family ☐ Consultation with PCP
☐ Genetic Counseling ☒ Laboratory Tests/Interpretation of Results
☐ No services provided in the past year ☒ Nutrition Management
☒ Parent Education ☒ Physical Examination
☐ Physical Therapy ☐ Psychological Services
☒ Respiratory Therapy ☐ School/Preschool-Related Issues
☒ Social Services ☐ Unknown (N/A)
☐ Other services:

*Total number of scheduled/planned patient visits kept at your cystic fibrosis center in the previous year: 2014 through 2015 including well care visits 2 Unknown (N/A)

*In the previous year, has the caregiver missed more than two scheduled appointments? No

If yes, what were the barriers to care?
☐ Clinic Communication Error ☐ Parent Working or Schedule Conflict
☐ No Insurance/Financial barrier ☐ Transportation
☐ No Response from Parent ☐ Unknown
☐ Language ☐ Other:

*Total number of hospitalizations in the previous year: 2014 through 2015 0 Unknown (N/A)

If more than 0, indicate date of admission, total days hospitalized and reason for admission:

Admission Date: - -
 Total Days Hospitalized:
 Reason:

Admission Date	Total Days Hospitalized	Reason
----------------	-------------------------	--------

*Total number of home IV Events in the previous year 1/4/2014 through 1/3/2015 0 Unknown (N/A)
 If more than 0, total days on home IV in the previous year 1/4/2014 through 1/3/2015 Unknown (N/A)

Cystic Fibrosis Therapies/Treatments

* Indicate all therapies prescribed to the patient in the previous year 1/4/2014 through 1/3/2015
☐ Enteral Supplemental Feeding ☐ Parental Supplemental Feeding
☐ Pancreatic Enzyme Supplements ☐ Oral Antibiotic Therapy
☐ Anti-Inflammatory Therapy ☐ Lung Percussion Therapy
☐ Oxygen Therapy ☒ Treatment Not Deemed Necessary
☐ Unknown ☐ Other therapies 1:(specify)
☐ Other therapies 2:(specify) ☐ Other therapies 3:(specify)
☐ Other therapies 4:(specify) ☐ Other therapies 5:(specify)

* Indicate all medications/supplements prescribed in the previous year 1/4/2014 through 1/3/2015

- ☐ Pancreatic enzyme supplements
- ☐ Ursodeoxycholic Acid
- ☐ Acetylcysteine/Mucomist
- ☐ Dornase alfa (i.e., Pulmozyme)
- ☐ Hypertonic saline (inhaled)
- ☐ Tobramycin (TOBI, inhaled)
- ☐ Other Aminoglycosides (inhaled, e.g. Gentamycin)
- ☐ Colistin (inhaled)
- ☐ Cayston (inhaled)
- ☐ Tobramycin (chronic, oral)
- ☐ Trimethoprim (chronic, oral)
- ☐ Ciprofloxacin (chronic, oral)

- ☐ Cephalosporins (chronic, oral, e.g., Penicillin, Amoxicillin)
- ☐ Macrolides (chronic, oral, e.g., Azithromycin)
- ☐ Ibuprofen (high-dose 25-30 mg/kg, oral)
- ☐ Corticosteroids (oral, e.g., prednisone)
- ☐ Corticosteroids (inhaled, e.g., Fluticasone, Flovent, Budesonide)
- ☐ Antifungals (oral, e.g., Itraconazole, Sporanox)
- ☐ Bronchodilators (oral)-beta agonists (e.g., Proventil Repetabs, Vomax)
- ☐ Bronchodilators (oral)-theophylline product (e.g., Theodur, Slo-bid, Uniphyll)
- ☐ Bronchodilators (inhaled)-beta agonist/short acting (e.g., Albuterol, Proventil, Ventolin)
- ☐ Bronchodilators (inhaled)-beta agonist/long acting (e.g., Salmeterol, Serevent, Foradil)
- ☐ Bronchodilators (inhaled)-anticholinergic/short acting (e.g., Ipratropium, Atrovent)
- ☐ Bronchodilators (inhaled)-anticholinergic/long acting (e.g., Tiotropium, Spiriva)
- ☐ Bronchodilators (inhaled)-combination (e.g., Combivent, Duoneb)
- ☐ Kalydeco (Ivacaftor)
- ☐ Vitamins (e.g. ADEKs, AquaADEKs, D-Vi-Sol, MVI, Vitamin D, Vitamin K, Multivitamins)
- ☐ Oral Nutritional Supplemental (e.g. Ensure, Scandishake, Calogen)
- ☐ Salt Supplementation
- ☐ Laxative (e.g. Miralax, ExLax, Milk of Magnesia, Dulcolax, Metamucil, Citrucel)
- ☐ H2 blocker (e.g. Tagamet, Zantac, Pepcid, Axid)
- ☐ Proton Pump Inhibitor (e.g. Omeprazole, Prilosec, Prevacid, Nexium, Lasoprazole, Zegard)
- ☐ RSV Immunoprophylaxis (e.g. Synagis, Palivizumab)
- ☐ Probiotics (e.g. Lactobacillus, Lactic Acid Bacteria, Bifidobacteria, Saccharomyces boulardii)
- ☐ Other medications/ supplements 1:(specify) _____
- ☐ Other medications/ supplements 2:(specify) _____
- ☐ Other medications/ supplements 3:(specify) _____
- ☐ Other medications/ supplements 4:(specify) _____
- ☒ No medication

Respiratory Cultures

*Total number of respiratory cultures performed in the previous year 1/4/2014 through 1/3/2015 ☐ Unknown (N/A)

If 0, select:

- ☒ Not Done ☐ Unable to Perform Reliable Culture

If more than 0, check all that apply and select types of cultures obtained

- | | | | |
|---|---------------------------------|---------------------------------|---------------------------------------|
| <input type="checkbox"/> No growth/sterile culture | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Normal flora | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Aspergillus (any species) | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Burkholderia cepacia | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Haemophilus influenzae (any species) | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> MRSA (Methicillin resistant Staph aureus) | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Pseudomonas aeruginosa (mucoid) | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Pseudomonas aeruginosa (non-mucoid) | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Pseudomonas aeruginosa (unknown) | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Staphylococcus aureus | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Stenotrophomonas (Xanthomonas) maltophilia | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Other Pathogen 1:(specify) _____ | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Other Pathogen 2:(specify) _____ | <input type="checkbox"/> Sputum | <input type="checkbox"/> Throat | <input type="checkbox"/> Bronchoscopy |
| <input type="checkbox"/> Unknown (N/A) | | | |

Health Problems

*Indicate all persistent health problems/symptoms the patient experienced in the previous year (check all that apply):

- ☒ None
☐ Unknown

- | | |
|--|--|
| <input type="checkbox"/> Air Trapping | <input type="checkbox"/> Pancreatic Insufficiency |
| <input type="checkbox"/> Bronchiectasis | <input type="checkbox"/> Abnormal Stool/Malabsorption |
| <input type="checkbox"/> Mucus Plugging | <input type="checkbox"/> Failure to thrive/Malnutrition |
| <input type="checkbox"/> Asthma | <input type="checkbox"/> Distal Intestinal Obstructive Syndrome (DIOS) |
| <input type="checkbox"/> Allergic Bronchial Pulmonary Aspergillosis (ABPA) | <input type="checkbox"/> Pancreatitis |
| <input type="checkbox"/> Nasal Polyps/Sinus Disease | <input type="checkbox"/> Gastroesophageal Reflux Disease (GERD) |
| <input type="checkbox"/> Electrolyte Imbalance | <input type="checkbox"/> Liver Disease |
| <input type="checkbox"/> Bone Disease | <input type="checkbox"/> Renal Disease |
| <input type="checkbox"/> Hearing Loss | <input type="checkbox"/> CF-Related Diabetes |
| <input type="checkbox"/> Vision Problems | <input type="checkbox"/> Edema/Hypoproteinemia/Hypoalbuminemia |
| <input type="checkbox"/> Other Problem 1:(specify) _____ | <input type="checkbox"/> Other Problem 2:(specify) _____ |

*Global Health Assessment (Select the number that describes the overall health status of the patient at the last visit compared to typical child without cystic fibrosis):

Comments _____

Appendix 5: Mountain States Genetics Collaborative medium chain acyl-CoA dehydrogenase deficiency (MCADD) QI Study

Table 1.

Documentation of the Following in Patient Chart	Charts Reviewed (N=11)
Fasting time identified and discussed	9/11
Updated Polycose protocol available	4/11
Emergency letter available	11/11
Management of illness and fasting discussed	8/11
Antiemetics prescribed	0/11
Car seat sticker discussed	2/5 (age appropriate)
Medic-Alert bracelet discussed	7/11
Chart Pop-Up in place	1/11

Epic Smart Phrase:

MCADD MANAGEMENT:

- Emergency Letter: {ED Ltr Provided:33113}
- Anesthesia Letter: {Anes Ltr Provided:33115}
- Current allowed fasting time WHEN WELL: *** hours. Patient/Family was counseled that there is no specific safe fasting time when ill.
- Acute Management Glucose Source: {Glucose Source:33117}
- Car Seat Alert Stickers: {Car Seat Stickers:33119}
- Medic Alert Bracelet: {Medic Alert Bracelet:33120}
- Flu Shot: {flu shot:33169}
- Patient Highlight/FYI: {FYI Alert:33122}
- Perceived Barriers to Care: {Barriers to care:33123}.

Appendix 6: Treating Congenital Hypothyroidism: 2016 Quick Guide for Primary Care Provider

Treating Congenital Hypothyroidism (CH)

2016 Quick Guide for Primary Care Providers



After Receiving Positive Newborn Screening (NBS) Results

- **Within 24 hours** of receiving a positive NBS result for CH (TSH ≥ 29 μ IU/mL), test serum TSH and free T4 (or total T4) for confirmatory diagnosis

- **If newborn screening TSH result is:**

> 40 μ IU/mL	29 – 40 μ IU/mL
Initiate treatment as soon as a serum sample is collected and refer to a pediatric endocrinologist	May wait for the results of confirmatory serum test to initiate treatment

If confirmatory serum TSH result is:

- **> 40 μ IU/mL**
Initiate levo-thyroxine treatment immediately
- **10 – 40 μ IU/mL**
Repeat the free T4 (or total T4) and TSH tests but do not start levo-thyroxine treatment yet
- **< 10 μ IU/mL**
Considered normal, no treatment needed

You are strongly encouraged to work closely with a pediatric endocrinologist to perform diagnostic evaluation, initiate treatment, and coordinate for ongoing care.

Treatment must begin within 2 weeks of age for confirmed cases.

Levo-thyroxine Treatment Dosing

- **Start at 10 – 15 μ g/kg per day**
(Use 15 μ g/kg if free T4 < 0.5 ng/dL or total T4 < 5 μ g/dL)
- **Either brand name or generic**, but stay with the same formulation, if possible. **USE TABLETS, DO NOT USE LIQUID FORM**
- Maintain TSH concentration in the age-specific reference range
- Maintain free T4 (or total T4) concentration in the upper half of the lab-provided reference range

Approximate daily dose by weight:

Weight (grams)	Daily Dose
2000 – 2499	25 μ g
2500 – 3999	37.5 μ g
4000 or more	50 μ g

Follow-up Frequency

- **2 weeks after treatment initiation:**
The first clinical follow-up examination and lab tests should take place
- **Then every 2 weeks:**
Evaluate clinical symptoms and thyroid function until **complete normalization of TSH** is reached

Recommended follow-up schedule by age, once TSH is normalized:

Age	Frequency
< 6 months	Every 1–2 months
6 months to 3 years	Every 2–4 months
After 3 years	Every 6–12 months

Reference: American Academy of Pediatrics, Rose SR, et al. **Update of newborn screening and therapy for congenital hypothyroidism.** Pediatrics. 2006;117:2290-2303.

- also:**
- **More frequent testing** when compliance is questionable and/or
 - **One month after** any dosage or formulation change

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Treating Congenital Hypothyroidism (CH)

2016 Quick Guide for Primary Care Providers

page 2



Special Considerations in Patient Case-Management

Transient Congenital Hypothyroidism

- **CH is estimated to be transient, not permanent, in 5% to 15% of cases diagnosed through newborn screening**
- **Causes**
 - Maternal anti-thyroid medications
 - Maternal anti-TSH-receptor antibodies
 - Excessive/deficient maternal iodine intake
 - Post-natal iodine exposure (povidone-iodine)

If you suspect Transient CH

When in doubt, start treatment to ensure adequate thyroid hormone for brain development

Consider trial off in patients who have not needed increases in levothyroxine dosage after age 1 year

After 3 years of age, safe to do a trial off levothyroxine for 4–6 weeks, and then retest free T4 (or total T4) and TSH

Reasons TSH May Remain Elevated During Treatment

- Non-compliance
- Improper delivery – suspension
- Malabsorption – lactose intolerance, milk allergy
- Other medications – iron, calcium, antacids
- Soy formulas and acidic juices
- Delayed maturation of hypothalamic-pituitary-thyroid axis

Regular (non-soy) formula should be given at least 30 minutes before or after medication

Important Information to Convey to Parents and Caregivers

Let them know that congenital hypothyroidism is usually permanent

Teach the correct technique of medication administration

Stress that adherence to treatment is key to normal development for all ages

Describe the follow-up lab testing schedule

The CDPH Parent Education Brochure (English/Spanish) for Congenital Hypothyroidism, as well as other resources, can be found at <http://www.cdph.ca.gov/nbs>.

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Appendix 7: the NBSTRN LPDR Public Health Questions and Research Activities

The NBSTRN has developed two sets of public health questions in their LPDR that form a short set of questions that apply to long-term follow-up of any child identified by a NBS program.

One set of questions focuses on the population of children with conditions identified by newborn screening served by a health department:

- Is the disorder on the 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 this disorder?
- 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?
- 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?

The other set focuses on individual children with priority for the first four questions:

- A. Diagnosis
- B. Condition Specific Care within the Past 12 Months
- C. Date or Age of Appropriate Intervention
- D. Alive or Deceased

- E. Reason reported for no treatment in the last 12 months?
- F. Cause of death?
- G. Developmentally appropriate?
- H. How many ER visits in the last 12 months?
- I. How many hospitalizations in the last 12 months?
- J. How many clinic visits in the last 12 months

The primary focus of the NBSTRN LDPR is on supporting research on individual NBS conditions. Quality measures provide an important contrast to research databases such as the NBSTRN Longitudinal Pediatric Data Resource (LPDR) that are comprehensive collections of data elements designed for new knowledge discovery and testing research hypotheses. Quality measures are indicators used to assess progress towards pre-defined quality targets. Research databases can play two important roles in the development of quality measures by helping to precisely define the components of a quality measure making them easier to implement, and by providing test data to validate new quality measures using existing populations of patients receiving care for a specific condition. Research databases have different consent requirements as quality measures are typically considered an essential and integral part of care delivery. Some research databases remove or conceal personal identifiers while quality measures more commonly rely on identified data to enable follow-up data collection and analysis and attribute responsibility. Both types of data may report population based de-identified results.

Appendix 8: Lessons for NBS from Quality Measures Developed for Other Childhood Chronic Diseases

Quality measurement activities for Asthma the Inner-City Asthma Consortium:

Asthma is the most common chronic disease in the United States, and children living in urban areas are disproportionately affected. Federal funding for the Inner-City Asthma Program began in 1991, with the goal of developing ways to more effectively treat asthma in inner city children. The initial National Cooperative Inner-City Asthma study, enrolling 1500 subjects, was followed by the NIEHS and EPA funded Inner-City Asthma Study, a CDC-NIAID funded collaborative on behavioral and patient education, and an Inner-City Asthma consortium, primarily focused on Immune based therapies. These intervention networks were successful in showing that structured ongoing follow up, access to medication, and environmental surveillance and mitigation could reduce symptom days and hospitalizations.

Program activities have focused on measurement of asthma severity and identification of barriers to adherence with asthma management guidelines. A large component of consortium activities was dedicated to measurement of environmental exposures and mitigation of risks for asthma exacerbation. More recently, the School Inner-City Asthma Intervention Study shifted attention to mitigation of environmental exposures in the school setting, through the School Inner-City Asthma Intervention Study.

Quality measures used in these studies include validated survey tools for families and schools, physical assessment of growth, spirometry, assessment of inflammation using expired nitric oxide, and epidemiology surveillance of microbial and viral pathogens. Interventional activities include environmental inspections, staff education, and allergen mitigation activities.

Implications for NBS follow-up: The consortium was successful in organizing primary care networks and school based contacts to successfully reduce exposure to asthma environmental triggers and promote adherence to controller medications. Although most newborn screen detected conditions are much rarer than asthma, these studies show that community engagement and school involvement can extend the reach of specialty providers in promoting adherence and providing outcome data.

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Appendix 9: Collation of Lessons Learned from NBS Follow-up Programs

1. Specific Conditions	
A. Sickle Cell Disease	<ul style="list-style-type: none"> therapies such as immunizations and prophylactic antibiotics that have strong evidence showing effectiveness have not been used for all children with sickle cell disease at the optimal time; programs for quality improvement can create higher compliance rates; sickle cell disease is cared for in a variety of settings; it is important to encourage cooperation and engagement of primary care, specialists, and emergency physicians to optimize care; long-term funding can support data collection, analysis, and quality improvement activities.
B. Registry and Surveillance System for Hemoglobinopathies	<ul style="list-style-type: none"> this method of longitudinal data collection, especially for a patient population that often receives healthcare outside of specialty care centers, provides a comprehensive understanding of the entire spectrum of patients; this surveillance system has provided helpful information to strengthen other projects, such as HRSA's SCDTDP program and NHLBI's Implementation Research project; maintaining consistent and long-term staffing, data sharing agreements, and funding are paramount to the success of this program.
C. Cystic Fibrosis Foundation Comparative Outcomes	<ul style="list-style-type: none"> CFF accredited centers were initially reluctant to allow transparency of their data, but accepted the change and have shared care strategies between centers for quality improvement purposes; families engaged in clinical care at a CFF accredited center will partner with personnel at that center to improve care and quality measures; understanding the course of the disorder over the lifespan requires well-defined and consistent measures collected across centers.
D. California Newborn Screening Follow-Up of Cystic Fibrosis	<ul style="list-style-type: none"> short- and long-term follow-up data collection about children with CF along with a burgeoning genetic database has helped inform appropriate diagnosis and interventions for CF through information sharing, scientific collaboration and publication; California has leveraged its relationships with the CF Foundation approved centers and utilized these state-contracted centers to provide patient access to centers of excellence for lifelong CF care; follow-up data collection and analysis has been successfully used to provide feedback to practitioners to improve timeliness of diagnosis and information on other outcomes of interest. replication of this model for other conditions would be challenging because collection of detailed and validated data with little

	<p>ascertainment bias benefits from the collaboration with nationwide private support from the CF Foundation for centers of excellence;</p> <ul style="list-style-type: none"> • other states may not be able to replicate this approach due to a lack of funding and there is a need to explore more limited strategies; • a limit of this model is collection of data only to age 5 years, before many of the long-term effects of CF (and of other conditions identified by NBS) become apparent.
E. Mountain States Genetics Collaborative medium chain acyl-CoA dehydrogenase deficiency (MCADD)	<ul style="list-style-type: none"> • integrating quality activities into routine care is feasible and can overcome the need for separate funding and duplicate data entry; • display of important newborn screening condition related information at the time of an emergency room visit or when seeing a new provider is feasible if appropriate care plans and data collection forms are integrated into an EHR (and assuming the new provider has access to that same EHR); • quality activities for one screened disorder can be modified to become applicable to other disorders.
F. CDC Early Hearing and Detection Intervention (EHDI)	<ul style="list-style-type: none"> • custodianship and maintenance of measures is important; • more organizations need to be encouraged and facilitated in navigating the process of creating standardized, validated, measures; • ascertainment bias or loss to follow up for data collection is a challenge to the quality improvement system.
G. California Long-term Follow-up of NBS Primary Congenital Hypothyroidism	<ul style="list-style-type: none"> • This approach using primary care providers, may be appropriate for states with smaller populations than California as a method to collect long term follow-up data for children with PCH and perhaps, other newborn disorders; • the model requires a minimal infrastructure investment, project management, recruitment staffing and part-time epidemiological consulting resources that might be already available within state health departments; • the model relies on identification of and payment to the data holders (primary care providers); • the software is agile and the database itself is maintained on the cloud. • this use case also illustrates the challenges of acquiring more complete follow up data.
H. Organic Acidemia Association Collects Data from Families	<ul style="list-style-type: none"> • patients and families are the only source for accurate input of certain kinds of data and outcomes. • ascertainment bias may occur as study participants are self-selected and may not be representative of the full spectrum of the disorder; • patients and families will not always be aware of or agree with "data dictionaries" for elements of reporting.

2. All Conditions Identified by NBS	
A. National Coordinating Center for Regional Genetics Networks	<ul style="list-style-type: none"> regional collaboratives can improve access to high-quality treatment of NBS conditions; telemedicine can help overcome issues related to access to specialists and can be used for education of providers.
B. California Department of Health Long-Term Follow-up	<ul style="list-style-type: none"> with appropriate resources and infrastructure, State NBS programs can provide LTFU of specific conditions.
C. University of Maryland Study of LTFU of Newborn Screening in Primary Care	<ul style="list-style-type: none"> It is feasible to carry out quality measurement in primary care settings; important future targets should focus on communication of information; practices can assess their medical home capabilities which is an important component of newborn screening follow-up. data collection can be labor-intensive and costly
D. Newborn Screening Translational Research Network (NBSTRN)	<ul style="list-style-type: none"> providers will participate without financial incentive; data aggregation and sharing is critical for rare diseases; standardized vocabularies are helpful; data security is important.
E. NewSTEPS	<ul style="list-style-type: none"> data that is locally identifiable but de-identified and combined at a national level can improve access to data at multiple levels without redundant mechanisms; a national database could aid in LTFU as patients move state to state.
F. NBS Connect: A Web-Based Self-Report Registry for Patients with Disorders Identified by Newborn Screening	<ul style="list-style-type: none"> The results presented in these two papers highlight the value of patient self-report registry data. Patients are willing to register and use a website for data entry and a single website can serve multiple conditions identified by newborn screening.
3. Children with Special Health Care Needs (CSHCN)	
A. National Survey of Children's Health (NSCH)	<ul style="list-style-type: none"> national surveys may be an important tool to gather data on the entire population and not just patients served by a specific provider or health department; there may be an opportunity to modify the survey to generate NBS data.

4. All Children	
A. HEDIS	<ul style="list-style-type: none"> requirements of insurance programs are strong motivators for participation; data collection and reporting tools are in place to make audits feasible; available measures focus on common problems such as adult chronic diseases or throat infections in children; measures relevant to children with NBS conditions are not included (e.g. transition to adult systems of care); children identified by NBS cannot easily be studied using this approach; NCQA tools to assess medical home status may be helpful, but credibility of this approach for NBS depends on considering the needs of children with special healthcare needs.
B. Pediatric Quality Measures Program	<ul style="list-style-type: none"> PQMP measures and also knowledge gained from their uptake and implementation can be leveraged to develop long-term follow up measures of NBS emphasis should not be placed solely on developing measures that are scientifically sound (reliable/valid) but measures must also be useable and feasible stakeholder input from those implementing the measures and also from families and others key users of the data/information that comes from measures is key to ensuring measure usability and feasibility
C. Health Resources and Services Administration, Maternal and Child Health Bureau (MCHB) - Title V	<ul style="list-style-type: none"> this type of long term and continuous measurement of outcomes can be used as a model for NBS quality improvement activities.
E. National Patient-Centered Clinical Research Network (PCORnet)	<ul style="list-style-type: none"> local and regional data collection can be combined across research networks to improve research efficiency and ease

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