

**Long-term follow-up after diagnosis resulting from newborn screening: statement of the US Secretary of Health and Human Services' Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children.**

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**Abstract:**

The US Secretary of Health and Human Services' Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children provides guidance to reduce the morbidity and mortality associated with heritable disorders, with a special emphasis on those conditions detectable through newborn screening. Although long-term follow-up is necessary to maximize the benefit of diagnosis through newborn screening, such care is variable and inconsistent. To begin to improve long-term follow-up, the Advisory Committee has identified its key features, including the assurance and provision of quality chronic disease management, condition-specific treatment, and age-appropriate preventive care throughout the lifespan of affected individuals. There are four components central to achieving long-term follow-up: care coordination through a medical home, evidence-based treatment, continuous quality improvement, and new knowledge discovery.

**Keywords:** neonatal screening | comprehensive health care | heritable disorders | genetic diseases | diagnosis

**Article:**

Newborn screening is an essential public health function provided to all newborns in the United States. The newborn screening system is intended to be comprehensive, including not only screening and diagnosis, but also long-term follow-up care through the medical home.<sup>1</sup> All of

the conditions identifiable through newborn screening are chronic, and therefore require medical care and other related services throughout the affected individual's lifetime. Unfortunately, the long-term follow-up activities within public health programs lack coordination and have been of low priority for funding compared with activities related to screening and diagnosis.<sup>2,3</sup>

In the treatment and management of a condition, affected individuals and their families may regularly interact with generalist physicians, specialist physicians, primary care and specialty nurses, clinical psychologists, dentists, nutritionists, genetic counselors, occupational and physical therapists, education specialists, public health nurses, social workers, and pharmacists. These interactions are complex because most conditions identified through newborn screening have a wide variety of manifestations and related comorbidities, the treatment evidence-base is not always definitive, effective models of comanagement are not in place, and there is a maldistribution and lack of accessibility of knowledgeable care providers. Further complicating long-term follow-up, affected individuals, families, and care providers must interact with both private and public payers, and insurance health benefits are variable and often incomplete.

To begin to improve the long-term follow-up component of the newborn screening system, this statement of the US Secretary of Health and Human Services' Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (ACHDGDNC; roster of members available at <http://www.hrsa.gov/heritabledisorderscommittee/governance/roster.htm>) defines the key features of long-term follow-up. The statement was developed from the comments of an expert panel of health care policy experts, public health specialists, generalist and specialist care providers, allied health care providers, and the families of affected individuals convened in April 2007, and approved by the ACHDGDNC. It greatly expands the concept of long-term follow-up from data management to systematic and comprehensive care of affected individuals. The statement does not address how these components will be implemented or supported; those discussions are ongoing.

## GOAL OF LONG-TERM FOLLOW-UP

The principal goal of long-term follow-up is to assure the best possible outcome for individuals with disorders identified through newborn screening. The time frame for long-term follow-up is the lifespan of the affected individual; however, the responsibility of the ACHDGDNC as set by its authorizing legislation is from birth to age 21 years.

## DEFINITION OF LONG-TERM FOLLOW-UP

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.

## COMPONENTS OF LONG-TERM FOLLOW-UP CARE

### Care coordination through a medical home

The ACHDGDNC supports the concept that all individuals diagnosed with a condition through newborn screening should have a medical home to integrate care and ensure quality and safety in care delivery.<sup>4,5</sup> As the usual place for sick and well care, the medical home should be family-centered, culturally effective, accessible, actively engaged in the coordination and provision of primary and subspecialty health care services within the health care system and across other community-based agencies and services (e.g., other clinicians, educational programs, and community-based counseling and support services),<sup>4,5</sup> and facilitate requisite referrals. Systems will need to be developed to assure that individuals transition to adult care services without losing a medical home; this is central to the receipt of long-term follow-up care.<sup>2,6</sup>

### Evidence-based treatment

Provision of condition-specific therapy is a critical component of long-term follow-up care. For many of the conditions identified by newborn screening, specific and often expensive therapeutic agents, including specialized nutritional formulas, are required over the life of the affected individual. For some of the conditions, the most effective treatment is uncertain. The current evidence base for these conditions is limited because their rarity and heterogeneity makes randomized controlled trials and even prospective observational cohort studies difficult to carry out.<sup>7</sup> More emphasis needs to be placed on collating and distributing “best practices” based on existing evidence, even for those conditions with evolving treatment protocols. This process will be central to the development of clinical care guidelines. A critical component of long-term follow-up care is a coordinated mechanism for both collecting and synthesizing information about effective treatments and connecting affected individuals with the most effective treatments or clinical research trials if the appropriate management is uncertain.

### Continuous quality improvement

Long-term follow-up data systems should allow clinicians, experts in quality improvement, researchers, affected individuals and their families, public health officials, and policy makers to evaluate the care that affected individuals and their families actually receive. These activities should focus on three separate areas: adherence to clinical guidelines or accepted best practice, evaluation of the effectiveness of various treatment/intervention protocols, and assessments of the experience with care of affected individuals and their families. These data could be analyzed to identify problems in quality of care across conditions and care settings that could be remedied with more broad-based actions such as technical assistance for quality improvement. Long-term follow-up information systems that capture appropriate clinical and other relevant care information will be required to assure quality improvement. Integrating such care information with public health data and the analysis of the information will provide knowledge to support informed decision-making that addresses clinical, public health, and health policy needs.

#### New knowledge discovery

Long-term follow-up information systems should provide a platform for basic science and clinical researchers to assess genotype-phenotype relationships, including the natural history of conditions, and to engage individuals and families as partners in research to evaluate the impact of interventions, including novel therapies and those current therapies that lack high-quality evidence of effectiveness. Such registry-based approaches to discovery are needed to overcome the challenges of research in rare conditions and fill in critical gaps in the evidence base. If we are to understand the many, often rare conditions detected by newborn screening, it will be important not only to track clinical and laboratory information as children grow, but also to provide their treatment within a research context that is family-centered and culturally competent. To be most efficient, the system developed for quality improvement should also be able to collect data regarding treatment effectiveness.

#### NEXT STEPS

This statement defines the major components of newborn screening long-term follow-up. Although this document addresses the long-term follow-up needs of children identified through the newborn screening system, it is acknowledged that some children will not be included from the outset because of limitations in the screening and short-term follow-up system (e.g., false-negatives) or to unique characteristics of the condition (e.g., late-onset hearing loss) The framework of long-term follow-up, however, applies for all children with a specific condition included in the NBS panel, regardless of how that child's condition is ultimately identified. The ACHDGDNC understands that there are many barriers to implementing long-term follow-up. These barriers are not only related to cost of implementation, but to other health care system factors (e.g., lack of public health infrastructure, maldistribution of providers, inefficient models of comanagement, variations in the availability and use of health information technology, lack of electronic health record interoperability) and to potential consequences for affected individuals

and their families (e.g., loss of privacy and other human subject protection concerns and risks to insurability). The ACHDGDNC will next develop a roadmap for implementation.

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