Newborn Screening

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KEYWORDS

- Newborn screening • Inherited metabolic disorders
- Early hearing detection and intervention • Critical congenital heart disease

KEY POINTS

- Newborn screening is a triumph of public health because of early identification of screened disorders permitting prompt initiation of therapy.
- Every newborn in the United States should have access to newborn screening for a recommended uniform screening panel of conditions.
- Uniform application of newborn screening in the neonatal intensive care unit requires attention to protocols ensuring completion of this essential test both with regard to assuring initial testing and for repeat testing required to assure diagnosis in premature and low-birth-weight infants.

WHAT IS NEWBORN SCREENING?

In 2013, practitioners, public health departments, and families in the United States celebrated 50 years of newborn screening. Beginning in 1963 with screening for phenylketonuria (PKU), now all 50 states, US territories, and the US military, and many countries around the world test newborns for conditions not evident on physical examination that, if not diagnosed and treated, result in disability, disease, or death. Predominantly, screening is undertaken by analysis of blood collected on filter paper spots; it also includes point-of-service tests for hearing loss and critical congenital heart disease (CCHD). Many of the conditions ascertained through newborn screening are inborn metabolic disorders, but screening also ascertains congenital hypothyroidism, congenital adrenal hyperplasia, severe T-cell immunodeficiency (severe combined immunodeficiency [SCID]), cystic fibrosis, and hemoglobinopathies. To a large extent, the disorders found by newborn screening are genetic conditions. The primary goal of newborn screening is the prevention of significant morbidity and mortality related to the screened disorders. The undertaking of newborn screening has been identified as a public health priority, and newborn screening programs are

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primarily the responsibility of state departments of health in the United States or national or regional health agencies in other countries.

NEWBORN SCREENING AND PUBLIC HEALTH

The general concept for justification of newborn screening is that improving outcomes for affected children is productive for society as well as for the individual child.[1] The practice of newborn screening was initiated with the underlying assumption that prevention of significant morbidity and mortality in infants due to detectable diseases was a responsibility of public health agencies. Public health organizations can be granted the authority for universal screening and have the potential for efficiency and quality control needed to ensure that every baby has an equal chance to receive this critical test. Departments of health also often have infrastructure that can facilitate continuing surveillance to ensure continued access to care and monitoring of outcomes (although this is not a potential that has been uniformly realized[2,3]). Newborn screening is thus a public health responsibility and the action of screening is a process, not an event. Screening is not simply the performance of the newborn heel prick that allows drops of blood to be spotted on filter paper and tested. Screening should be understood rather as a system, beginning with the sampling, including delivery of samples to the Department of Health; testing, analysis, and reporting of test results; communication of the results to the family, primary provider, and specialist; follow-up diagnostic testing and initiation of therapy; and long-term follow-up to assure the promise of newborn screening (**Fig. 1**).[4] Newborn screening involves the partnership of many participants such as the baby's parents, hospital providers, and the associated laboratory teams; primary providers; and specialists with departments of health to monitor and assure proper functioning of all aspects of the system. Improvements and efficiencies that allow better functioning of the system are important to continue to protect infants from the consequences of the screened conditions and to realize the societal benefits of early identification and treatment.

Because blood spot testing is undertaken in departments of health, tests used for newborn screening need to be rapid, cost-efficient, and suitable to scaling for high-throughput analysis. Sample size is small, and tests need to be developed with a high degree of rigor to assure appropriate levels of detection while minimizing

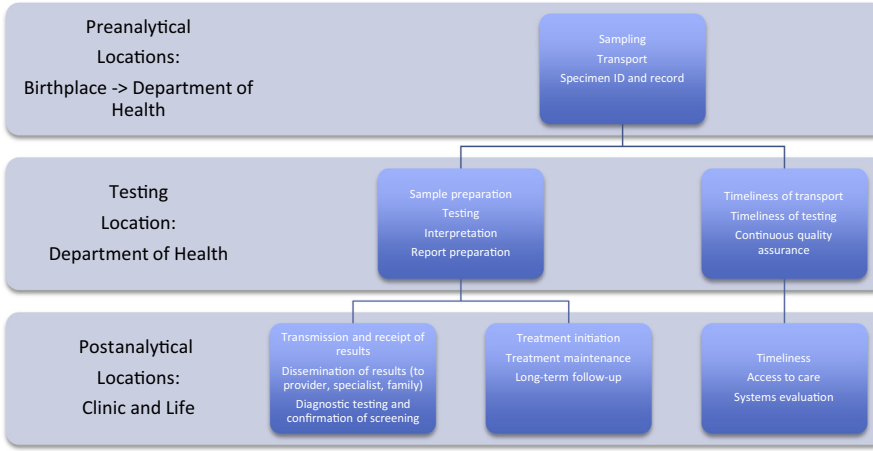

Fig. 1. Components of a newborn screening system.