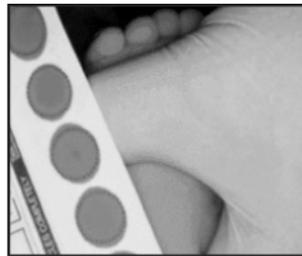




# Newborn Screening Services: Then and Now

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## **Newborn Screening And Genetic Services Then**

The diaper test for phenylketonuria developed 76 years ago marked the beginning of newborn screening and the delivery of genetics services within the system of public health. The development of a system of State-based newborn screening programs is considered one of the greatest accomplishments in public health. However, it took more than 30 more years for the beginning of a system of universal newborn screening when in 1962 the U.S. Children's Bureau funded a national trial for PKU screening.

Public health genetic services traditionally have included not only clinical services like newborn screening and prenatal testing, but also policy development, training and education. However, it was not until the 1950s that a system of genetic services began to be developed. During this time, Congress earmarked funds for children with developmental delays (mental retardation) and congenital heart disease. Later, during the 1960s, maternal and infant health care projects were initiated to provide care for pregnant women at high risk of giving birth to an infant with special health care needs. Programs for the education of health care providers in the practice of genetic medicine also were established at this time.

The maternal and child health program began its focused efforts to improve access to genetic services because of the National Sickle Cell Anemia Control Act of 1972: Statewide support for screening programs for sickle cell disease and other genetic disorders were established. In addition, a system of education and family counseling began that was structured to intersect with the newborn screening process. Thus began a systems approach to the development of newborn screening programs, which is in place today. Throughout 1960s and 70s, many newborn screening tests were developed; state regulations governing the newborn screening programs were enacted. However, there was much variability in the newborn screening systems leading sometimes to social inequities.

## **Newborn Screening And Genetic Services Now**

Until 2005, expanded screening occurred slowly and variably due, at least in part, to the lack of a national newborn screening policy. Now, newborn screening across the States is largely uniform and is coordinated with input from the federal Secretary's Advisory Committee on Heritable Disorders in Newborns and Children.

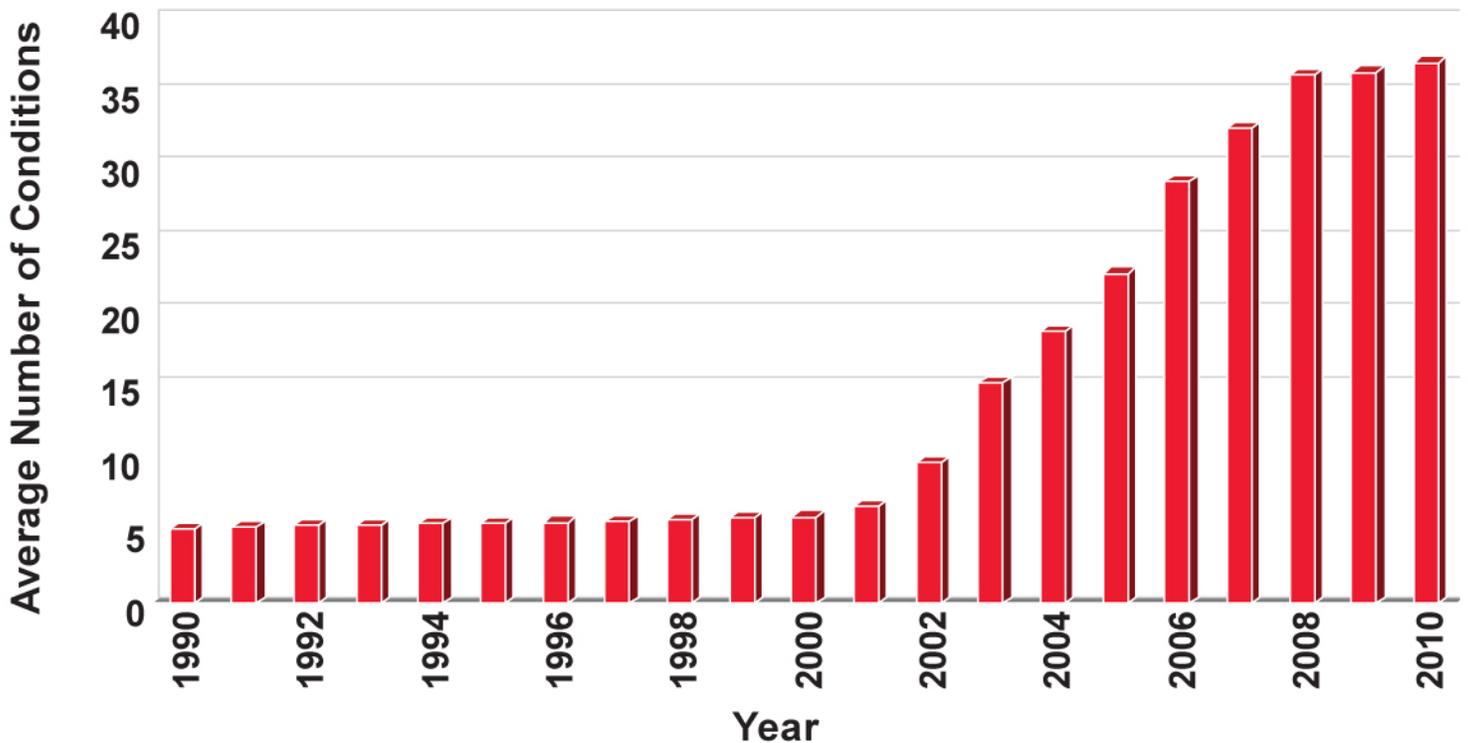
Genetic services and our understanding of genetic medicine have evolved rapidly in the past 20 years. Besides newborn screening, genetic services now include a broad range of clinical and public health activities such as individual and community health assessment; education and training; diagnosis; genetic screening, testing, and counseling for inherited conditions; measuring susceptibility and prevalence of gene-related conditions that result from specific environmental exposures; and helping individuals with genetic related conditions or risk factors in the management of their health.

What does the future bring for genetics and newborn screening? In 2003, the human genome was sequenced and today the cost for full sequencing is rapidly approaching the predicted \$1000 benchmark. Knowledge of genetics and newborn screening is increasing almost as rapidly as the technology and our understanding of single gene disorders has evolved to realize the impact of environment and other factors on those gene expressions. For instance, there are tests for various drug sensitivities, which in some cases can even predict response to cancer treatment based on genetic information. Clinicians are beginning to examine the genetic/biologic risk and protective factors and the relationship between those factors and their interactions with the environment that cause a given person to express a given condition at a given point in time in his or her developmental history. Health educators and health care and public health professionals need to seize the challenge to assist individuals and communities to utilize genetics information and services in ways that will be helpful to them and for the effective translation of these scientific advances into medicine and public health.

## Newborn Screening And Genetic Services History

1950	1960	1970	1980	1990	2000	2010
1930s-Diaper test for PKU	1953-Discovery of the "double helix"	1959-Identification of chromosomal cause of Down and Turner Klinefelters syndrome	1960s-NBS for PKU with filter paper	1970s-Sickle Cell Disease Act and National Genetic Disease Act	1970s-Thyroid testing sickle cell testing, CAH testing; technology for chromosomal banding and cytogenetic analysis	1986-NIH-HRSA Sickle Cell Disease Consensus conference: screening for SCD begins
1990s-NBS includes DNA from filter paper, application of tandem mass spectrometry (MS/MS) to allow simultaneous detection of multiple disorders, CF studies, infectious diseases	AAP Newborn Screening Task Force	2000-Newborn hearing screening initiated. Expansion of MS/MS screening. Children's Health Act authorization: Heritable Disorders Program	2003-Secretary's Advisory Committee on Heritable Disorders in Newborns and Children chartered	2008-Newborn Screening Saves Lives Act	2010-Endorsement of SACHDNC Recommended Screening Panel and addition of SCID to the panel	1987-1992-Congress earmarked funds in the 1987 to 1989 MCH Block Grants and federal MCH program to initiate federal and state partnerships to support newborn screening for SCD and other genetic disorders. MCH program and state infrastructure for genetic services were established.

Average Number of Newborn Screening Conditions Required in United States Programs, 1990-2010





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**Suggested Citation:**

Lloyd-Puryear, M and Therrell, B. *Newborn Screening Services: Then and Now*. A 75th Anniversary Publication. Health Resources and Services Administration, Maternal and Child Health Bureau. Rockville, Maryland: U.S. Department of Health and Human Services; 2010.

This publication is available online at <http://www.mchb.hrsa.gov/>

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