

## Screening and Public Health

Population-based screening is an important component of public health. Screening has the goal of identifying early disease (as in a non-palpable breast cancer detected by mammography) or a risk state (as in hypertension) in order to initiate treatment to reduce morbidity and mortality. Most current screening recommendations are implemented in outpatient settings, after a physician recommendation.

**Newborn screening.** An important exception to outpatient screening is newborn screening, which is implemented universally in all states through state-based public health programs. The blood draw for newborn screening usually occurs in the hospital shortly after birth, with testing and other aspects of the program (follow-up, education, management, and treatment) under state oversight. Most conditions tested for in newborn screening programs are genetic. All states provide universal newborn screening for phenylketonuria (see [OMIM#261600](#), [National PKU News](#), [GeneReview](#)), congenital hypothyroidism, and galactosemia ([OMIM#230400](#), [GeneReview](#)). All but one provide universal screening for sickle cell diseases ([OMIM# 603903](#)). Most states also screen for congenital adrenal hyperplasia ([GeneReview](#)), biotinidase deficiency ([GeneReview](#)), and maple syrup urine disease ([OMIM#248600](#), [OMIM#248611](#), [OMIM#248610](#)).

Newborn screening allows detection of diseases for which early treatment is necessary, available, and efficacious. For example, children with phenylketonuria (PKU) develop mental retardation on a normal diet. With a phenylalanine-restricted diet, they achieve normal intelligence.

Many additional newborn screening tests are currently under consideration or have been added to some state screening panels. These include rare biochemical disorders that are detectable with tandem mass spectrometry, congenital hearing loss ([Case 10](#)), and cystic fibrosis ([Case 12](#)). Some states are involved in research projects — for example, developing screening tools for such diseases as diabetes type 1 ([Case 19](#)), lysosomal storage diseases, and fragile X syndrome ([Case 16](#)).

Several Web resources provide additional information about newborn screening:

- [National Newborn Screening and Genetics Resource](#)

- [Newborn Screening Quality Assurance Program](#)
- [Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children](#)

**Screening in adults.** No genetic tests are currently recommended for adult population screening. Some experts have recommended screening for [hemochromatosis](#). However, the CDC currently recommends screening only for persons who have a close blood relative with hereditary hemochromatosis, or for persons experiencing otherwise unexplained symptoms compatible with hemochromatosis (these symptoms include severe weakness or fatigue; unexplained joint or abdominal pain; signs of liver disease, diabetes, or heart problems; impotence; infertility; and loss of menstrual periods). See [CDC recommendations](#).

**Genetics services.** Public health has an important role in assuring that needed health care services are provided. Many states help support genetic counseling services, to ensure their availability to the public. Federal support has played an important role in ensuring availability and quality of services.

Health Resources and Services Administration (HRSA) News Briefs:

- [Remarks: Integrating Genetics into Public Health Care Policy, Research, and Practice \(1999\)](#)
- [HRSA Report Calls for Expanded Genetics Education for Nurses \(2002\)](#)

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