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## Newborn Genetic Screening

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State newborn screening systems were the first and remain the largest genetic programs for children. Nationwide, state public health programs screen an estimated 4 million infants annually for genetic disorders. Each year approximately 3,000 babies with severe disorders are detected due to newborn screening programs. Undetected and untreated abnormalities can result in mental retardation, severe illness and premature death. Some genetic disorders occur rarely (e.g. maple syrup urine disease—once in 120,000 live births) while others are more common (e.g. congenital hypothyroidism—once in 4,500 births). Comprehensive state newborn screening programs involve testing, follow-up, diagnosis, treatment and evaluation.

**Testing.** As public health initiatives, newborn screening programs focus resources on treatable conditions that occur relatively frequently. Currently, tests are available for about 30 genetic or metabolic diseases, but most babies are not tested for all of these disorders because policies regarding testing vary from state to state. Advances in technology, particularly in genetics and metabolic research, will enable testing for numerous abnormalities.

**Costs.** Although funding newborn screening programs requires expenditures by the states, proactively treating congenital abnormalities may save states money by avoiding more financially burdensome medical costs and state institutional services. Most states fund screening programs by collecting fees for the tests. Fees range from \$15 in some states to \$50 in others. When fees for screening are charged, several vehicles exist for payment, including patient payment, private insurance, Medicaid and the State Children's Health Insurance Program. Some states, however, use public dollars or general funds instead.

### Federal Action

Federal legislation supporting screening for genetic diseases passed in 1976. More recently, the Children's Health Act of 2000 (Public Law 106-310) authorizes funds to establish, expand, or improve systems or programs for newborn and child genetic screening.

The Newborn Screening Task Force (convened by the American Academy of Pediatrics, with funding from the federal Maternal and Child Health Bureau) published recommendations in August 2000 concerning newborn screening. The report, *Newborn Screening: A Blueprint for the Future*, calls for states and the federal government to work together to create a national model standardizing the structure and function of newborn screening systems.

### State Action

Even though newborn screening became available to infants through state programs in the 1960s, and all states screen for some conditions, the extent of screening varies among the states. State experiences vary

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regarding laws or regulations, conditions tested, oversight responsibilities, state advisory boards, processes for informing parents, exemptions, storage policies and use of blood samples and payment for newborn screen procedures. Many state laws include exemptions for parents who object to genetic testing for religious reasons.

All states screen for at least two disorders—phenylketonuria (PKU) and congenital hypothyroidism. More than half the states screen for galactosemia, sickle cell disease or hearing loss. Massachusetts screens for almost 30 genetic conditions (some as part of a research study), while West Virginia requires screening for just four disorders—PKU, congenital hypothyroidism, galactosemia and sickle cell disease (only in selected populations). The other most common screening requirements include adrenal hyperplasia, biotinidase deficiency, branched-chain ketonuria, cystic fibrosis, homocystinuria and maple syrup urine disease.

**Issues for State Policymakers.** As newborn screening programs continue to evolve, state policymakers face a number of issues. Currently, no uniform national policy exists for the selection of newborn screening tests, although various organizations have released recommendations. For example, the March of Dimes recommends that programs use nine core tests plus newborn hearing screening: PKU, biotinidase deficiency, hypothyroidism, congenital adrenal hyperplasia, sickle cell, maple syrup urine disease, galactosemia, homocystinuria and medium-chain acyl-CoA dehydrogenase deficiency (MCAD). On the other hand, the American Academy of Pediatrics supports newborn genetic screening, but has not yet recommended a core number of tests.

Since no uniform national policy exists, great variations exist among state newborn screening systems. When formulating legislation regarding newborn screening programs, lawmakers must determine:

- Which disorders to screen—especially if the diseases are very rare or untreatable;
- Whether to integrate the program with other infant screening and public health programs in order to foster coordination and prevent duplication;
- Whether to provide follow-up services and treatment, or ensure that treatment is accessible;
- How to include public health laboratories as part of a system of detection, intervention and follow-up, but still ensure confidentiality and regulation, or how to maintain quality if private labs do the testing.

## Selected References

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