



## FROM BIRTH TO DEATH AND BENCH TO CLINIC

# THE HASTINGS CENTER BIOETHICS BRIEFING BOOK

for Journalists, Policymakers, and Campaigns

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### CHAPTER 27

## Newborn Screening

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# newborn screening

by Mary Ann Baily

## Framing the Issue

State newborn screening programs test nearly all infants born in the United States for selected inherited and congenital conditions that may cause disability or death. Screening is mandatory in all but a few states. In addition to screening, the programs provide education, follow-up to definitive diagnosis, and long-term treatment and management, or link affected infants and parents to these services.

The programs began in the 1960s after a simple blood test was developed for a genetic metabolic disorder called phenylketonuria (PKU). If infants with PKU are identified soon after birth and immediately put on a special diet, they can be protected from permanent mental retardation and other health problems. States began screening newborns for PKU and, over time, added other disorders in response to advocacy by parents of affected children, health professionals, and organizations concerned with child health. As individual state programs evolved, they came to vary considerably from each other in the conditions screened for, the services provided, and the modes of financing.

Recently, a dramatic expansion of state newborn screening has been underway. The expansion is a response to new medical knowledge, new testing technologies, and political pressure by advocacy groups; however, its form and timing have been heavily influenced by the work of a federally funded expert group convened by the American College of Medical Genetics (ACMG). The group's charge was to develop a uniform test panel that could be adopted by all states. Before the work began, the majority of states screened for fewer than 10 conditions. The ACMG group's report recommended screening for 29 core conditions and another 25 conditions that could be detected incidentally in screening for the core group. By May 2008, most states had started screening for all or nearly all of the core conditions (see box, "Core Conditions").

This expansion has been accompanied by ethical debates related to cost, evidence, and parental rights. While supporters believe that the expanded screening is important for children's health, critics charge that the ACMG group made its recommendations without a solid evidence base or adequate consideration of competing demands on public resources. Critics also object to the group's movement away from newborn screening's traditional reliance on significant benefit to the newborn from very early diagnosis and treatment as the criterion for adding a condition to a state screening panel. They argue that when this criterion is not met, there is no justification for omitting parental informed

*Mary Ann Baily, PhD, is a research scholar at The Hastings Center.*

## HIGHLIGHTS

- Newborn screening programs test nearly all infants born in this country for selected inherited and congenital conditions that can cause disability or death.
- These state programs began in the 1960s with a blood test for phenylketonuria (PKU), a metabolic disorder that can cause permanent mental retardation and other problems unless it is diagnosed and treated early.
- A dramatic expansion of newborn screening programs is under way, with most states testing for about 29 core conditions, up from fewer than 10 several years ago.
- The expansion of newborn screening raises ethical controversies about its cost, evidence of its efficacy, and parental informed consent.
- Policymakers have an ethical obligation to weigh benefits against costs when directing resources to newborn screening.
- Although state governments create and manage screening programs, the federal government supports them with funding, help with improving program performance, and other kinds of assistance.
- The Newborn Screening Saves Lives Act, signed into law in 2008, will help support state efforts to coordinate and improve their programs.

## EXPERTS TO CONTACT

**Mary Ann Baily, PhD**, *Research Scholar, The Hastings Center* • [bailym@thehastingscenter.org](mailto:bailym@thehastingscenter.org), 845-424-4040, x 200

**Jeffrey R. Botkin, MD, MPH**, *Professor of Pediatrics, Division of Medical Ethics, University of Utah School of Medicine* • [jeffrey.botkin@hsc.utah.edu](mailto:jeffrey.botkin@hsc.utah.edu), 801-581-7170

**Ellen Wright Clayton, JD, MD**, *Codirector, Center for Biomedical Ethics and Society, Vanderbilt University School of Law* • [ellen.w.clayton@vanderbilt.edu](mailto:ellen.w.clayton@vanderbilt.edu), 615-322-1338

consent, and no obvious reason to give screening and treatment for a condition priority over other ethically important health care not readily available to all children, such as diagnosis and treatment of asthma or juvenile diabetes.

## The Ethical Controversies

The debates about the expansion of newborn screening programs concentrate on three key issues: cost, evidence, and parental consent.

**Cost.** Concern about cost is often seen as opposed to ethics in debates about newborn screening policy. Many advocates say that it is wrong to consider cost at all when infants' lives are at stake. In contrast, ethicists say that it is wrong to ignore cost. Cost is an ethical issue because newborn screening uses collective resources (public and private) to pay for the screening, as well as the follow-up and treatment. Although screening newborns may be desirable, there are always other uses for resources that would also save lives and prevent disability—there is an opportunity cost to their use. Policymakers thus have an ethical obligation of stewardship to weigh the benefits against the costs when directing resources to newborn screening.

In stewardship, the total net benefit from screening isn't the only consideration; the fairness of the distribution of benefits and costs also matters. On the benefit side, fairness often comes up in discussions of state differences in test panels. To many people, it seems unfair that for an infant with a genetic disorder, being born in one state rather than another can be a matter of life or death. A major goal of the ACMG group was to make access to screening less variable from state to state. The variation in the quantity and quality of follow-up services received by affected children and their families is also a fairness issue. On the cost side, it is important to recognize that cost isn't just the screening test but also the entire cost of the program. There are also time and anxiety costs to families of newborns with "false positive" tests (infants who test positive but are shown by follow-up testing to be healthy). Currently, most people involved in newborn screening would agree that the distribution of these costs is arbitrary and does not conform to any reasonable standard of fairness. It imposes excessive burdens on some families and fails to distribute the total cost of the system equitably across the entire nation.

**Evidence.** If cost is an ethical issue, then evi-

dence is also an ethical issue. One cannot assess the opportunity cost of resources and the distributional fairness associated with a policy decision without detailed information about the decision's positive and negative effects. Some advocates are uncomfortable with calls for evidence-based newborn screening policy, however. They argue that it sets too high a bar, given that conditions considered for screening are relatively rare and, in the absence of screening, may be underdiagnosed.

Since gathering evidence itself uses resources, everyone understands that policy must often be made without complete information. The debate is about how much information is enough. Strong advocates for screening are likely to say that screening should go ahead without any hard evidence if there is some hope of benefit. Others argue that ethics requires a cost-conscious, systematic effort before the introduction of a mandatory new test to gather and evaluate evidence on the consequences of the decision. There is less controversy over what should happen after testing is introduced: most consider cost-conscious, systematic collection and evaluation of evidence on the effects of newborn screening to be an essential part of the ongoing management of these public programs.

**Parental consent.** The United States has sturdy societal values respecting the rights of individuals to decide what treatments they will have, whether they will participate in research, and what can be done with their personal information and their bodily tissues, including blood samples. (Because only a fraction of each blood sample taken for newborn screening is used in the screening, the remainder is a valuable potential resource for research and program evaluation.) Since parents are normally considered the appropriate people to make decisions on behalf of their children, parental informed consent is ethically required for the medical treatment of children and for the involvement of children in research.

Because parental consent is the ethical standard, the mandatory status of public newborn screening has always been controversial. Mandatory screening for PKU was originally sought on the grounds that the urgent need for early diagnosis and the great benefit of the treatment justified omitting parental informed consent. This criterion guided newborn screening programs for many years (although some ethicists maintained that states should obtain parental consent even in these cir-

cumstances). The ACMG report argued that it was appropriate to depart from this criterion and consider benefits to the family or society, rather than to the infant. For example, early diagnosis of an untreatable genetic condition may allow parents to plan ahead for the time when the child's symptoms appear and perhaps to alter their reproductive decisions to avoid the birth of another affected child. Early identification of affected children can also benefit research on the condition by providing potential human research subjects and residual blood samples. When the traditional criterion is modified in these ways, however, parental informed consent to screening is the usual ethical standard.

## Looking to the Future: Federal and State Policy

The ethical issues above are likely to persist in the debates over newborn screening policy. The policy action is at both the state and federal levels. Although state governments create and manage the programs, the federal government has played a supportive role in newborn screening from the beginning. This role includes funding research on newborn screening, promoting the development of policies and guidelines, and partnering with states to improve their programs' performance. The federal government also supports newborn screening indirectly through Medicaid, a medical assistance program for certain categories of the poor that is financed with federal and state funds. Many state newborn screening programs charge fees to third party payers for testing and diagnostic services, and Medicaid is the third party payer for about one-third of babies born in the United States.

The federal Health Resources and Services Administration (HRSA) funded the ACMG expert group's work. Through the Advisory Committee on Heritable Disorders in Newborns and Children, which advises the Secretary of Health and Human Services, HRSA and other federal agencies are now working with states as they implement the recommendations. Many states already had difficulty providing adequate follow-up services for the conditions in their existing test panels, and for all states, expanding programs to cover so many conditions is a major managerial and financing challenge. The Newborn Screening Saves Lives Act, signed into law in April 2008, will help support state efforts to coordinate and improve their programs.

## CORE CONDITIONS

### Blood disorders

- Sickle cell anemia
- S-beta thalassemia
- Sickle-C disease

### Amino acid disorders

- Phenylketonuria
- Maple syrup urine disease
- Homocystinuria
- Citrullinemia
- Argininosuccinic acidemia
- Tyrosinemia type I

### Fatty acid disorders

- Medium-chain acyl-CoA dehydrogenase deficiency
- Very long-chain acyl-CoA dehydrogenase deficiency
- Long-chain L-3-OH acyl-CoA dehydrogenase deficiency
- Trifunctional protein deficiency
- Carnitine uptake defect

### Organic acid disorders

- Isovaleric acidemia
- Glutaric acidemia type I
- Hydroxymethylglutaric aciduria (also called HMG-CoA lyase deficiency or 3-OH 3-CH<sub>3</sub> glutaric aciduria)
- Multiple carboxylase deficiency
- Methylmalonic acidemia due to mutase deficiency
- 3-Methylcrotonyl-CoA carboxylase deficiency
- Methylmalonic acidemia cblA and cblB forms
- Propionic acidemia
- Beta-Ketothiolase deficiency

### Other conditions

- Congenital hypothyroidism
- Biotinidase deficiency
- Congenital adrenal hyperplasia
- Hearing loss
- Cystic fibrosis
- Classical galactosemia

Going forward, the debate about the criteria to be used and the evidence required for adding new conditions to the nationally recommended newborn screening panel will continue. It is important to reach a societal consensus on an appropriate process. The Advisory Committee on Heritable Disorders has taken responsibility for overseeing the development and implementation of such a process. It would seem desirable to have the process include an evidence review that follows

## RESOURCES

### Web sites

- <http://genes-r-us.uthscsa.edu> – The National Newborn Screening and Genetics Research Center. Includes fact sheets, a clickable map of state newborn screening programs, an up-to-date list of conditions screened for in the United States, and literature for parents and providers.
- [www.nlm.nih.gov/medlineplus](http://www.nlm.nih.gov/medlineplus) – Medline Plus, a service of the National Library of Medicine and the National Institutes of Health. Includes a gateway page under health topics for newborn screening that features an overview and tutorial, links to latest news and journal articles, research, and information on specific conditions.
- [www.aap.org](http://www.aap.org) – American Association of Pediatrics. Includes a gateway page under health topics for newborn screening that features overview articles, fact sheets and policy guidelines, information on specific conditions, and links.
- [www.ncsl.org](http://www.ncsl.org) – National Conference of State Legislatures. Includes a page on newborn genetic and metabolic screening with links to newborn screening legislation in all fifty states.
- [www.hrsa.gov/heritabledisorderscommittee](http://www.hrsa.gov/heritabledisorderscommittee) – the Advisory Committee on Heritable Disorders in Newborns and Children. Includes reports and correspondence, governance, and educational materials.

### Recent news

- Shirley S. Wang, “Addressing the Fallout of Newborn Screening,” *Wall Street Journal*, October 30, 2007.
- Jamie Talan, “Early Action Proving Crucial to Hearing

Success,” *New York Times*, September 4, 2007.

- Erin Cline, “Infant Screening Rates Rise in U.S.,” *Los Angeles Times*, June 11, 2006.
- Darshak Sanghavi, “Newborn Screening Benefits Not So Clear,” *Boston Globe*, October 10, 2005.

### Further reading

- Mary Ann Baily and Thomas H. Murray, “Ethics, Evidence, and Cost in Newborn Screening,” *Hastings Center Report*, May-June 2008.
- Virginia A. Moyer et al., “Expanding Newborn Screening: Process, Policy, and Priorities,” *Hastings Center Report*, May-June 2008.
- Bruce K. Lin and Alan R. Fleischman, “Screening and Caring for Children with Rare Disorders,” *Hastings Center Report*, May-June 2008.
- “Newborn Screening: Toward a Uniform Screening Panel and System,” eds. Michael S. Watson et al., *Genetics in Medicine* supplement, May 2006.
- Scott D. Gross et al., “From Public Health Emergency to Public Health Service: The Implications of Evolving Criteria for Newborn Screening Panels,” *Pediatrics*, March 2006.
- Jeffrey R. Botkin, “Research for Newborn Screening: Developing a National Framework,” *Pediatrics*, October 2005.
- American Academy of Pediatrics Newborn Screening Task Force, “Serving the Family from Birth to the Medical Home,” *Pediatrics* supplement, August 2000.

accepted scientific standards and is done (or is thoroughly reviewed by) people outside the field of newborn screening.

Debate will also continue over the extent to which priority should be given to newborn screening given the constraints on the resources available for children's health. In today's health care system, there is no institutional structure that can force consideration of opportunity cost and no way to ensure responsible stewardship. Without stewardship, special interest groups such as health professional organizations, consumer groups, and makers of screening technologies can exercise inappropriate influence on policy. Many advocates provide important perspectives and are committed to the well-being of affected children; however, in the absence of a structure that forces comparison of newborn screening with other uses of public and private resources, it is difficult to make cost-conscious, evidence-based, fair decisions that balance the needs of all children. This situation is unlikely

to change without meaningful reform of American health care.

The issue of parental consent will remain controversial. Many ethicists will continue to hold that programs must maintain clear benefit to the infant as the essential criterion for mandatory public newborn screening; if the benefit is to anyone else, or if the benefit to the infant is uncertain, parental informed consent is required. In the meantime, the expansion of the test panels has made the task of informing parents significantly more complex.

Finally, there is an urgent need to clarify the ethical requirements with respect to parental consent for using leftover blood spots for newborn screening quality improvement, research related to newborn screening, and research on questions not directly related to newborn screening. The use of newborn screening blood spots is simply a specific instance of the larger issue of achieving a societal consensus on the ethical rules that should govern the use of bodily tissues for social purposes. 