

Ethics, Evidence, and Cost in NEWBORN SCREENING

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When deciding what disorders to screen newborns for, we should be guided by evidence of real effectiveness, take opportunity cost into account, distribute costs and benefits fairly, and respect human rights. Current newborn screening policy does not meet these requirements.

In 2000, a child named Ben Haygood died in rural Mississippi from a rare, inherited, undiagnosed metabolic disorder known as medium chain acyl-coenzyme A dehydrogenase deficiency, or MCADD. Children with MCADD seem healthy, but if they go without food for too long, they can suddenly become seriously ill, and they may even die. Children diagnosed with MCADD must avoid prolonged fasting. If Ben had been tested for MCADD as a newborn and his parents had known to take this simple precaution, perhaps his life could have been saved.

In the United States, state-based public health programs screen all infants shortly after birth for selected conditions that can have serious health consequences if not identified and treated very early in life. After Ben died, his father became a passionate

advocate for expanding Mississippi's newborn screening program to add MCADD and other disorders. Within a few years, the Mississippi legislature had passed the Ben Haygood Comprehensive Newborn Screening Act, the state's test panel had gone from only five disorders to forty, and a three-person team had been created in each of nine state districts to manage the cases of children with abnormal results.¹ Most newborn screening program funding came from a fee for each newborn screened; to help pay for the expansion, the state doubled the fee to seventy dollars. This meant that a substantial share of the resources for expansion came from Mississippi's Medicaid funds, since Medicaid covers more than half of Mississippi births.² In the first year of expanded screening, three cases of MCADD were identified along with twelve cases of other new disorders, out of a total of 116 newborn screening diagnoses.³

Around the same time, according to a *New York Times* article, Mississippi experienced a worrying change in overall infant mortality.⁴ The state's rate

Mary Ann Baily and Thomas H. Murray, "Ethics, Evidence, and Cost in Newborn Screening," *Hastings Center Report* 38, no. 3 (2008): 23-31.

had long been above the national average but had recently been falling. Between 2004 and 2005, however, it increased, especially among blacks, and in 2005, 481 infants died, sixty-five more than in the previous year.

A new governor had taken office in 2004 with a promise to keep taxes steady and bring Medicaid costs down. Medicaid eligibility requirements were tightened, and some programs were cut. Were the changes in Medicaid a factor in the increase in infant deaths? Could infant lives have been saved if the state had increased the availability of Medicaid services and provided state funds to subsidize transportation for low-income rural black women so they could access prenatal care more easily? It is hard to know; however, the *Times* article points to the dramatically lower infant mortality rate achieved in one very poor Mississippi county from 1991 to 2005.⁵ The county's rate fell sharply after a private charity began providing intensive in-home visits using local women as counselors and busing pregnant black women to pre- and postnatal classes.

Our goal here is not to single out Mississippi for criticism or to focus on a specific pair of alternatives for improving the health of children. Newborn screening, home visits, and prenatal care are all means to the end of helping children. There are many others as well. There could be a systematic effort to identify children with asthma and manage the condition better, especially in poor children, who often end up in emergency rooms in asthmatic crises. There could be efforts to reduce smoking by pregnant women, increase car seat use, or prevent childhood accidents. We do not know which of these programs would produce the greatest benefits for children—but that ignorance is itself a major problem. The problem is heightened when resources available for children's health, such as Medicaid, are decreased or fail to keep up with growing need.

The experience in Mississippi serves to highlight an important ethi-

cal issue in child health policy. Although resources for children's health are scarce, too often there is no systematic effort to identify and compare alternative ways to use those precious resources to help children; moreover, the information needed to make an intelligent and informed comparison among such programs is often lacking.

In this article, we explore this and other ethical and policy issues that arise in debates about public newborn screening programs. Our analysis is based on a Hastings Center project funded by the National Human Genome Research Institute, and it has been shaped by the deliberations of the people with varied disciplinary backgrounds and practical newborn screening experiences who participated in the project.⁶ This article is not a consensus report from our project, however. The period during which the project ran—2002 to 2007—was a time of unexpectedly rapid and often controversial change in newborn screening. Participants were able to reach agreement on the general requirements for ethical newborn screening policy, but they disagreed strongly about the extent to which developments in newborn screening conformed to the requirements. The opinions and recommendations expressed here are therefore our own.

An Overview of Newborn Screening

Newborn screening began in the 1960s after Dr. Robert Guthrie developed a simple blood test for phenylketonuria. PKU is a genetic metabolic disorder that leads to mental retardation and other symptoms; the treatment is a special dietary regime that, if begun early in life, before any symptoms occur, can reduce or eliminate the major symptoms associated with the condition. To screen newborns for PKU, a small blood sample is taken from each newborn's heel, deposited as spots on a special filter paper card, and transported to a testing facility. Children who test pos-

itive receive further diagnostic services to identify those who actually have the disorder ("true positives") and refer them for treatment.

State newborn screening began with a voluntary PKU testing program in one state. Over time, other states introduced PKU testing and made it mandatory, partly in response to intensive grass-roots lobbying by children's advocates. Some states also began expanding newborn screening to include tests for other inherited and congenital disorders. The federal government has made substantial contributions to the development of newborn screening, but the actual testing remains primarily a state public health activity.

Today, all states have newborn screening systems that provide initial screening and follow-up services, which may include diagnostic services, short- and long-term treatment and management, parent education, and program evaluation. Since state governments make the decisions about program structure and content, the systems vary along many dimensions. For example, all fifty states and the District of Columbia test for PKU, sickle cell disease, congenital hypothyroidism, and galactosemia, but they vary in the selection of additional conditions. States have also made different decisions about the content of treatment protocols, the services available for follow-up, and the extent to which the cost of the system falls on the families of the newborns screened. All but a few states make screening mandatory and do not obtain parental informed consent. A number of these states give parents some freedom to opt out, but that freedom is rarely exercised: many parents do not even realize that they have it.

The heel-stick blood sample is used to screen for all newborn screening program conditions except hearing impairment. In the past, when a program added a new condition, it added a new laboratory blood test. In the 1990s, a new screening technology, tandem mass spectrometry, be-

came available. It can test for PKU, MCADD, and a number of other disorders simultaneously. Tandem mass spectrometry measures the levels of various metabolites in the blood, and abnormalities in the levels suggest the presence of metabolic disorders. Advocates have pressed states to invest in the new technology; a federally funded expert group has recommended that all states adopt a uniform list of conditions, including many disorders detectable with the new technology; and the uniform list has been endorsed by key public and private entities involved in newborn screening. A major expansion of state newborn screening programs is now under way.

Another new technology is visible on the horizon: screening for genetic disorders using DNA-based “microchips.” Chip technology will allow newborns to be screened directly, simultaneously, and at relatively low cost for many disorders, likely leading to further expansion of newborn screening.

Requirements for Ethical Newborn Screening Policy

The overall conclusion of the Hastings Center project is that newborn screening policy is ethically acceptable when it is evidence-based, takes into account the opportunity cost of the newborn screening program, distributes the costs and benefits of the program fairly, and respects human rights. Many would agree that these are sensible requirements for prudent public policy, but some may not immediately see all of them as *ethical* requirements. In particular, the ethical dimensions of the clauses about opportunity cost and evidence may not be clear. In public debates about newborn screening, concern about cost is often seen as opposed to ethics, and some advocates assert that when infant lives are at stake, considering cost at all is morally wrong.⁷

In fact, cost is an ethical issue in newborn screening policy because, as the experience in Mississippi suggests, the collective resources used for

screening programs could always be used in other ways to improve the length and quality of human lives. The resources used for newborn screening have an opportunity cost, and policy-makers have an ethical obligation of *stewardship* to take that into account when they make resource allocation decisions. Policy-makers also have an ethical obligation to ensure that the costs and benefits of their allocation decisions are fairly distributed across individuals.

Evidence is an ethical issue because opportunity cost and distributional fairness are ethical issues. Information about the existence, the reliability, the size, and the distribution of the bene-

propriate people to make decisions on behalf of their children.

Given these values, the mandatory status of public newborn screening has always been ethically controversial. Mandatory screening for PKU was originally sought on grounds that the urgent need for early diagnosis and the great benefit of the treatment justified omitting informed consent. If mandatory screening requires this kind of justification, then a new condition should be added to the mandatory panel only when there is an established screening test and good evidence that the condition causes serious harm and that the harm can be avoided if the infant is diagnosed and

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fits and costs of screening is critical in evaluating the opportunity cost of devoting scarce resources to it and the extent to which the costs and benefits are fairly distributed. Since gathering evidence consumes resources in itself, policy decisions must often be based on incomplete information. Nevertheless, policy-makers have an ethical obligation to use all available evidence and to support systematic, cost-conscious, ongoing efforts to develop additional evidence where necessary.

Evidence and cost are also factors in developing newborn screening policies that respect human rights. The United States has sturdy societal values supporting 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. In these matters, parents are normally considered the ap-

propriate people to make decisions on behalf of their children. To develop the evidence base for ethical policy decisions, research and data-guided quality improvement activities are essential; however, these activities must be designed to respect individual rights related to participation in research and use of blood spots.

Even when screening is mandated, parents deserve to receive some information about the screening. To determine how much, the cost of the information process to the program and to parents should be weighed against the value of being informed. Newborn screening programs should maintain the confidentiality of personal health information in program evaluation and research, which may require weighing the value of privacy protection against the cost of security measures and the benefits foregone if security measures make a research or evaluation activity impracticable.

Finally, if society is ethically required to engage in some activity no matter what it costs, still, the necessary resources must come from somewhere. Since resources ultimately come from people, not from an abstract entity called “society,” policy-makers must always have an ethical justification for the pattern of individual sacrifice that results when resources are devoted to meeting societal ethical obligations.

Assessment of Current Policies

Does the newborn screening policy process produce policies that meet these ethical requirements? Unfortunately, the answer is no—not in the past, and not now. To be fair to newborn screening, this is part of the larger disorder and confusion in health policy in general. Below, we take each of the four clauses in the project conclusion and discuss them with reference to what actually happens in current newborn screening programs.

Newborn screening policy should be evidence-based. Is newborn screening policy based on solid evidence on the nature, size, and distribution of the benefits and costs of newborn screening? Obviously, the information requirements for ethical policy are formidable. For each candidate condition, detailed evidence on the disorder’s natural history, its incidence, and the variation in its incidence and expression within the population would be desirable. There should be evidence on the scientific validity, clinical utility, and resource cost of the screening test. There should also be evidence on the effectiveness, resource cost, and availability of treatment. Finally, the positive and negative effects of introducing newborn screening must be measured, aggregated, and compared. This requires evidence on personal values and preferences and how they vary across individuals. Collecting and carefully evaluating all this information is a major challenge.

Currently, the challenge is not being met. The information is far from complete even for conditions that have been included in newborn screening panels for years. Yet instead of addressing the significant gaps in knowledge about conditions already on the screening panels, states are adding new conditions, many of them only poorly understood.

The nature and extent of the expansion is somewhat unexpected. In a comprehensive report published in 2000, the federally funded Newborn Screening Task Force emphasized the importance of using an evidence-based approach in decisions about the introduction of new tests, and it recommended introducing tests on a pilot basis when evidence was limited. In 2002, the American College of Medical Genetics convened a working group (funded by a federal agency, the Health Resources and Services Administration) to evaluate the available evidence on a long list of conditions proposed for newborn screening and to make evidence-based recommendations for a uniform panel of conditions that were suitable for inclusion in state newborn screening programs.⁸

In its final report, the group recommended a uniform panel of twenty-nine primary disorders and an additional twenty-five secondary disorders that would be detected incidentally while screening for the primary disorders. The Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (the principal national body concerned with newborn screening since June 2004) immediately endorsed the ACMG report in a statement to the Secretary of Health and Human Services and called for state newborn screening programs to conform to the report’s recommendations. Other organizations, including the March of Dimes and the American Academy of Pediatrics, also endorsed the recommendations.

The ACMG report acknowledges that there are serious limits to the information available on many of the

conditions it recommends for screening. Unfortunately, the information shortfall may be worse even than acknowledged. As discussed in detail in the accompanying paper by Virginia Moyer and colleagues, as well as in other published work, the ACMG’s methodology, content, and working process appear to be deeply flawed.⁹ Critics have argued that the methodology used to develop the recommended panel was highly idiosyncratic and did not conform to established standards for evidence-based reviews. They have also noted that the working group had extensive expertise in metabolic genetics and laboratory medicine but lacked essential expertise in evidence-based medicine, bioethics, primary care, and health economics.¹⁰

The Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children has responded to criticism of the ACMG methodology by obtaining expert advice on accepted standards and methods for evidence-based medicine. It is developing a new and more rigorous evidence-based process for deciding whether to add new conditions to the uniform panel. The effort to improve a seriously flawed process is commendable; however, the process is unlikely to be in place and ready for evaluating new conditions in the very near future. For now, the committee might be better advised simply to urge states to deal adequately with the management issues in their current programs. Moreover, before evaluating additional conditions, the conditions in the current recommended panel should be reevaluated.

In the committee’s discussion of the evaluation process, some members of the committee suggested that the criteria used by the United States Preventive Services Task Force are too strict for newborn screening programs.¹¹ The USPSTF reviews a broad array of health services, including screening tests, to recommend whether they are appropriate for routine use in primary care practice, subject to patient consent. The ACMG is

recommending tests for use on every newborn in the country, without full informed consent, and at the expense of public and private third-party payers and individual families who will incur new costs without consultation. Surely, in such circumstances, the standard of evidence should be higher, not lower.

Newborn screening policy should take opportunity cost into account. Could greater benefits or a fairer distribution of benefits be achieved by reallocating newborn screening resources to another use? In answering this question, all costs should be considered, and costs are often understated in newborn screening policy debates. A common error is to consider only the cost of the individual test. Advocates may say: “If Baby A had only had a fifty-dollar screening test for MCADD, his life could have been saved. Surely a child’s life is worth fifty dollars!” That would indeed be a small price to pay to save an infant’s life. But newborn screening programs must test many newborns in order to identify the few with MCADD. Thus, even if only the cost of testing itself is counted—ignoring for a moment many other activities that must accompany the testing—saving one life costs much more than the price of a single test.

Cost is also understated when advocates claim that the cost of testing all newborns for an additional condition is low because the blood sample is already being collected and the infrastructure is in place. For example, if tandem mass spectrometry is already used for PKU and MCADD, then adding one more metabolic disorder seems to add only a little to the cost. The real cost is more than the cost of testing, however. After we factor in the full costs of parental education, follow-up of all positives to a definitive diagnosis, treatment of affected children, and ongoing data collection and evaluation, adding a new disorder to an existing panel can be very expensive. Moreover, if the natural history of the condition is poorly understood and plainly effective treatments

are lacking, children may receive no benefit, or may even be harmed by unnecessary interventions.

Another way to understate the cost of newborn screening is to count only the net cost of state budget appropriations earmarked for the program. In fact, adding a test to a mandatory newborn screening panel automatically imposes costs on private insurance (which is expected to pay for the screening test, follow-up, and treatment for insured infants) and on both the state and the federal government (which through the Medicaid program cover about a third of births in

underestimates the total cost of newborn screening.

Once the full costs of newborn screening are understood, the benefits must be assessed and compared to the benefits that could be achieved from other ways of using the resources. The framework for equitable allocation of health care resources we used in our project starts from the premise that society has a moral obligation to ensure that every child has access to adequate health care and to distribute the cost of achieving this outcome fairly. The adequate level of care should be determined by considering

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the United States). Other costs fall directly on families. These include the cost in time and money that families of children who test positive must bear to obtain a definitive diagnosis, and the unnecessary worry and anxiety experienced by families whose children turn out either not to have the disorder or to have a clinically insignificant form of it.

Finally, the full cost of newborn screening includes the cost of program-related research and quality improvement. The ACMG report concludes that the development and evaluation of evidence before and after introducing a test is an essential part of a national screening system, and it makes recommendations for incorporating ongoing research and quality improvement activities into newborn screening programs. It does not attempt to estimate the cost of all this work, however, and thus inevitably

the relative merits of different health services in the light of the reasons for the special importance of health care. This means that the resources devoted to newborn screening and treatment for genetic disorders should be established in the context of determining the entire adequate level of health care and the importance of health care relative to other important social goods.

Unfortunately, the policy process is biased against doing this. The American health care system is not really a *system*. It has no institutional structure to take responsibility for stewardship of collective resources and force consideration of opportunity costs of decisions about public health programs or additions to standard clinical care. In newborn screening, this system-level problem has been made worse because each state makes its own decisions on newborn screening; furthermore, program financing is plagued by a lack of transparency. Advocacy

by health professional groups, makers of screening technologies, and consumer groups such as the March of Dimes and associations supporting parents of children with genetic conditions also affects policy development. These advocates provide important perspectives, but often no one steps up to advocate for the programs that will not be undertaken and the people who will not be helped because health care resources have been directed elsewhere. Think again of Mississippi: a state expands newborn screening at the same time that it cuts support for prenatal care for poor women. The advocates for those women, and the children they were carrying, were either silent or ineffective.

Newborn screening policy should distribute the costs and benefits of the program fairly. Would changes to newborn screening policies produce a more fair distribution of benefits and costs within the programs? The comprehensive Newborn Screening Task Force report, advocacy groups, and other observers have identified many longstanding fairness issues associated with program structure. These relate primarily to the selection and implementation of tests and the financing and delivery of screening, follow-up, diagnosis, and services for treatment and management.¹²

The variation in the composition of test panels across states means that being born on one side of a state border instead of the other can mean life or death for a child with a genetic disorder. The incidence of a condition may vary across ethnic groups, and fairness is an issue in decisions about how this variation should influence the selection of a test and whether only the members of specific ethnic groups should be screened. Technical decisions made within newborn screening laboratories can also have implications for fairness; for example, a laboratory's decisions about the cut-off level that constitutes a positive test result for a disorder can affect different ethnic groups differently. The same is true for decisions about what

mutations to include in DNA-based screening.

Currently, the services received by individual families of affected children vary substantially and inequitably both across and within states. Also, the cost of the various elements of newborn screening programs is arbitrarily distributed, largely because of the patchwork nature of health care financing in America. This creates excessive burdens for some families and fails to distribute the burden of the total cost of newborn screening, including follow-up diagnosis and care, equitably across the entire nation.¹³

As noted, the traditional public health justification for newborn screening was that a very important benefit to the child would be lost if screening did not occur soon after birth, and that the risk to the child was minimal at worst. One of the most surprising features of the ACMG report is its departure from this basic principle, which has guided newborn screening from the beginning and was reaffirmed by the highly respected Newborn Screening Task Force only a few years before the ACMG began its work.¹⁴ The ACMG's expanded framework for justifying screening allows consideration of benefits to the family and to society. For example, benefits to the family might include the provision of information that could help the family make future reproductive decisions or avoid the so-called diagnostic odyssey (the lengthy pursuit of an explanation for a child's persistent ill health). Benefits to society might include the identification of potential research subjects for the study of currently untreatable disorders. Some newborn screening advocates now argue that the decision is not what to include in screening panels, but what to *exclude*; in this view, the default position is to screen newborns for everything possible.

Both the change and the way it came about are troubling. The ACMG working group adopted the new criteria with little discussion or

justification and immediately began using them to select the new uniform panel, which was then released as a *fait accompli*. As a result, the uniform panel includes conditions that do not urgently need treatment in the newborn period, or for which no proven treatment is available, or for which the benefit of treatment is much less significant and certain than the benefit of treatment for a condition like PKU. This means that the inequity of the difference in access across states has been exchanged for another kind of inequity. For some of the new conditions, it is less obvious that newborn screening for the condition is truly part of an adequate level of care, or if it is, that it should take priority over other ethically urgent health care not readily available to all children at this time.

Moreover, it seems imprudent as well as unfair to expand quickly, without the necessary support services in place for the new disorders and without first addressing the inequities in access to services for the conditions already included in screening panels. The work involved in expanding programs to deal with the long list of conditions in the uniform panel is monumental. It includes educating health care providers and parents about the conditions, resolving many technical issues associated with the new tests, building the infrastructure for follow-up to diagnosis and long-term treatment and management, and providing for ongoing research and quality improvement. HRSA is making a significant investment in helping states to do this work, but much more remains to be done. A child harmed because she was screened for a condition but the test failed to detect it, or because there was no proper follow-up and treatment, is no better off than a child who is harmed because screening for the disorder was never initiated.

The ACMG report's family benefit justification for screening also raises fairness questions. For example, although some families might benefit from an early diagnosis that lets them

avoid a diagnostic odyssey, that benefit should be weighed against the burdens of different kinds of odysseys that other families might have to endure. Families whose healthy children are inaccurately identified as having a condition must endure a period of anxious searching and wandering until new tests can reassure them that their children are well. Less fortunate families may experience more disturbing long-term outcomes. Suppose parents are told that testing confirms that their child has an abnormal laboratory finding associated with serious illness in some children, but as it turns out, their child never becomes symptomatic. Perhaps the child has a mild or subclinical form that was unknown before newborns were routinely screened for the disorder. (This very plausible scenario underscores the need for evidence on the natural history of disorders recommended for newborn screening.) Meanwhile, the family reorganizes its life around medical monitoring and planning for something terrible that never happens. Or a family may be told that a child has a genuinely serious disorder for which there is no proven treatment. The family begins a treatment odyssey—searching the Internet, visiting specialists, running up debt, medicalizing the child's life—only to have that life end in early death anyway. Or perhaps treatment options exist, but they are terribly expensive and burdensome—perhaps to the child as well as to the parents—and bring at best a slight, fleeting improvement in the child's condition. A family with limited resources and no comprehensive health insurance may be forced to choose between seeking a way to gain access to the services and making the painful decision to forego them. With programs expanding to include more conditions, many of them poorly understood, unhappy medical wanderings such as these have become more likely.

Newborn screening policy should respect human rights. Does newborn screening policy take appropriate account of fundamental and widely re-

spected American values concerning confidentiality, privacy, and informed consent? The mandatory nature of newborn screening seems inconsistent with these values. The standard rationale for mandating public health measures such as mandated immunization or treatment of infectious disease is that the measure will avert serious, imminent harm to others, but this rationale does not apply to newborn screening. Instead, the justification for requiring screening without parental informed consent has been that the risk is minimal and the child will lose a vital benefit if screening is not done immediately. Even under these circumstances, not all ethicists think that omitting informed

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consent is acceptable. Broadening the rationale makes the omission even more questionable. If the rationale is a family benefit, such as information that can inform reproductive decisions or help avoid diagnostic odysseys, or a societal benefit, such as identifying potential research subjects for the study of currently untreatable disorders, then the ethical requirement is clear: parents should be informed and allowed to make their own decisions.

Oddly enough, the new rationale for screening is frequently justified with references to “what parents want.”¹⁵ For example, some argue that parents want to know if the results of tandem mass spectrometry show that a child has a metabolic abnormality, even if there is no treatment—in fact, even if its clinical sig-

nificance is unknown. In practice, “what parents want” usually means “what some of the parents of children already identified as having a specific genetic condition are advocating for.” We should all be deeply sympathetic to parents whose children suffer from diseases that could have been treated by newborn screening. Indeed, in our view, where the evidence that such screening can reliably prevent harm is solid, screening should usually be initiated. Unfortunately, in more ambiguous cases, there is very little hard evidence of what parents typically want. Hearing from parents frightened by a false positive result, or presented with a diagnosis of a condition whose natural history is not clearly

understood and for which no reliable and effective treatment is available, would be especially helpful. Moreover, as important as the views of parents are, there are others who should also have a say. When collective resources are used, taxpayers and those paying insurance premiums also deserve to be heard.¹⁶

Society has not systematically asked individual parents, taxpayers, or premium payers what newborn screening policies they think are appropriate. If society decides to ask, framing the question properly will be a challenge. The question should not be: “Do you want to be able to refuse screening for a specific genetic disorder on behalf of your newborn given that the program is already in place?” Nor should the question be: “Do you think screening newborns for life-

threatening genetic disorders is a good idea?” The right question is not a yes or no question, but one about alternative paths for pursuing good and valuable ends. For example, we could pose the question in a way that helps people understand that screening has an opportunity cost and that gives them examples of what could be obtained if the resources were used differently. We could collect information on how respondents’ views vary with the specific characteristics of the disorder. Probably, most people would agree that screening for PKU is worthwhile; but judgments about the desirability of using scarce resources to screen newborns for conditions that have only minimally effective treatments or whose clinical significance is unclear—especially when those same resources might be devoted to some other activity—are less easy to guess.

Fortunately, there are alternatives to screening newborns on a mandatory basis for all conditions. States can establish pilot programs that offer voluntary screening for a condition and generate the evidence needed to support an informed policy decision to include the condition in a mandatory universal testing program. States can also make screening for a condition available to all newborns in a public program, but on a voluntary basis with informed consent. Physicians can offer parents the option of newborn screening for a condition in the clinical setting, with informed consent. Attractive and practical alternatives to mandatory screening exist for those conditions for which crucial evidence is lacking. It is vital to understand that these alternatives do not deny a known, lifesaving benefit to newborn infants. Instead, under conditions of uncertain benefit, they promote parents’ informed choices while simultaneously allowing us to gather crucial evidence.

Finally, in considering the parental role in newborn screening, we err if we focus entirely on the debate between the opposing claims that “informed consent is an ethical absolute”

and “informed consent is too expensive and time-consuming, and babies will be harmed because some parents will make bad decisions.” The reality is that even in a mandatory program, families have to know enough about newborn screening to understand what is at stake and to cooperate appropriately with the enterprise if newborn screening’s goal of preventing harm to the child is to be achieved.

What kind of information is needed? Ideally, expectant parents should be aware that their newborn baby will be screened for a variety of disorders before leaving the hospital. During pregnancy, there should be basic education designed to convey a few simple messages: “Newborn screening will happen soon after your baby is delivered; your obstetrician recommends it; most babies picked up by screening for a disorder do not have it, but those few who do need urgent treatment; you must follow up immediately if notified of a positive result.” Obstetricians do not have to provide detailed information about all the individual disorders and their consequences, but they should be able to tell parents where to find more information if they want it. In the hospital, mothers should be notified that the screening is being done, and they should be reminded about how important it is to follow up a positive result, even though most babies turn out to be unaffected. Parents receiving a positive result should receive basic information about the specific condition, and of course, the parents of a child with a confirmed diagnosis should receive the detailed information and support they need to understand their child’s condition and manage its impact on the child and the family.

In our project, we focused especially on the key position of obstetricians in the educational process. As part of the project, the March of Dimes collaborated with the New York state newborn screening program to prepare educational materials for obstetricians, and it also produced an educational video on newborn

screening that can be used by obstetricians to educate parents. At the same time, HRSA funded the development of educational materials for parents and health professionals. Individual state programs, the March of Dimes, and parent advocacy groups have all engaged in efforts to educate the public, parents, and health professionals. Thus, some progress is being made in this area, even as the rapid expansion of test panels makes the task of informing parents significantly more complex.

There remains the issue of whether and how blood spots can be stored and used after newborn screening is conducted. Newborn screening blood spots potentially represent a national repository of genetic material from the entire newborn population, making them an invaluable resource for program research and quality assurance activities and for health research in general. Do state policies for blood spots appropriately respect the human right to decide whether to participate in research and how one’s bodily tissues may be used? The answer is complex. Opinions differ about the extent to which it is ethical to use leftover blood samples taken for clinical care purposes with consent, and it is even less clear what the ethical rules should be for leftover samples collected from children for a mandatory public health service. What is clear is that state policies vary dramatically with respect to how long blood spots are stored and the purposes for which they can be used. The variation creates inefficiencies in the implementation of quality control projects and research studies, but more important, the lack of established, enforceable societal ethical norms on matters of informed consent, confidentiality protections, and the use of bodily tissues tends to undermine public trust in the research enterprise.

Some Policy Goals

We can and should improve the process by which newborn

screening policy decisions are made. We should have a rigorous evaluation of evidence before adding a condition, and we need a systematic, cost-conscious plan for collecting evidence afterwards. The evidence review should follow accepted standards and should include the perspectives of experts on evidence-based health policy analysis outside the field of newborn screening. To take adequate account of opportunity cost, we should collect evidence on both costs and benefits, and we should structure the policy process to compare newborn screening with other uses of resources, both within and outside of health care. To fairly distribute costs and benefits, we need more transparency in the financing of programs, better data on what the distribution of costs and benefits looks like, and more uniformity in access to follow-up and treatment. To respect human rights, programs should insist that benefit to the infant is an essential criterion for making newborn screening mandatory. If the benefit is to anyone other than the infant, or if the benefit to the infant is uncertain, then parental informed consent should be required. Finally, we must clarify obligations with respect to consent for using blood spots in newborn screening quality improvement, research related to newborn screening, and research on questions not directly pertaining to newborn screening.

All of these goals would be far easier to achieve in a health care system with certain key elements. All persons should have access to a socially accepted, morally adequate level of health care. The system should have institutional structures to allocate health care resources across the entire spectrum of care with due regard to opportunity cost and stewardship. It should have an integrated electronic health information system with security measures that protect the confidentiality of personal health information and clear rules that govern data use. It should have a comprehensive program of quality improvement and research, using data accumulated in

routine system operation to efficiently generate information that allows patients to better understand their options and health care professionals and organizations to provide better care.

Protecting the health of children is a noble goal. Newborn screening has made significant contributions to children's health since its humble beginnings decades ago. New technologies, new voices, and new opportunities have recently challenged the original ethical foundations of newborn screening programs. We need to meet these challenges honestly and justly.

Acknowledgments

The writing of this article was funded by grant number R01 HG002579-03, "Ethical Decision-Making for Newborn Genetic Screening," from the National Human Genome Research Institute.

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