TALKING WITH PARENTS BEFORE NEWBORN SCREENING

Ellen Wright Clayton, MD, JD

Parents typically make all sorts of decisions about their children's health care.[1] They decide when to provide care themselves or to seek help and guidance. They decide whether to follow the clinician's advice about child-rearing, medications, and tests. Efforts are rarely made, either by clinicians or by the state, to override parents' decisions. There are a lot of reasons for this relatively deferential stance toward parents. Some are quite practical, even utilitarian. Clinicians recognize that parents, who almost always initiate the contact, will not come as readily when they fear they are going to be overruled. Parents are usually in the room when children are poked, prodded, and stuck, and it would be odd not to say something about what is going on and why. Further, parents are less likely to adhere to recommendations when they do not understand them or feel that they were treated rudely. Many clinicians believe that empowering patients and parents to take a greater role in their health care leads to better outcomes. Other reasons for deferring to parents reflect an evolving ethical understanding of the foundations of medical care. Health care decisions often reflect values that vary in socially acceptable ways among individuals. The relatively recent notion that patients, and in the case of children, their parents, have a role to play in their health care reflects a commitment to honoring the values of patients, who are, after all, the ones primarily affected by health care choices.

Many of the practical and ethical factors that shape typical clinician-parent interactions simply did not apply in the same ways to newborn screening (NBS). Perhaps the most important is that NBS is largely invisible to parents. Numerous things happen to babies born in hospitals in the United States: antibiotic ointment is put in their eyes and triple dye is put on their umbilical cords; they receive shots of vitamin K and Hepatitis B vaccine; increasingly they undergo hearing screening; and after 24 hours of age, samples of blood are obtained and sent to laboratories to be screened for a growing array of disorders. Almost all these interventions occur in the nursery, often while the parents are not present. Nor is this a time when parents' attention is focused solely on what is going on with their child, because they are dealing with the biological and social changes that parents experience with labor and delivery.

NBS is a public health activity that discourages parental involvement. These programs were first put into place in the 1960s,[2] just as the notion of patient choice in health care was first emerging. The concept of shared decision-making was not normative at the time for either legislators or public health officials. As a result, only 3 states enacted laws requiring parental permission for NBS. Even now, public health officials are not subject to the same practical and ethical pressures that clinicians feel to defer to parental/patient choice. This is because public health officials have no direct contact with families. Typically these programs are mandatory, altering the role of clinicians as well. It is easy for them simply to say that NBS is required. The major risk from the health care provider's perspective is permitting a parent to refuse NBS only to find that the child was affected with 1 of the disorders that would have been detected.

However, the forces that altered the physician-parent relationship have begun to touch the public health sector as well, leading officials to place more emphasis at least on education. Parents have said for decades that they want to know about NBS.[3,4] However, because public health officials decide what parents are to be told while health care providers are the ones who have to convey the information, responsibility for education is diffused. A recent survey of state NBS programs, for example, revealed that most states used informational brochures and conversation and reported that a combination of health professionals shared responsibility in conveying this information to parents. Twenty-three states (45%) indicated that primary care physicians had some responsibility in informing parents about NBS. Most commonly, states reported that parents were informed just before specimen collection. Three states were unable to report a procedure for informing parents about NBS before testing.[5]

As a result, accountability is diffused as well. This probably explains why some state programs seek written confirmation from parents that they have received educational

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CF	Cystic fibrosis		

From the Center for Genetics and Health Policy, Vanderbilt University, Nashville, Tennessee.

Reprint requests: Ellen Wright Clayton, 507A Light Hall, Center for Genetics and Health Policy, Vanderbilt University, Nashville, TN 37232-0165. E-mail: ellen.w.clayton@Vanderbilt.Edu.

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materials. Such documentation is the only tangible evidence available to state officials, who are not directly involved with parents, but it provides no assurance that effective communication has actually occurred. A major problem with current efforts to educate parents about NBS is that they typically take place in the peripartum period, reflecting the origin of NBS in pediatric practice. This, however, is a terrible time for education because of the many transitions that occur. As a result, parents generally know little about NBS.

Recently, the Newborn Screening Task Force, led by the Health Resources and Services Administration and the American Academy of Pediatrics, recommended that "prospective parents should receive information about newborn screening during the prenatal period. Pregnant women should be made aware of the process and benefits of newborn screening and their right of refusal before testing, preferably during a routine third trimester prenatal care visit."[6] This makes a lot of sense. Birth and the health care needs of the newborn are hardly unforeseeable. One of the goals of the prenatal period is preparation for parenthood, which includes the duties of obtaining appropriate health care for their children.[7,8] One could argue that it is easy for 1 group of physicians to recommend what another group of physicians ought to do, but in 2003, the Committee on Genetics of the American College of Obstetrics and Gynecology (ACOG) endorsed counseling about newborn screening by obstetrical providers.[9] This is good news, but much work will need to be done to convince these providers to incorporate this education into their other responsibilities,[10] to give them the tools they need to succeed,[11] and to ensure that education actually occurs.

Although states increasingly acknowledge the importance of educating parents about NBS, parents in most states still effectively have little choice about whether their child is screened.[12] Some state statutes permit parents to refuse NBS, usually only for religious reasons, but these provisions are relatively empty, because despite current educational efforts, parents rarely know that screening is even occurring. Only 3 states actually require parental permission. Opponents of seeking parental permission argue that no reasonable parent would refuse screening, that it is too expensive to seek consent, and that it is too difficult to document effective informed refusal.[6] Others have argued that parents simply should not be permitted to expose their children to risk by refusing screening.[13] The latter line of reasoning, however, fails to recognize that the risk of any particular child being affected with 1 of the disorders sought in NBS is quite low and that the state appropriately overrides parental choices only to avert a high probability of serious harm.[14]

Recent developments in NBS and in health care more generally, moreover, make the arguments in favor of seeking parental permission increasingly compelling. Until recently, state-run programs screened for disorders, such as phenylketonuria and congenital hypothyroidism, for which early identification and treatment had an enormous impact on outcome. The focus was on the health of the affected child, and public health officials could perhaps be forgiven for believing that the decision was clear. Even these programs have had to deal with false-positive results, children who failed screening only later to be determined to be unaffected.[15] These results can have adverse consequences for both children and their parents, although most families ultimately do well.[16-19] No studies have examined the effect of pre-screening education on assuaging parental anxiety about abnormal screening results. Little is known about the impact of post-screening communication and counseling on ameliorating these risks of NBS, although it does appear that face-to-face conversation with a specialist is helpful.[18,20] Studies of predictive and predisposition testing in adults and of prenatal diagnosis suggest that many factors affect the effectiveness of counseling in reducing distress.[21] Unfortunately, however, a recent report revealed that states vary widely in what sort of counseling they provide, with many having no programs at all to counsel parents of children with false-positive results and some failing to provide counseling even when children are affected.[22]

More recently, programs have begun to identify an array of children for whom the benefits of early detection are not so clear. The largest group is carriers for autosomal recessive traits, first for hemoglobinopathies and now for cystic fibrosis (CF). These children are not and will not be ill. They may be at reproductive risk in the future if they procreate with another carrier, but it is hard to ensure that they have this information when it might be relevant to them. Parents are sometimes confused about the implications for their child of being a carrier,[23] which is hardly surprising because many states do not provide counseling to these families.[22] There is also some degree of irony in having the government require screening that yields results about an infant's carrier status when current norms preclude honoring parental requests for such testing of their children.[19,24] These problems have led some programs to decide not to report carrier status, a choice that may cause some parents to complain that they were denied information to which they feel entitled.

The benefits of early identification of some disorders that have been added more recently are not always compelling. Tandem mass spectrometry, which is being adopted in many states, is a means of identifying many conditions for which there is no effective therapy.[25] Although evidence is mounting that early diagnosis and treatment improves outcome for CF, as is well-demonstrated by many of the articles in this supplement, the magnitude of the benefit of early intervention generally pales in comparison with that for some of the disorders in the original screening programs. Perhaps as a result, people are now discussing benefits to parents primarily, such as informing their subsequent reproductive decision-making and averting delay in diagnosis.[19]

The point of listing these considerations is not to assess their validity, but rather to demonstrate that the calculus of the risks and benefits of current NBS programs is complex and that people of good will can reach different conclusions about the best course of action. Some parents, for example, may not want to know about untreatable disorders or about reproductive risks they or their child may face. Other parents