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George J. Annas

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# Mandatory PKU Screening: The Other Side of the Looking Glass

GEORGE J. ANNAS, JD, MPH

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**Abstract:** The challenge that PKU screening programs face is to be effective without sacrificing individual liberty. Most states have assumed that this is impossible, and have enacted mandatory PKU screening tests. It now appears that in fact voluntary screening for PKU can be effective. Accordingly, it seems appropriate to reexamine existing mandatory screen-

ing statutes to determine if we can replace government coercion with voluntary informed consent. Focus should be placed on the proper role of the government in screening, and on improving the consent process, and not on those few couples who withhold consent. (*Am J Public Health* 1982; 72:1401-1403.)

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At one point in Lewis Carroll's *Through the Looking Glass*, Alice finds herself discussing the preventive possibilities of the law. The Queen tells her that the King's Messenger is in prison, being punished but that his trial doesn't begin until next week, "and of course the crime comes last of all." Alice is shocked and asks, "Suppose he never commits the crime?" The Queen responds, "That would be all the better." Alice agrees that it would be better if the crime was not committed but, "it wouldn't be all the better his being punished for it." The Queen insists she's wrong; that it's always better if people don't commit bad acts, even if they're punished as if they did. If they didn't commit the acts, "that would have been better still; better and better, and better!" The conversation is interrupted when Alice begins to say, "There's a mistake somewhere . . ."

Elsewhere in this issue of the Journal, Dr. Ruth Faden and her coworkers report on their study which demonstrates that requiring informed consent for PKU (phenylketonuria) screening is well-accepted by the public, improves the public knowledge about PKU screening, and does not make the program any less cost-effective.<sup>1</sup>

I would have thought the natural conclusions to such a study would have been a sigh of relief followed by an affirmation of present Maryland consent policy and, perhaps, a suggestion that states with mandatory PKU screening programs can begin to consider making them voluntary. So it is puzzling that we are treated instead to a commentary by three of the study's four authors arguing that the 27 parents who did refuse to have their children screened were not morally justified.<sup>2</sup>

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Address reprint requests to George J. Annas, JD, MPH, Edward Utley, Professor of Health Law, and Chief, Health Law Section, Boston University School of Public Health, 80 East Concord Street, Boston, MA 02118. This paper, submitted to the Journal July 26, 1982, was accepted for publication August 2, 1982.

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It is because I believe these parents were morally justified in their refusal, and because the study demonstrates that PKU screening programs can be both effective and voluntary, that I agreed to write this commentary. As Alice put it, "There's a mistake somewhere . . ."

In fact, I think that in arguing that things can get "better and better, and better" by returning to mandatory screening, Dr. Faden and her coworkers make two fundamental mistakes that cloud their laudatory instincts; the first is concluding that PKU screening must entail a choice between two competing values; and the second is uncritically applying the "harm principle" to a whole population scenario.

## *Values in Conflict?*

The dilemma for those who want to reduce genetic disease has traditionally been seen as one of governmental effectiveness versus individual freedom. The notion has been that to be effective, individual liberty must be sacrificed. As Daniel Callahan noted almost a decade ago, the real challenge is to find a "way of combining both logics." This would be difficult, he opined, "if only because most people find it easier to cope with one idea than with two at the same time." And he described what would be involved:

It will mean taking the idea of free choice seriously, allowing parents to make their own choice without penalizing them socially for the choices they make, or condemning them for those choices which will increase the financial costs to society. Part of the very meaning of human community, I would contend, entails a willingness of society to bear the social costs of individual freedom.<sup>3</sup>

The most remarkable finding of the Faden PKU survey is not that irreconcilable values are in conflict, but that in Maryland at least, we can simultaneously protect the values of both beneficence and autonomy. We can permit PKU screening programs to be voluntary without running any substantial risk of missing an affected infant. Rejoicing rather than anguishing seems a more appropriate response. As the authors themselves note, "the chance of missing a

PKU infant at the observed rate of parental refusal (0.05 per cent) is 100 times less than the chance of missing a PKU infant because of a false negative test result.<sup>11</sup> In fact, at the observed rate of refusal, it would take 500 years before one case is missed for this reason. Efforts aimed at decreasing the number of false negatives rather than decreasing the small number of parental refusals would seem more reasonable.

### *Parental Autonomy*

The authors correctly argue that the primary purpose of parental autonomy in decision-making for their children is so that they can provide their children with the opportunity to grow and develop to be able to exercise their own autonomy.<sup>4,5</sup> Accordingly, when parents refuse standard medical treatment for a child who will die or become seriously ill if he or she is not treated, the state properly intercedes to ensure treatment.<sup>4,6</sup>

But what if the child is not sick and the parents have not withheld treatment for the child's condition? What if, as Alice asked the Queen, no negligent or intentional detrimental act has been committed by the parents? Should the state nonetheless force their children to be screened for diseases which they almost certainly do not have? This is a much more difficult question than the authors suggest.

The screening scenario is quite different than the dying child scenario. With the defective newborn, for example, there is usually almost 100 per cent probability the child will die without treatment.<sup>4,5</sup> In PKU, of 14,000 infants, 13,999 will be healthy. The odds are overwhelming that if any particular set of parents refuse screening, no detriment at all will befall their child. The proper question is thus not the individual-centered one which the authors pose: "Do parents have the right to consign their children to a state of irreversible mental retardation?", but rather the societal one: "Is mandatory screening for PKU a legitimate exercise of the state's public health powers?"<sup>7\*</sup> This requires an analysis of testing and treatment technologies, the incidence of the disease, resource allocation, and the role of law in promoting the nation's genetic well-being.

Of all mandatory newborn screening laws, PKU laws have been the most vigorously defended, and with good reason. The test is simple, safe, cost-effective, accurate, and detects a severely disabling condition that can be successfully treated if identified early. Newborns should be screened for PKU. But, as the new data from the Faden study reveal,

\*Stating the question in terms of parental rights to refuse smacks of "blaming the victim" in this context. Although only seven of the 27 refusers were studied, the authors note that five of the seven had a "poor understanding of the screening." Perhaps the explanation lies in the poor manner in which it was or was not explained to them. Two of the mothers did not speak English, and we do not know if an interpreter was available. The other two women actually did have their children tested later, apparently by their private pediatrician. These data not only lower the actual number of "refusers" but also suggest that it could be lowered even more by better explanations and the use of interpreters.

none of this necessarily means we need *mandatory* screening laws.<sup>1</sup> What it could just as well indicate is that, while mandatory laws helped to initially set up reasonably good laboratories and testing procedures and helped alert practitioners and the public to the disease and its test and treatment, such mandatory laws are no longer necessary. We may be able to return the public's autonomy without sacrificing the program's efficiency.

The authors seem to miss this point by assuming that any parental refusal must be both immoral and irrational. The immorality has been dealt with previously. What about rationality? First, people may have religious reasons for refusing screening and, if their child is in no danger, these should be respected. Secondly, parents may simply believe that for them the risk is not worth the benefit.

Currently, the rate of false positives is much higher than the rate of true positives, so it is much more likely that the child will be falsely identified as affected with PKU than that he/she will be accurately identified. This strikes the authors as a trivial risk, although they do recognize a state "obligation to ensure adequate follow-up." Given their thoughtful empirical work, I would have guessed they might propose a study of the health implications of being identified as a false positive. A recently concluded study of the parents of 60 infants in Massachusetts who were retested in a newborn screening program found that the more accurate information the parents obtained about the test and reason for the retest, the less anxious and depressed they were. Nevertheless, 36 per cent of the parents of these normal infants reported heightened concern about the health of their infant because of the repeat testing. The authors could not determine how long these concerns would last, or what actions they would take.<sup>\*\*</sup>

This may not strike one as an adequate reason for refusing PKU screening. But look into the future when we will be able to screen for 1,000 more diseases. Suppose, for example, a computerized screening test for 1,000 conditions. Suppose further that each of these tests has been so perfected that the false positive rate is only 1 per cent. Each infant screened will then be diagnosed initially as suffering from 10 disorders, even though he/she suffers from none. If the false positive rate is 5 per cent per test, he/she will appear to have 50 disorders, etc. The tests that are performed for rare diseases, the more likely it is that pathology will be generated from the retesting procedures, and the more rational a decision not to screen initially becomes. This is independent of any stigma that may accompany a true positive diagnosis.<sup>8</sup> The rare parent who refuses newborn screening, both today and in the future, is likely to be viewed as either a child neglecter or an irrational anti-science fanatic. Neither label seems accurate or helpful. Such labels seem to be the result of uncritically applying the public health model, with its

\*\*Personal communication from Dr. James Sorenson referring to research that is completed but not yet published, i.e., Sorenson JR, Levy HL, Mangione TW, Sepe SJ: "Parental Response to Repeat Testing of 'False Positive' Infants in a Newborn Screening Program."

emphasis on the good of the entire population, to family decisions where the medical model, with its emphasis on the good of the individual patient, is more appropriate.

### *The Role of Law in Public Health*

Law can effectively promote the public's health. But in the United States, law also promotes individual liberty. Liberty should be sacrificed only when clearly necessary for the public good.<sup>9-14</sup> As the Faden study indicates, PKU screening may not necessitate such a sacrifice.<sup>1</sup> The 1975 recommendations of the National Academy of Sciences regarding genetic screening seem even more relevant today:

*"Genetic screening is appropriate when:*

*"(1) There is evidence of substantial public benefit and acceptance, and acceptance by medical practitioners.*

*"(2) Its feasibility has been investigated and it has been found that benefits outweigh costs; appropriate education can be carried out; test methods are satisfactory; laboratory facilities are available; and resources exist to deal with counseling, follow-up, and other consequences of testing.*

*"(3) An investigative pretest of the program has shown that costs are acceptable; education is effective; informed consent is feasible; aims have . . . been defined . . . qualified and effective counselors are available in sufficient number. . . .*

*"(4) The means are available to evaluate the effectiveness and success of each step in the process."<sup>13</sup> (Emphasis added.)*

In regard to mandatory statutes, the Academy's Committee specifically recommended that "participation in a genetic screening program should not be made mandatory by law, but should be left to the discretion of the person tested or, if a minor, of the parents or legal guardian."<sup>9</sup> This latter recommendation has been generally ignored, and almost all states now have mandatory PKU screening programs. Maryland is an exception, but not without controversy. When its consumer-majority Commission on Hereditary Disorders recommended that its mandatory screening law be changed to a voluntary one, the Maryland Chapter of the American Academy of Pediatrics asked that the requirement for written parental consent be deleted because "it feared that informed consent would be extended to other routine procedures and also that many parents would refuse to allow their infants to be screened."<sup>14</sup>

Their second fear turned out to be baseless, but the Faden study should cause us to reexamine other "routine" newborn procedures to determine if they are susceptible to an informed consent approach. Dr. Faden, for example, uses silver nitrate prophylaxis as an example of a procedure parents should not be able to refuse.<sup>2</sup> But why not? The procedure helps the infant only if the mother suffers from

venereal disease at the time of birth. There are also less irritating alternative drugs to silver nitrate. Why shouldn't the mother be able to refuse this treatment (provided she agrees to be tested for venereal disease), and why shouldn't she be able to choose among drug treatment alternatives if the child's eyes need treatment? Morally and legally, if the parents can demonstrate that the treatment is not necessary, they should be able to refuse it.<sup>6</sup>

Maryland's screening statute has long been considered the best in the country and has been proposed as a model for other states to emulate.<sup>7</sup> Its most controversial provision—the requirement for written informed consent—has now been proven feasible.<sup>1</sup> The proper response is not to change the Maryland statute, but to renew the effort to get states with mandatory statutes to reexamine the feasibility of running an effective screening program on a voluntary basis.

Alice's legal conversation with the White Queen began with a discussion of the advantages of "living backwards." The Faden study should encourage us to at least look backwards to the principles of voluntary screening so boldly trumpeted in the mid-1970s and see if it is not possible to implement effective screening programs that preserve liberty in the 1980s.

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