

2014

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Sonia M. Suter, *Did You Give the Government Your Baby's DNA? Rethinking Consent in Newborn Screening*, 15 MINN. J.L. SCI. & TECH. 729 (2014).

Available at: <https://scholarship.law.umn.edu/mjlst/vol15/iss2/3>

Did You Give the Government Your Baby's DNA? Rethinking Consent in Newborn Screening

Sonia M. Suter*

ABSTRACT

Newborn screening (NBS) has long offered the possibility of identifying rare conditions, which can be lethal or debilitating if not detected and treated quickly in the newborn period. These screening programs, usually mandatory, have been well established in every state since the 1960s. In the last decade, the number of conditions screened for has risen exponentially to include more than fifty inborn errors of metabolism, blood disorders, genetic, or other conditions. Not surprisingly, newborn screening programs have been widely accepted for their potential to save the lives of countless children.

Despite their valuable public health benefits, however, old approaches to, and more recent expansions of, NBS raise important privacy and policy concerns. NBS samples are collected in most states without affirmative, or sometimes any, consent from parents. NBS programs now screen for an ever-broadening range of diseases—sometimes without careful assessment of the risks and benefits—including conditions for which there is no treatment. NBS samples are retained for long periods or indefinitely. And finally, few, if any, limits prevent potentially invasive uses of these samples by the government or third parties. Indeed, evidence suggests that a great deal of research is being conducted on these stored blood spots, the

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collection and storage of which many parents are simply unaware. Only a few lawsuits and legislatures have addressed the legality of these practices.

With recent expansions in the scope of NBS and increased interest in these samples for research, it is time to take a fresh look at this long-standing public-health system and to reexamine some of the underlying philosophies and practices associated with it. While NBS offers important public health benefits, it also threatens some of the civil liberties of the parents and children involved. This piece argues for the need to strike a careful balance between the public goods and private interests, and describes a methodology that allows these competing values to be recognized in policymaking. It concludes by suggesting ways to balance the important values of maximizing the well-being of newborns and promoting research, while also protecting autonomy and privacy as much as possible.

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INTRODUCTION

If you ask parents whether their child should undergo genetic testing or participate in research, most would probably say, consistent with legal norms in most areas of medicine, “only with my consent!” Yet the majority of parents do not realize that in every state, a small blood sample is collected from newborns to test for inborn errors of metabolism (many of which are inherited).¹ Nor do they realize that, in many states,

1. See Taralyn Tan, *Newborns' DNA: Don't Deny Scientists This Useful Resource*, GENETIC ENGINEERING & BIOTECH. NEWS (Apr. 13, 2010), <http://www.genengnews.com/gen-articles/newborns-dna-don-t-denyscientists-this-useful-resource/4377> (“[I]n most cases, parents are not aware that the blood sample from their child is being kept at all.”).

the dried blood spots (DBS) are retained for long periods or indefinitely, with few, if any, limits on third-party access to and uses of these samples.² Indeed, evidence suggests that a great deal of research is being conducted on these stored blood spots by the state and other entities.³ All of this, from collection to retention of samples, often comes without parents' affirmative, let alone informed, consent.⁴

The impetus for mandatory newborn screening (NBS) is the fact that rarely, but quite significantly, a child will be born with abnormal levels of enzymes, metabolites, or other chemicals, which can be lethal or debilitating if not detected and treated in time.⁵ NBS offers the possibility of identifying some of these conditions before clinical symptoms manifest and "before developmental disabilities or death occurs."⁶ These, usually mandatory, screening programs have been well established in every state since the 1960s, potentially saving the lives of countless children.⁷ The scope of NBS programs has expanded dramatically in recent years, with most states screening for between twenty-seven⁸ and over fifty inborn errors of

2. See Lori Andrews, *Public Choices and Private Choices: Legal Regulation of Genetic Testing*, in JUSTICE AND THE HUMAN GENOME PROJECT 46, 55 (Timothy F. Murphy & Marc A. Lappé eds., 1994) (noting that genetic information can change lives, "precipitated by the release of genetic information to third parties—such as when insurers or employers make adverse decisions against people based on genetic information"); Tan, *supra* note 1 (discussing DNA warehousing and the indefinite retention of samples).

3. See, e.g., Tan, *supra* note 1 ("[S]torage . . . allows geneticists and neonatology researchers access to an incredible genetic database. These blood spot samples can be utilized to develop new genetic tests, to learn more about existing genetic disorders, and to study factors such as the mother's health and in utero environment in relation to rare disorders.").

4. *Id.*

5. See *Newborn Screening*, *Pediatric Genetics*, CENTERS FOR DISEASE CONTROL & PREVENTION, http://www.cdc.gov/ncbddd/pediatricgenetics/newborn_screening.html (last updated May 13, 2013) (discussing the importance of newborn screening and the benefits derived from the process).

6. See Michael S. Watson et al., *Newborn Screening: Toward a Uniform Screening Panel and System*, 8 GENETICS MED. 1S, 1S (Supp. May 2006) ("States and territories mandate newborn screening of all infants born within their jurisdiction for certain disorders that may not otherwise be detected before developmental disability or death occurs.").

7. *Id.* (discussing the importance of the state-based newborn screening programs that began over forty years ago).

8. STEFAN TIMMERMANS & MARA BUCHBINDER, SAVING BABIES? THE CONSEQUENCES OF NEWBORN GENETIC SCREENING 59 (2013) ("By 2010, all states screened for 27 . . . conditions."); Wylie Burke et al., *Genetic Screening*,

metabolism.⁹ Some of these conditions have been added to the list without careful assessment of the risks and benefits,¹⁰ and some are identified and reported with no known effective treatment.¹¹

Even so, NBS has been a well-accepted part of our public health system for nearly half a century.¹² Recently, a few lawsuits have challenged the consent requirements with respect to NBS and related research. In 2003, a couple claimed that Nebraska's efforts to compel the screening of their newborn violated their religious freedom and parental rights.¹³ The Nebraska Supreme Court found no such violation.¹⁴

33 EPIDEMIOLOGIC REVIEWS 148, 149 (2011) (“In the United States, most states screen for at least 29 conditions . . .”); *see also* ASSESSING GENETIC RISKS: IMPLICATIONS FOR HEALTH AND SOCIAL POLICY 66 (Lori B. Andrews et al. eds., 1994) [hereinafter AGR] (“Newborns are usually screened today for several inborn errors of metabolism . . .”).

9. *See* Louise Moody & Kubra Choudhry, *Parental Views on Informed Consent for Expanded Newborn Screening*, 16 HEALTH EXPECTATIONS 239, 239 (2011) (mentioning that all states now screen for fifty-three core conditions to detect inherited metabolic diseases). This range of conditions includes what are described as twenty-nine core conditions and a secondary group of twenty-five targets that can be identified by screening for the core set. TIMMERMANS & BUCHBINDER, *supra* note 8, at 50, 63.

10. *See* Beth A. Tarini et al., *Waiving Informed Consent in Newborn Screening Research: Balancing Social Value and Respect*, 148C AM. J. MED. GENETICS 23, 23–24 (2008) (mentioning that “new NBS tests have rarely been subjected to population-based study” and demonstrating the difficulties of assessing risks and benefits).

11. *See* Andrews, *supra* note 2, at 58 (“Given the current state of development of medical genetics, . . . effective treatment for genetic disorders is rare . . .”); Ellen Wright Clayton, *Currents in Contemporary Ethics: State Run Newborn Screening in the Genomic Era, or How to Avoid Drowning When Drinking from a Fire Hose*, 38 J.L. MED. & ETHICS 697, 698 (2010) (noting that for many of the reported results of newborn screening, “the efficacy and utility of therapeutic and preventative interventions are not clear”).

12. Watson, *supra* note 6, at 1S.

13. *See* Douglas Cnty. v. Anaya, 694 N.W.2d 601, 604 (Neb. 2005) (discussing the Anaya's argument that the requirement violated their “First Amendment right to free exercise of religion and their fundamental rights as parents”).

14. *Id.* at 608 (concluding that the requirement did not “unlawfully burden the Anayas' right to freely exercise their religion” or “unlawfully burden their parental rights,” mentioning the lack of evidence that the state had an anti-religious purpose in enforcing the law and the valid policy interests in addressing the health and safety of children born in Nebraska).

The more recent “Baby DNA Lawsuits”¹⁵ have challenged the involuntary collection and dissemination of NBS samples to researchers for purposes other than NBS.¹⁶ In Minnesota, the state Supreme Court ruled that the state’s dissemination and use of newborns’ DBS for research without obtaining written informed consent violated its Genetic Privacy Act.¹⁷ Two similar lawsuits were brought in Texas. The state settled with the five plaintiff parents in the first suit after agreeing to destroy all samples collected without parental consent since 2002.¹⁸ A class action filed in late 2010 in Texas was dismissed as moot because there was no evidence that the parties’ newborn samples were actually used or distributed for research.¹⁹

I argue in this Article that these lawsuits and other developments in NBS should give pause to the presumption that parental consent is not necessary with respect to NBS. We already obtain much more information from NBS than we did in the past and we are on the cusp of being able to obtain substantially more information in the near future. Moreover, the nature of the information we will be able to glean will be of varied value, certainty, and complexity, raising issues not only about what diseases we should screen for, but whether parents should be required to consent to some or all parts of the NBS process. In addition, the fact that newborn samples are increasingly used for research, and that anonymization of biospecimens is increasingly difficult, supports the need to

15. K.J. Mullins, *Bill to Ban Unauthorized Use of Infant DNA Clears Senate Committee*, DIGITAL J. (Feb. 11, 2010), <http://www.digitaljournal.com/article/287446>.

16. *See id.* (pointing out that NBS samples are used for unauthorized research).

17. *Bearder v. Minnesota*, 806 N.W.2d 766, 776 (Minn. 2011) (holding that there is no authority in the statute to disseminate blood samples or genetic information, without consent, “beyond that expressly authorized for the reporting of newborn test results”). *See generally* MINN. STAT. §13.386 (2010) (Minnesota’s Genetic Privacy Act).

18. *See Higgins v. Tex. Dep’t of Health Servs.*, 801 F. Supp. 2d 541, 545–46 (W.D. Tex. 2011) (discussing the settlement of the earlier *Beleno* lawsuit, and the agreement to “destroy all blood specimens taken as part of the newborn screening program” prior to May 2009, for which no written consent existed); Allison M. Whelan, Note, *That’s My Baby: Why the State’s Interest in Promoting Public Health Does Not Justify Residual Newborn Blood Spot Research Without Parental Consent*, 98 MINN. L. REV. 419, 430–31 (2013).

19. *Higgins*, 801 F. Supp. 2d at 554 (“Plaintiffs never refute Defendants’ evidence that Plaintiffs’ children’s blood samples were not distributed and have in fact been destroyed. Accordingly . . . their claims are now moot.”).

rethink the role of consent in NBS, at least with respect to storage and research uses of DBS. As I will argue, the case for consent with respect to research also supports, in part, the notion of consent for NBS itself.

Yet, just as changing circumstances provide reasons to rethink parental consent with respect to NBS, the increasing scope of information we can glean from NBS makes the possibility of obtaining fully informed consent that much more problematic logistically, practically, and economically. In addition, the DBS are potentially valuable resources for research that can benefit the common good, generally, and the pediatric population, in particular. Thus, the question of consent in NBS raises issues about how to strike the right balance between the public good and private interests.

This Article offers a proposal for finding the right balance of consent for NBS itself, and for the storage and use of DBS. Part I offers a history of NBS and its evolution. Part II explores the rationales for the limited consent provisions for NBS as well as the growing practice of retaining these samples and using them for purposes that go beyond the original goals of NBS. Part III highlights the ways in which the public good comes into conflict with the private interests and describes a methodology that allows for these competing values to be recognized in policymaking. It concludes by suggesting that requiring affirmative consent for NBS and for research on DBS best balances the values of protecting the newborn's well-being and promoting research, while also protecting autonomy and privacy as much as possible.

I. THE EVOLUTION OF NEWBORN SCREENING

NBS begins with a heel prick and the collection of a few drops of blood on filter paper, or Guthrie cards.²⁰ It is a preventive health measure that involves the analysis of the newborn's blood for various medical conditions, many of which are inherited, including certain inborn errors of metabolism and

20. See AGR, *supra* note 8, at 39 (“This test could be performed on a spot of blood obtained from a heel prick before the infant left the hospital nursery.”). The Guthrie cards are named after Dr. Robert Guthrie, who developed the first NBS assay for phenylketonuria. Clayton, *supra* note 11, at 697; *Spotlight on NBS Researchers, Robert Guthrie, MD, PhD*, NEWBORN SCREENING TRANSLATIONAL RES. NETWORK, <https://www.nbstrn.org/about/spotlight/Guthrie> (last visited Mar. 6, 2014).

blood disorders.²¹ The value of conducting screening during the newborn period is both practical and clinically significant. Most infants are born in hospitals, which makes the systematic collection of samples easier at this stage of life than nearly any other.²² In addition, for many of the diseases screened, treatment must be started in the newborn period to prevent the development of clinical symptoms.²³

As its name suggests, NBS is a *screening* program in which an abnormal result does not necessarily identify the presence of disease. It merely indicates an increased risk that the child has the condition, necessitating confirmation through diagnostic testing.²⁴

With its inception nearly fifty years ago, NBS is the longest program of genetic screening in the history of genetics.²⁵ The first state program screened for phenylketonuria (PKU), a disease in which the child lacks a vital enzyme that breaks down the amino acid, phenylalanine.²⁶ Without this enzyme, phenylalanine can accumulate in the brain, causing mental retardation, unless the affected child eats a phenylalanine-free diet.²⁷ The first program, developed in Massachusetts, was

21. *E.g.*, *Newborn Screening*, *supra* note 5.

22. *See* MARIAN F. MACDORMAN ET AL., NAT'L CTR. FOR HEALTH STATISTICS, CTRS. FOR DISEASE CONTROL & PREVENTION, HOME BIRTHS IN THE UNITED STATES, 1990–2009, at 1 (2012) (showing that only 0.72% of births took place in the home in 2009).

23. *E.g.*, *Newborn Screening Tests*, KIDSHEALTH, http://kidshealth.org/parent/system/medical/newborn_screening_tests.html# (last visited Mar. 1, 2014) (“[E]arly diagnosis and proper treatment can make the difference between lifelong impairment and healthy development.”); *see also* Clayton, *supra* note 11, at 697 (discussing the policy behind newborn screening and the rationale of “adding disorders to the newborn screening panel only if early detection and treatment could avert serious harm”).

24. AGR, *supra* note 8, at 65 (“These screening tools are not definitive diagnostic tests, however, and positive results must be confirmed through specific testing for the disease in question.”).

25. *See* Nancy S. Green et al., *Newborn Screening: Complexities in Universal Genetic Testing*, 96 AM. J. PUB. HEALTH 1955, 1955 (“Newborn Screening (NBS) is the first and largest example of systematic, populationwide genetic testing . . .”).

26. AGR, *supra* note 8, at 66.

27. *See id.* (stating that “high phenylalanine levels” can lead to mental retardation, and that a phenylalanine dietary restriction is “highly effective in preventing mental retardation”). The deficient enzyme is called phenylalanine hydroxylase. *Id.*

voluntary.²⁸ This is in sharp contrast, as I will address in Part II, to what is essentially mandatory screening in many states. Most states do not require affirmative parental consent under the theory either that the police powers justify this public health measure or under the doctrine of *parens patriae*.²⁹

While PKU was the primary disease screened for in the early days of NBS, the panel of NBS diseases has expanded considerably in the last few years. The initial expansion, however, was quite slow, with only a few diseases added per decade.³⁰ As late as 2003, the number of diseases screened for in most states was still quite low—eight or fewer diseases.³¹ Technological advances, however, changed that. While initial NBS required a separate assay for each disorder, the development of tandem mass spectrometry (MS/MS) in the 1990s allowed for the identification of over forty conditions through a single test,³² contributing greatly to the expansion of

28. See Newborn Screening Task Force, Am. Acad. of Pediatrics, *Serving the Family from Birth to the Medical Home: Newborn Screening: A Blueprint for the Future—A Call for a National Agenda on State Newborn Screening Programs*, 106 PEDIATRICS 389, 389 (2000) [hereinafter NBSTF] (“By 1962, Massachusetts launched a voluntary newborn PKU screening program that demonstrated the feasibility of mass genetic screening.”). Initially, “the American Medical Association (AMA) and its state organizations opposed mandatory screening as an infringement of physicians’ rights to regulate their professional practice.” TIMMERMANS & BUCHBINDER, *supra* note 8, at 38.

29. See *infra* Part II.A.

30. See Burke et al., *supra* note 8, at 149 (providing background information on the expansion of NBS).

31. U.S. GOV’T ACCOUNTABILITY OFFICE, GAO-03-449, NEWBORN SCREENING: CHARACTERISTICS OF STATE PROGRAMS 2 (2003) [hereinafter GAO], available at <http://www.gao.gov/new.items/d03449.pdf> (“While the number of genetic and metabolic disorders included in state newborn screening programs range from 4 to 36, most states screen for 8 or fewer disorders.”).

32. See Cecilia I. Kaye et al., *Introduction to the Newborn Screening Fact Sheets*, 118 PEDIATRICS 1304, 1307, 1310 (2006) (discussing how MS/MS has led to additional disorders added to screening panels and the essential role played by pediatricians throughout the process). See generally Bridget Wilken et al., *Screening Newborns for Inborn Errors of Metabolism by Tandem Mass Spectrometry*, 348 NEW ENG. J. MED. 2304, 2309 (2003) (“It is now possible to screen rapidly, simultaneously, and inexpensively for a number of very rare disorders with the use of tandem mass spectrometry.”). Tandem mass spectrometry screens for inborn errors of metabolism by measuring the levels of various metabolites in the blood. *Id.* at 2305. Abnormalities in the levels of these metabolites suggest the presence of metabolic disorders. Mary Ann Baily & Thomas H. Murray, *Ethics, Evidence, and Cost in Newborn Screening*, HASTINGS CENTER REP., May–June 2008, at 23, 25. MS/MS can also screen for

NBS.³³ After several years of much variability in screening practices, a consensus began to emerge about the need for more uniformity in NBS, especially with respect to screening panels.³⁴ The American College of Medical Genetics (ACMG) issued recommendations for the standardization of the selection of NBS diseases in 2005, which were endorsed by several professional groups.³⁵ Now every state tests or will test for a minimum of twenty-nine conditions.³⁶ Some panels include over fifty disorders.³⁷

As technologies allow us to test for more diseases more efficiently, the question of what diseases should be included in each state's NBS panel remains difficult and, as we shall see later, has some bearing on the question of whether parental consent should be required. Among the relevant criteria are, of course, scientific considerations, such as the prevalence of the condition in the population, the validity of the NBS test, and the efficacy of available treatments.³⁸ But other non-scientific considerations also play a vital role. Political concerns—such as

PKU and other amino acid disorders, but it does not allow for the testing of all NBS disorders. Kaye et al., *supra* at 1310.

33. TIMMERMANS & BUCHBINDER, *supra* note 8, at 17. Interestingly, in the United Kingdom, “there was insufficient evidence and cost-effectiveness to support tandem mass spectrometry technologies for newborn screening,” whereas in the United States, these factors did not inhibit the use of this technology because “cost-effectiveness is often neglected within health policy discussions, due to cultural anxieties about healthcare rationing.” *Id.* at 58.

34. *Id.* at 34 (“The United States is one of only two industrialized countries without a national newborn screening policy.”).

35. *Id.* at 50, 59. Although the report was one of the most controversial reports on NBS issued by an advisory body, it was also one of the most influential, in large part because it was strongly endorsed by such groups as the March of Dimes Foundation; The American Academy of Pediatrics; the Association of Women's Health, Obstetric and Neonatal Nurses; and the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. *Id.* at 59.

36. *Id.* at 50; Watson et al., *supra* note 6, at 1S (“[T]he expert panel identified 29 conditions for which screening should be mandated.”). I should note that I was part of the panel.

37. Moody & Choudhry, *supra* note 9, at 239 (“All states in the USA now screen for 53 core conditions . . .”).

38. The “classical” criteria used by states in determining which conditions to include in their NBS panels were derived from a seminal paper for the World Health Organization by Wilson and Jungner. See Heather Harrell, *Currents in Contemporary Ethics: The Role of Parents in Expanded Newborn Screening*, 37 J.L. MED. & ETHICS 846, 846–47 (2009) (discussing Wilson and Jungner's ten criteria to apply when considering population screening).

the existence of advocacy groups³⁹ and cost-benefit analysis⁴⁰—are also hugely influential. And, of course, ethical considerations should and often do come into play.⁴¹ For example, because the benefits to the newborn, to the family, and to society do not necessarily overlap, decision makers must decide whose benefits should determine the selection of the screening panel.

If the goal of NBS is to benefit the newborn, the panel of diseases should be limited to those for which we have effective treatments or early intervention and whose natural history we understand well. If we also consider the benefits to the family, however, the panel of diseases might be broader because it would include diseases with no treatment that might help parents make better informed reproductive decisions about

39. In the context of NBS, parents have been strong advocates for expanding the array of tests. Advocacy and lobbying have been strong forces in the development and evolution of NBS. As Ellen Wright Clayton observes, NBS laws were influenced more by individual practitioners and political groups than anything else. Clayton, *supra* note 11, at 697–98 (discussing how most programs in the United States were driven by a report endorsed by the government committees and parent advocacy groups); see TIMMERMANS & BUCHBINDER, *supra* note 8, at 39, 44–48, 59–61 (describing the powerful role of advocacy in promoting NBS and its expansion).

40. See Harrell, *supra* note 38, at 846–47 (explaining that the criteria when considering population testing boils down to screening “illnesses that are sufficiently understood” and can be tested in a cost-effective manner). One of the reasons PKU screening was so widely applauded was its high cost savings of \$93,000 per detected case. *Report of the NIH Consensus Development Conference on Phenylketonuria (PKU): Screening & Management: Chapter II*, NAT’L INST. CHILD HEALTH & HUM. DEV., <https://www.nichd.nih.gov/publications/pubs/pku/Pages/sub30.aspx> (last updated Dec. 21, 2011). The costs of screening per detected case, however, can sometimes be quite large. See OFFICE OF TECH. ASSESSMENT, U.S. CONG., HEALTHY CHILDREN: INVESTING IN THE FUTURE 106–11 (1988) (demonstrating the variability in cost amongst different screening and testing strategies). Some groups, such as the March of Dimes, have taken the view that newborns should be screened regardless of how rare the disorder is, in essence rejecting considerations of cost-benefit analysis. See *Newborn Screening*, MARCH OF DIMES, <http://www.marchofdimes.com/baby/newborn-screening.aspx> (last visited Mar. 26, 2014) (expressing their desire for mandatory testing of extremely rare diseases, most of which, but not all, can be treated or dealt with). This perspective is more political or ethical than scientific, since it may not result in the greatest health benefit to the community, though it is quite a sympathetic position from the perspective of the individual families who benefit from such an approach. See Press Release, N.Y. Dep’t of Health, State Health Department Receives March of Dimes Award for National Leadership in Newborn Screening (Dec. 14, 2007) (lauding New York’s comprehensive NBS program).

41. See *infra* text accompanying notes 42–44.

whether to undergo prenatal testing with future pregnancies.⁴² In addition, such information can avoid diagnostic odysseys, when parents search long and hard for the diagnosis of a rare condition.⁴³ Finally, if we focus on the benefits to society, the panel of diseases would be even larger, including conditions about which we have limited knowledge and no effective treatments so that we can identify potential research subjects to learn more about the natural history of the disease.⁴⁴

For some time, the consensus has been that the benefits to the newborn should be decisive in selecting conditions for NBS since the *raison d'être* of the program is to protect infants from debilitating diseases.⁴⁵ Despite this consensus, these criteria have not always been followed in practice.⁴⁶ Because state health departments have substantial discretion to decide which

42. Many parents would seek prenatal testing with future pregnancies, even if they did not plan to terminate affected pregnancies. Peter T. Rowley, *Parental Receptivity to Neonatal Sickle Trait Identification*, 83 PEDIATRICS 891, 892 (1989) (noting that most women at risk for having a child with sickle cell anemia wanted prenatal testing even though only one quarter would terminate the pregnancy if the fetus were affected). *But see* Ranjeet Grover et al., *Newborn Screening for Hemoglobinopathies: The Benefit Beyond the Target*, 76 AM. J. PUB. HEALTH 1236, 1236–37 (1986) (reporting that fourteen out of twenty-three women at risk for having a child with sickle cell anemia had an amniocentesis and three of the four affected pregnancies were terminated). Some have observed that this rationale for NBS makes it less about protecting the newborn and more about eugenic goals of eradicating undesirable conditions in the population. *See, e.g.*, TWILA BRASE, CITIZENS COUNCIL ON HEALTH CARE, NEWBORN GENETIC SCREENING THE NEW EUGENICS? THE CASE FOR INFORMED CONSENT REQUIREMENTS FOR GENETIC TESTING, BABY DNA STORAGE AND GENETIC RESEARCH 1 (2009), available at http://www.cchfreedom.org/pr/NBS_EUGENICS_REPORT_Apr2009_FINAL.pdf.

43. Baily & Murray, *supra* note 32, at 28–29.

44. TIMMERMANS & BUCHBINDER, *supra* note 8, at 51 (describing how consideration of not just individual benefits, but also benefits to the family and society is an example of “benefit creep”).

45. *See* J.M.G. WILSON & G. JUNGNER, WORLD HEALTH ORG., PRINCIPLES AND PRACTICE OF SCREENING FOR DISEASE 14 (1968) (stating that the aim of early detection is to protect the individual). For a broader discussion and criticism of the shift in focus of some NBS programs from benefit to the infant to benefit to the family and society, see generally PRESIDENT’S COUNCIL ON BIOETHICS, THE CHANGING MORAL FOCUS OF NEWBORN SCREENING: AN ETHICAL ANALYSIS BY THE PRESIDENT’S COUNCIL ON BIOETHICS (2008), available at http://bioethics.georgetown.edu/pcbe/reports/newborn_screening/index.html (discussing the shift from focusing primarily on what benefits the infant to a “broader conception of benefit”).

46. *See generally* COMM. FOR THE STUDY OF INBORN ERRORS OF METABOLISM, NAT’L ACAD. OF SCIS., GENETIC SCREENING: PROGRAMS, PRINCIPLES AND RESEARCH 228 (1975) (listing unacceptable aims of NBS).

tests to include for NBS, there is little oversight.⁴⁷ Even the ACMG recommendations, which expressly declare that the benefit to the newborn should drive the selection of disease,⁴⁸ include a panel of diseases, not all of which directly or indirectly benefit the newborn.⁴⁹

Several factors have contributed to, and will likely further contribute to, the expansion of NBS, not all of which directly benefits the newborn. Technological advances, such as MS/MS, have contributed to this expansion.⁵⁰ Other technologies, like DNA microarrays, will make it possible to screen for a slew of genetic conditions.⁵¹ With the possibility of ever-cheaper whole genome sequencing, it is not hard to imagine a time, in the not too distant future, when NBS will be expanded to include whole genome sequencing.⁵² Indeed, the National Institutes of Health (NIH) recently funded pilot programs to “explore the promise—and ethical challenges—of sequencing every newborn’s

47. See AGR, *supra* note 8, at 67 (stating that typically state health departments have broad discretion to introduce tests, often with little oversight, which can lead to testing for genetic conditions with little clinical significance).

48. Watson et al., *supra* note 6, at 2S. The approach to selecting diseases awarded points for clear benefits to family and society, as well as points for individual benefits, which were weighted more heavily. TIMMERMAN & BUCHBINDER, *supra* note 8, at 51–52.

49. Specifically the group proposed mandated screening for a panel of twenty-nine conditions and suggested that an additional twenty-five be reported to families. Watson et al., *supra* note 6, at 1S. Because there is no treatment for some of these diseases, they did not meet the standard criteria for NBS. Baily & Murray, *supra* note 32, at 26; Watson et al., *supra* note 6, at 1S; see also Jeffrey R. Botkin et al., *Newborn Screening Technology: Proceed with Caution*, 117 PEDIATRICS 1793, 1796 (2006) (discussing the issues with offering results for a large number of conditions for which limited or no evidence of benefits exist).

50. This would not be the first time that medical diagnostics have been driven as much or more by technology than by need. See Sonia Mateu Suter, *The Routinization of Prenatal Testing*, 28 AM. J.L. & MED. 233, 233 (2002) (“A product of the technology era, genetics has, in a short time, offered vast amounts of information.”).

51. DNA microarrays allow researchers to analyze thousands of active genes at a time, which could allow them to search for huge numbers of genetic disease mutations at one time. *DNA Microarray Technology*, NAT’L HUM. GROWTH RES. INST. (Nov. 15, 2011), <http://www.genome.gov/10000533>.

52. See FRANCIS S. COLLINS, *THE LANGUAGE OF LIFE: DNA AND THE REVOLUTION IN PERSONALIZED MEDICINE* 208 (2010) (“[It is] almost certain . . . that complete genome sequencing will become part of newborn screening in the next few years.”).

genome.”⁵³ This is consistent with the development of personalized medicine and the belief that it is responsible and empowering to get as much medical information as possible.⁵⁴

So far, most of the expansions of NBS have been beneficial, although the data about “long-term clinical outcomes” are limited.⁵⁵ The lives of many children, who might have died years ago because their state did not screen for medium chain acyl-coenzyme A dehydrogenase deficiency (MCADD), for example, have been saved by the introduction of MCADD testing in all states.⁵⁶ Even so, the expansion of NBS is not without costs. The more conditions we screen for, the greater the risk of the inevitable artifacts of any screening program: false negatives, false positives, and clinical and diagnostic uncertainty. False negatives may create false reassurance and slow the process of diagnosis; because pediatricians know that NBS is done for all children, they may assume that the child does not have one of the NBS diseases based on the negative NBS result.⁵⁷

False positives present the opposite problem.⁵⁸ When a child is reported as being positive for one of the NBS conditions,

53. Jocelyn Kaiser, *NIH Studies Explore Promise of Sequencing Babies' Genomes*, SCI. MAG. (Sept. 4, 2013, 2:45 PM), <http://news.sciencemag.org/biology/2013/09/nih-studies-explore-promise-sequencing-babies%E2%80%99-genomes>.

54. See Suter, *supra* note 50, at 233–34 (noting the strong desire to use technology to get as much information as possible, but also cautioning that knowledge can be toxic at times).

55. TIMMERMANS & BUCHBINDER, *supra* note 8, at 184.

56. See Baily & Murray, *supra* note 32, at 23–24 (discussing Mississippi's response to MCADD and the benefits to its newborn population). However, not all deaths due to MCADD have been eliminated with NBS. See TIMMERMANS & BUCHBINDER, *supra* note 8, at 185.

57. False negatives can occur because of failures in the administration of NBS: failure to perform the test properly, to record the results, or simply to test. But false negatives can also occur even if everything is done correctly because NBS is a *screening* test—it is not diagnostic. AGR, *supra* note 8, at 40. False negatives may have become less of a problem in the last five to ten years, but state health departments recognize the possibility of false negatives. ARIZ. DEPT OF HEALTH SERVS., ARIZONA NEWBORN SCREENING PROGRAM: GUIDELINES 42–43 (2010), *available at* <http://www.azdhs.gov/lab/aznewborn/documents/providers/AZ-Newborn-Screening-Provider-Guidelines.pdf> (revised Jan. 2011).

58. False positives may result from errors in the testing process (testing/analysis or reporting), but in general, false positives are an unavoidable consequence of screening for extremely rare disorders. But like false negatives, they are also inevitable artifacts of any screening program. The

the family can experience a great deal of anxiety and confusion. Some studies have shown that false positives can have an adverse effect on the relationship between parent and child, including parents' continued worries about the child's health even after learning that she did not have the condition after all.⁵⁹ In addition, false positives may have a negative health impact on the child by requiring follow-up testing and treatment until it is determined that the child is unaffected; further testing and treatment both pose potential medical risks.⁶⁰ Children who have false positive results are often mislabeled as ill even though they do not display any clinical symptoms.⁶¹

The recent and rapid expansion of NBS panels may also result in the diagnosis of conditions for which there is no treatment, which may create unnecessary stress and anxiety for the family and affect the parent-child relationship. For example, parents may pursue costly treatment odysseys, hoping to find a cure even though no proven treatment exists.⁶² While such information may help parents with future reproductive decision making, this rationale moves NBS away from its stated purpose of benefitting the newborn. Moreover, it undercuts the

incidence of false positives can be quite high. "Some states have a [positive predictive value] of only 3%, meaning that 97% of infants who initially test positive do not actually have the disease." Whelan, *supra* note 18, at 438.

59. See K. Fyrö & G. Bodegård, *Four-Year Follow-up of Psychological Reactions to False Positive Screening Tests for Congenital Hypothyroidism*, 76 ACTA PAEDIATRICA SCANDINAVICA 107, 107, 111 (1987) (finding that a significant portion of families experienced persistent anxiety months and years after false positives); James R. Sorenson et al., *Parental Response to Repeat Testing of Infants with 'False-Positive' Results in a Newborn Screening Program*, 73 PEDIATRICS 183, 185-86 (1984). One study also found that about half of the children demonstrated difficulty adjusting psychologically to the false positives as the mother-child relationship was negatively impacted. Karin Fyrö & Göran Bodegård, *Difficulties in Psychological Adjustment to a New Neonatal Screening Programme*, 77 ACTA PAEDIATRICA SCANDINAVICA 226, 229-31 (1988) (noting, however, that other factors may have played a role in the dysfunction, which were unveiled by the NBS results).

60. Harrell, *supra* note 38, at 847-48 (describing the general concern and her family's experience with a false positive when her son was screened as a newborn).

61. *Id.* at 847 (discussing the effects of a ten to one ratio of false positives to true positives, coupled with a lack of visible symptoms, on parents' decision making, and the fact that false positives create the belief that the child is ill and that it is neglectful not to proceed with additional testing).

62. See Baily & Murray, *supra* note 32, at 28-29.

justification for the mandatory nature of NBS, as we shall see in Part III.B.

Even more complicated issues arise when laboratories make incidental findings of “abnormalities” or clinically ambiguous findings.⁶³ This problem has increased with tandem mass spectrometry, which looks for a group of core conditions by identifying unusually high levels of metabolites related to these conditions.⁶⁴ An artifact of this technology is the incidental identification of elevated levels of certain metabolites, which the laboratory was not even trying to identify,⁶⁵ or the identification of screening values that lie outside the normal range but that do not always clearly correlate with defined disease categories.⁶⁶ These findings can lead to a new kind of diagnostic odyssey, where children become, to use the terminology of Timmermans and Buchbinder, “patients-in-waiting,” who hover “for extended periods of time under medical attention between sickness and health, or more precisely, between pathology and an undistinguished state of ‘normality.’”⁶⁷

Several problems arise when these incidental or diagnostically uncertain findings are made and reported to

63. TIMMERMANS & BUCHBINDER, *supra* note 8, at 12 (“Newborn screening is a technology expected to provide actionable knowledge, yet it generates uncertainty in the clinic . . .”).

64. Baily & Murray, *supra* note 32, at 25 (“Tandem mass spectrometry measures the levels of various metabolites in the blood, and abnormalities in the levels suggest the presence of metabolic disorders.”).

65. TIMMERMANS & BUCHBINDER, *supra* note 8, at 104 (describing the identification of ACADM variants of unknown significance). Indeed, one of the debated aspects of MS/MS is how many of the metabolic variants to report to families. The ACMG proposed that in addition to a core panel of twenty-nine conditions identified through MS/MS, twenty-five others should be disclosed to families. *See supra* note 49. Some countries report only a limited number of conditions identifiable through MS/MS. Clayton, *supra* note 11, at 697 (“Many countries have chosen to report only a limited number of disorders detectable by MS/MS . . .”). The argument for this approach is that, if the family knows about these conditions, they might avoid diagnostic odysseys. In addition, such information might be useful for reproductive decision making, and following such children might help us deepen our understanding of these conditions. These arguments, however, depart from the traditional NBS philosophy by placing societal benefits above the needs of the child. Baily & Murray, *supra* note 32, at 28. On the other hand, not everyone wants such information and there can be harm in receiving ambiguous information or information about conditions for which there is no treatment. *See Clayton, supra* note 11, at 698 (“Some parents simply will not want all these results.”).

66. TIMMERMANS & BUCHBINDER, *supra* note 8, at 65.

67. *Id.*

parents. The child might be stigmatized as a “sick child” before symptoms develop, if they ever will. This label has been shown to have a harmful effect on the parent-child relationship and on the family as a whole.⁶⁸ Indeed, in some cases, the child might never become clinically affected by the abnormal levels of the metabolite or the mutation.⁶⁹ There may be a considerable time lag before physicians can determine whether high metabolites or certain mutations are clinically significant, hence the phrase “patients-in-waiting.”

Timmermans and Buchbinder’s ethnographic study of a genetics clinic describes the complexities and anxieties that such diagnostic uncertainties present and the ways in which entire families are affected during this period.⁷⁰ If families learn of these findings, they might embark on treatment odysseys, investing significant money and time in search of treatments that may not exist or that are unproven. Sometimes the heightened vigilance that parents exhibit during this period is difficult to “tone down” once it becomes clear that the child is not clinically affected.⁷¹ NBS programs may also spend added dollars to report and follow up on conditions for which treatments may not exist. It has also presented challenges for clinicians who have to contend with the fact that expanded screening has “identified more patients than anticipated,” most of whom are asymptomatic, and which requires a collective

68. See *supra* note 59.

69. In fact, with little knowledge of the disease’s natural history, it is difficult to know the rate of false positives or negatives or even, at times, to determine whether there is a false positive or negative.

70. TIMMERMANS & BUCHBINDER, *supra* note 8, at 65–96 (describing the full experience of “patients-in-waiting” and their families).

71. *Id.* at 88 (“When, after time passed, the baby remained fine, clinicians sometimes had trouble getting the parents to tone down their level of vigilance.”); *id.* at 91 (“[W]hile geneticists could be ready to let the condition fade away, family members could nevertheless perpetuate the medicalization of their child.”); *id.* at 226 (“The most striking emotion we observed in the clinic was anxiety, but parents also expressed shame, anger, and sadness.”). Even so, “nearly all of the families in [Timmermans and Buchbinder’s] study regarded the screening program favorably.” As one parent said, “[w]e would rather go through 10 weeks of the hell we went through than a lifetime of having a special needs child without having the opportunity to know from day one or day five.” *Id.* at 219.

learning process and the development of new knowledge to determine who is truly affected.⁷²

If NBS ultimately includes whole genome sequencing, similar issues will arise on an even greater scale. We are unlikely to fully understand for some time the clinical implications of many mutations, let alone the complex interactions of different mutations within a particular genome and environment. In many instances, it will be difficult to determine whether a genetic variant is likely to have a significant clinical impact, or what the degree or timing of such impact would be.⁷³ As a result, whole genome sequencing would likely provide a great deal of data of limited value, which could increase parental anxiety and confusion.

Although the *raison d'être* for NBS was to promote the wellbeing of newborns, some of the expansions of NBS can only be justified by other considerations, such as allowing parents to make better informed reproductive decisions and benefiting society by allowing us to better understand the conditions. The more these other rationales are used to justify expansions of NBS, the more we should question whether screening infants without the consent of parents can be justified. I turn now to an explanation for the enduring lack of consent in NBS before discussing the issues of consent that arise with respect to the storage and dissemination of newborn samples for research and other uses.

II. THE LACK OF CONSENT IN NBS

Consent has long been absent in NBS, making it in essence a mandatory screening program. Recently, the public and scholarly communities have focused largely on the lack of consent with respect to the storage and future uses of DBS. But although the lack of consent with respect to the collection of blood samples and screening itself has not been challenged as strongly, there are reasons to question the presumption against requiring consent for NBS itself. I begin by describing the general rationales for lack of consent in NBS and then turn to the practices with respect to storage and future uses before

72. *Id.* at 94–95; see *id.* at 119 (“[E]xpanded newborn screening has prompted a tremendous knowledge explosion about rare metabolic conditions.”).

73. Clayton, *supra* note 11, at 698.

offering my recommendations, in Part III, regarding consent in these two areas.

A. CONSENT (OR LACK THEREOF) FOR NEWBORN SCREENING ITSELF

NBS is quite unusual in being one of the few areas where the state can require medical testing of an individual or child without affirmative consent.⁷⁴ Even so, the mandatory nature of NBS has long been well accepted with only minimal criticism.⁷⁵ Although most states do not require affirmative parental consent for newborn screening, there is some variability with respect to what amounts to presumed consent. The majority of states allow parents to opt out, although the reasons they allow differ. Some will only allow parents to refuse for religious reasons.⁷⁶ Many will allow parents to opt out for any reason.⁷⁷ At one extreme, NBS is mandatory without exception.⁷⁸ One state actually imposes criminal penalties for refusing to undergo NBS.⁷⁹ Even in states where there is an opt-out provision, there is serious doubt as to whether parents truly have an opportunity to refuse in these jurisdictions,⁸⁰ making

74. Parents are generally allowed to refuse medical treatment or testing on behalf of their child, unless their decision puts a child at grave risk. See Andrews, *supra* note 2, at 59 (“Only when their decisions put their children at grave risk are parental decisions overridden by the state.”).

75. See, e.g., Clayton, *supra* note 11, at 697 (discussing the rapid development of the screening programs and stating that they “were almost always mandatory, in response to advocacy by geneticists and parents”).

76. See TENN. CODE ANN. § 68-5-403 (2013) (allowing parents to opt out of testing or medical treatment if they file a written statement that states such tests or treatment conflict with their “religious tenets and practices”); WIS. STAT. ANN. § 253.13(3) (West 2010) (stating that the statute shall not apply “if the parents or legal guardian of the child object thereto on the grounds that the test conflicts with their religious tenets and practices”).

77. See, e.g., FLA. STAT. ANN. § 383.14(4) (West 2007) (“The provisions of this section shall not apply when the parent or guardian of the child objects thereto.”); N.M. STAT. ANN. § 24-1-6(A) (West 2011) (stating that parents, after being informed of the reasons for the tests, may waive the requirements for the tests in writing).

78. MICH. COMP. LAWS ANN. § 333.5431 (West 2001); MONT. CODE ANN. §§ 50-19-201 to -211 (2013); NEB. REV. STAT. §§ 71-519 to -524 (2009); S.D. CODIFIED LAWS §§ 34-24-17 to -25 (2011); W. VA. CODE §§ 16-22-1 to -6 (2010).

79. S.C. CODE ANN. § 44-37-30(G) (1991).

80. Ruth Faden et al., *A Survey to Evaluate Parental Consent as Public Policy for Neonatal Screening*, 72 AM. J. PUB. HEALTH 1347, 1347 (1982) (describing the screening as “compulsory for all practical purposes”).

the provision “opt-out” more in name than practice. Only two states require affirmative parental consent.⁸¹

Not only is a requirement of consent for NBS rare, but parents are often woefully uninformed about NBS. Often states provide limited information about the nature of NBS testing⁸² or that there is an option to opt out (when there is such an option).⁸³ Sometimes parents are not even informed that the child will be tested.⁸⁴ If a child tests positive through NBS, parents often do not learn that the newborn *screening* results are not diagnostic and that there may be false positives or negatives.⁸⁵ And many are not adequately educated about the nature of the condition or offered genetic counseling, even when the child tests positive.⁸⁶

81. D.C. CODE §§ 7-831 to -840 (LexisNexis 2012); WYO. STAT. ANN. §§ 35-4-801 to -802 (2013). In the last few years, Maryland switched from its opt-in, informed consent approach, to an opt-out approach. See MD. CODE REGS. 10.52.12.07 (2013); Rachel L. Schweers, *Newborn Screening Programs: How Do We Best Protect Privacy Rights While Ensuring Optimal Newborn Health?*, 61 DEPAUL L. REV. 869, 891 n.130 (2012). The rationale for this change was to bring testing in line with the national Newborn Screening Taskforce, to be like the vast majority of states, and to lighten the paperwork burden on hospitals and providers because parental refusal is so rare. MD. DEP'T OF HEALTH & MENTAL HYGIENE, 2008 LEGISLATIVE REPORT: SHOULD A COORDINATED STATEWIDE SYSTEM FOR SCREENING NEWBORN INFANTS BE APPLIED TO ALL NEWBORN INFANTS IN MARYLAND? 2-3 (2008).

82. See Schweers, *supra* note 81, at 869 (discussing the lack of knowledge about screening policies amongst health care providers, and the need to initiate a discussion in order to address concerns).

83. See, e.g., MD. CODE REGS. 10.52.12.07 (2013) (providing an example of an opt-out regulation); Rachel Grob, *Parenting in the Genomic Age: The 'Cursed Blessing' of Newborn Screening*, 25 NEW GENETICS & SOC'Y 159, 159, 163 (2006).

84. AGR, *supra* note 8, at 67 (stating that at this point, most parents receive brochures or some general information at the time of screening, although in many cases this is very thin, token information); see Terry C. Davis et al., *Recommendations for Effective Newborn Screening Communication: Results of Focus Groups with Parents, Providers, and Experts*, 117 PEDIATRICS S326 (Supp. May 2006) (providing that one-third of patients in a study in California never received NBS materials from their prenatal providers even though California requires them to provide patients with such information); Lisa A. Faulkner et al., *The Newborn Screening Educational Gap: What Prenatal Care Providers Do Compared with What Is Expected*, 194 AM. J. OBSTETRICS & GYNECOLOGY 131 (2006).

85. AGR, *supra* note 8, at 65, 67.

86. See Clayton, *supra* note 11, at 697 (“While some people may value this information, other parents who specifically chose not to have carrier screening for themselves may be less pleased when they involuntarily learn their carrier status from their child’s newborn screen.”).

NBS laws and practices go very much against legal and ethical norms in the United States, which recognize an individual's right to choose whether to undergo medical treatment or testing and to refuse treatment even when it can result in death.⁸⁷ Not only is consent required for most medical interventions and treatments,⁸⁸ generally consent must be *informed*.⁸⁹

There is considerable irony in the fact that parental decision making and education are so limited with NBS since it is essentially a form of *genetic* screening. Mandatory genetic testing is extremely unusual,⁹⁰ in large part because a strong consensus has existed for some time that genetic screening programs should not be compulsory and should involve informed consent.⁹¹ After all, genetics and especially genetic counseling are among the disciplines in medicine most deeply committed to individual autonomy in medical decision making and informed decision making for genetic testing.⁹²

87. The Supreme Court, in *Cruzan v. Mo. Dep't of Health*, 497 U.S. 261, 269–70 (1990), discussed the long common law tradition of protecting bodily integrity through battery actions and the informed consent doctrine, which is now “firmly entrenched in American tort law.” Based on this common law tradition, the court inferred that a competent person has a constitutionally protected right to refuse lifesaving hydration and nutrition. See *Winston v. Lee*, 470 U.S. 753, 753, 766 (1985) (holding the surgical removal of a bullet from a defendant's body was an unreasonable search violating the Fourth Amendment); *Rochin v. California*, 342 U.S. 165, 172–73 (1952) (holding that evidence obtained through the forceful use of a stomach pump violated the Due Process Clause).

88. Treating a patient or imposing some medical intervention without a patient's consent could easily be the basis for a battery claim. BARRY R. FURROW ET AL., *HEALTH LAW: CASES MATERIALS AND PROBLEMS* 357–58 (5th ed. 2004).

89. *Id.* at 357.

90. Andrews, *supra* note 2, at 58 (providing that some unfortunate exceptions to this rule have included the mandatory testing for carriers of the gene for sickle cell anemia); see AGR, *supra* note 8, at 40–42.

91. Faden et al., *supra* note 80, at 1347–48 (describing various policy committees that have expressly rejected “public health justification[s] for mandatory [genetic] screening” and noting that “[t]he Genetic Disease Title of Public Law 94-278, which provides assistance in the establishment of genetic testing and counseling programs, requires that the ‘participation by an individual in any program or portion thereof under this part shall be wholly voluntary’”).

92. See TIMMERMANS & BUCHBINDER, *supra* note 8, at 19 (noting how inconceivable it seems in “an era infused with bioethical concern about patient autonomy and genetic discrimination” to screen “the overwhelming majority of

NBS is not, however, the only example in which the state has made medical decisions on behalf of individuals. The state has intervened either to protect the well-being of the public or the individual himself. In *Jacobson v. Massachusetts*, for example, the Supreme Court upheld the state's right to mandate its citizens to be vaccinated against smallpox.⁹³ The Court reasoned that vaccinating an individual against his will did not violate the individual's liberty interests.⁹⁴ This was so because a "community has a right to protect itself against an epidemic of disease which threatens the safety of its members,"⁹⁵ as long as the means of doing so are "reasonably required for the safety of the public."⁹⁶ The court located the state's right to compel vaccination within its police powers because it protects the public health⁹⁷ by preventing the spread of highly contagious smallpox.⁹⁸ The state has also exercised its police powers to impose medical treatment against a person's will when someone has been deemed mentally ill and a threat to others.⁹⁹ In both instances, the government intervenes to prevent one individual from threatening physical danger or harm to another.¹⁰⁰ In spite of possessing these potentially broad powers, the states have tended to be fairly limited in using them.¹⁰¹

infants . . . for *genetic* conditions without informed consent"); Suter, *supra* note 50, at 242–43.

93. *Jacobson v. Massachusetts*, 197 U.S. 11, 38–39 (1905).

94. *Id.* at 27.

95. *Id.*

96. *Id.* at 28.

97. *Id.* at 24–25 ("The authority of the State to enact this statute is to be referred to what is commonly called the police power—a power which the State did not surrender when becoming a member of the Union under the Constitution. [T]his court . . . has distinctly recognized the authority of a State to enact quarantine laws and 'health laws of every description;' According to settled principles, the police power of a State must be held to embrace, at least, such reasonable regulations established directly by legislative enactment as will protect the public health and the public safety.").

98. *Id.* at 35 (finding "strong support" for the view that vaccination is an effective "means of protecting a community against smallpox").

99. *Washington v. Harper*, 494 U.S. 210, 236 (1990).

100. Ellen Wright Clayton, *Screening and Treatment of Newborns*, 29 HOUS. L. REV. 85, 126 (noting that the police power "has historically been invoked only to protect others from physical harm").

101. Andrews, *supra* note 2, at 54 (noting, for example, that the government has not tended to track people down with infectious diseases, quarantined them, or forced them to undergo treatment, but observing that in some limited

The mandatory nature of NBS has been justified by these police powers because NBS is touted as a public health effort.¹⁰² In fact, however, NBS does not neatly fit into this model. NBS screening is conducted primarily to prevent harm to the *individual* who is being screened, rather than to prevent harms to others.¹⁰³ To be sure, identifying a child's metabolic disorder in time to provide treatment can minimize suffering for the family overall, reduce societal health care costs, and expand families' reproductive options. These rationales, however, are not typically what we think of as public health efforts of the sort that justifies the police powers. Of course, if we conceive of the public health more broadly as the public good, then this justification is more powerful.

Even so, the better rationale for the mandatory nature of NBS is the doctrine of *parens patriae*, which allows the state to limit a person's liberty to protect the individual.¹⁰⁴ The basic principle of this doctrine is to preserve human life.¹⁰⁵ Although there is a common law and constitutional presumption that parents have the right to make medical decisions on behalf of their children,¹⁰⁶ the state can intervene if parental decisions constitute abuse or neglect.¹⁰⁷ Classic cases in which the state

cases people have been required to be tested to HIV infection if convicted of certain crimes).

102. Faden et al., *supra* note 80, at 1347.

103. Whelan, *supra* note 18, at 435 (describing the police powers as infringing "on individual rights primarily to protect the public from other individuals' actions or behaviors").

104. STEVEN OLSON & ADAM C. BERGER, INST. OF MED., CHALLENGES AND OPPORTUNITIES IN USING RESIDUAL NEWBORN SCREENING SAMPLES FOR TRANSLATIONAL RESEARCH: WORKSHOP SUMMARY 7 (2010) ("Newborn screening programs are authorized through the legal doctrine known as *parens patriae*, which gives the state the right to assume certain roles of parents based on benefits to the child and to society as a whole."); Clayton, *supra* note 100, at 126.

105. *Newmark v. Williams*, 588 A.2d 1108, 1115 (Del. 1991).

106. *Id.* at 1115–16 ("[T]he common law recognizes that the only party capable of authorizing medical treatment for a minor in 'normal' circumstances is usually his parent or guardian."); e.g., *Santosky v. Kramer*, 455 U.S. 745, 753 (1982); *Parham v. J.R.*, 442 U.S. 584, 602 (1979).

107. *Newmark*, 588 A.2d at 1116 ("[T]he State can intervene in the parent-child relationship where the health and safety of the child and the public at large are in jeopardy."); BARRY R. FURROW ET AL., *BIOETHICS: HEALTH CARE LAW AND ETHICS* (6th ed. 2008); Lainie F. Ross, *Predictive Genetic Testing of Children and the Role of the Best Interest Standard*, 41 J.L. MED. & ETHICS 899, 901 (2013) (noting that in the United States, as compared with the United Kingdom, the best interest standard tends to give "considerable deference to

has successfully intervened include parental decisions to withhold lifesaving transfusions or chemotherapy.¹⁰⁸

The *parens patriae* justification for NBS is the urgent need for early diagnosis of conditions for which early treatment can reduce morbidity and mortality. It is further supported by the fact that the risks of testing and treatment are generally minimal. Thus, the argument goes, the state must intervene because parental refusal to test for various inborn errors of metabolism and other serious conditions could be potentially life threatening or seriously debilitating by preventing an affected child from being diagnosed during the newborn period. The underlying presumption is that without a mandate, parents will refuse to participate in NBS, leaving children undiagnosed and therefore untreated for treatable conditions.¹⁰⁹ Because NBS fits better within a medical model—where the focus is the risk/benefit calculus with respect to the individual—than a public health model, the *parens patriae* justification is more appropriate than the police powers rationale.

Even so, as some scholars pointed out in the earlier years of NBS, and as is even truer now as NBS expands, the *parens patriae* rationale is somewhat questionable for many reasons. First, as I discuss in Part III, empirical data challenge the presumption that a mandate is necessary to ensure that newborns are screened. Second, definitive treatments are not available for all of the conditions identified;¹¹⁰ a problem that

childrearing decisions made by parents or guardians, with state intervention generally confined to instances of abuse or neglect”) (citing Lainie F. Ross et al., *Technical Report: Ethical and Policy Issues in Genetic Testing and Screening of Children*, 15 GENETICS MED. 234, 236 (2013)); June Carbone, Legal Applications of the “Best Interest of the Child” Standard: Judicial Rationalization or a Measure of Institutional Competence 10 (unpublished manuscript) (on file with author) (noting that, although “the treatment of children starts with deference toward parental preferences” parental rights “are not absolute”).

108. Andrews, *supra* note 2, at 59; Seema Shah, *Does Research with Children Violate the Best Interests Standard? An Empirical and Conceptual Analysis*, 8 NW. J.L. & SOC. POL’Y 121, 125, 156 (2013) (finding that courts ordered blood transfusions over parental objections in all but two cases).

109. See PRESIDENT’S COMM’N FOR THE STUDY OF ETHICAL PROBLEMS IN MED. & BIOMEDICAL RESEARCH, A REPORT ON THE ETHICAL, SOCIAL, AND LEGAL IMPLICATIONS OF GENETIC SCREENING, COUNSELING, AND EDUCATION PROGRAMS (1983), available at <http://kie.georgetown.edu/nrcbl/documents/pcemr/geneticscreening.pdf>.

110. See TIMMERMANS & BUCHBINDER, *supra* note 8, at 183 (describing how the genetics clinic saw many “symptomatic patients who did not seem to

will likely grow as the panel of diseases expands. Third, in some cases interventions can save lives, but the children still “face significant developmental delays, frequent hospitalizations, and serious risks of mortality.”¹¹¹ Sometimes, newborn screening may not occur in time to protect those at greatest risk.¹¹² Given the ongoing morbidity and mortality for many children screened positive, some scholars predict that “the health payoff of screening is likely to be lower than the number of true positive might otherwise imply.”¹¹³

Even when treatments are available, the state often does not actually provide treatment to the affected children; the programs merely provide families with the information to seek out treatment.¹¹⁴ The success of newborn screening in preventing disease depends largely on day-to-day efforts to manage the conditions and “the ability [of families] to tap into available medical services and social resources,” which is as much a function of socioeconomic factors as anything else.¹¹⁵ As

improve” and how “for the most severe disorders associated with the worst outcomes . . . newborn screening [is] unlikely to make a difference in outcomes”); Clayton, *supra* note 11, at 698 (“Other disorders are identified for which there is no effective therapy.”).

111. TIMMERMANS & BUCHBINDER, *supra* note 8, at 179; *id.* at 184 (“[S]ome children did poorly despite the advance knowledge provided by newborn screening.”); *id.* at 189 (describing conditions for which early interventions “could prevent only some negative consequences”).

112. *Id.* at 162 (“[B]etween July 2005 and April 2009, 62 screen positive infants died in California before follow-up care could be started in a metabolic center.”); *id.* at 180 (“In some cases, newborn screening results arrived too late, after a child had already sustained a devastating metabolic crisis and permanent brain damage.”).

113. *Id.* at 216.

114. See Burke et al., *supra* note 8, at 152 (“Although most states provide informational brochures, many parents are unaware that their infant has been tested unless they are notified of a positive result.”); see also R. Rodney Howell, *We Need Expanded Newborn Screening*, 117 PEDIATRICS 1800, 1802 (2006) (“The facilities vary widely for such follow-up around the country, and it is incumbent on the state programs to work in their regions to provide follow-up support in terms of funding and organization.”). In such cases, we may simply be labeling more children as ill without actually providing much clinical benefit to many of these children, especially if parents are not adequately educated or cannot afford the treatment. Moreover, it exacerbates concerns about whether the resources devoted to NBS could be better used to address the urgent health care needs of many children that have still not been met.

115. TIMMERMANS & BUCHBINDER, *supra* note 8, at 195; see *id.* at 170, 194–210 (describing the effects of insurance, access to transportation, language, education and bureaucratic barriers on parents’ abilities to manage their children’s metabolic conditions).

a result, the state's efforts work only partially toward the goal of eliminating the deleterious effects of the diseases, leading some to question whether the true motivation for mandatory NBS is actually the well-being of the child.¹¹⁶

Finally, even if the state is motivated primarily by the well-being of each child, it is not clear that the risks are great enough to justify state intervention. While many of the NBS conditions could lead to grave, even life-threatening, harm if undetected, these conditions are extremely rare. This means that the probability that any one child who is not tested through NBS will suffer a grave or life-threatening illness by failing to undergo NBS is statistically quite low, although clearly the magnitude of harm could be quite great.¹¹⁷ In contrast, both the probability and magnitude of harm (death or serious debilitation) in failing to provide blood transfusions or chemotherapy, for example, will often be considerable.¹¹⁸ As Professor Lori Andrews has noted, the risks of refusing NBS screening "is far less than the risks inherent in many other decisions that parents are routinely allowed to make," such as allowing their children to play on high school sports teams.¹¹⁹ Moreover, the probability of false positives is quite high; the rate of false to true positives can be as high as, or higher than, ten to one.¹²⁰ As noted above, false positives are often not inconsequential. They can potentially lead to psychological,

116. See Burke et al., *supra* note 8, at 151 ("However, growing test capacity has led to calls to expand not only the number of disorders screened for but also the goals of newborn screening."). "In the past, . . . infrastructural problems and healthcare costs had tempered enthusiasm for expanding newborn screening, but the separation of the scientific issues from those affecting healthcare delivery had the effect of decontextualizing the viability of screening." TIMMERMANS & BUCHBINDER, *supra* note 8, at 55.

117. NBSTF, *supra* note 28, at 414.

118. Andrews, *supra* note 2, at 60. Of course, the calculus can often be complicated by other factors. In *Newmark v. Williams*, 588 A.2d 1108 (Del. 1991), for example, the Delaware Supreme Court ruled that it was not neglectful for parents to refuse chemotherapy treatment for their three-year-old child, who suffered from "an aggressive and advanced form of pediatric cancer," because the proposed treatment was "highly invasive, painful, involved terrible temporary and potentially permanent side effects, posed an unacceptably low chance of success, and a high risk that the treatment itself would cause his death." *Id.* at 1109–10, 1118.

119. Andrews, *supra* note 2, at 60.

120. See Harrell, *supra* note 38, at 847 ("Given such real life consequences of a false positive and that the rate of false positives to true positives is as high as 10 to 1 (or higher) for many of the newborn screens . . .").

relational, and even physical harms from follow-up testing and/or treatment.¹²¹ While the magnitude of such harms is lower than failing to detect the condition, the probability of such harms is likely much greater than the probability of identifying the conditions screened for.

Despite these concerns and a general presumption against compulsory genetic screening in virtually every other context, mandatory NBS remains the norm, even when opportunities arise to change the nature of this institution.¹²² As I argue in Part III, it may be time to rethink the role of consent in NBS, particularly with the potential of NBS to expand even further and as NBS samples are used more widely in research, as the next section shows. In addition, consent requirements may go far in promoting the NBS education that parents, providers, and scholars believe is woefully inadequate.¹²³

B. STORAGE AND SECONDARY USES OF NBS SAMPLES

Once the newborn blood spots are analyzed for the various NBS conditions, residual blood remains in the form of DBS.¹²⁴ Increasingly, states retain these samples for future uses, although the retention time varies significantly from state to state. Some states have provisions to retain samples for only one to four weeks, some for months, some for years, some for decades, and others indefinitely.¹²⁵ Often these samples are stored with identifying information.¹²⁶

121. *See id.* at 847–48.

122. MICH. COMM'N ON GENETIC PRIVACY & PROGRESS, FINAL REPORT AND RECOMMENDATIONS 4, 33 (1999). This Author was a member of the Michigan Commission on Genetic Privacy and Progress. Despite many months of deliberation, a majority of the committee voted to retain mandatory NBS, with an opt-out provision, although efforts were made to ensure that parents were to receive information about NBS.

123. Sandra J. Carnahan, *Biobanking Newborn Bloodspots for Genetic Research Without Consent*, 14 J. HEALTH CARE L. & POL'Y 299, 303, 322–25 (2011) (“Although educational pamphlets about the screening program are typically distributed to the parent, guardian, or managing conservator . . . state statutes, almost universally, do not require NBS programs to obtain the informed consent of the newborn’s parent prior to extracting the blood sample.”).

124. *Id.* at 301.

125. *See* Michelle H. Lewis et al., *State Laws Regarding the Retention and Use of Residual Newborn Screening Samples*, 127 PEDIATRICS 703, 704 (2011) (“A total of 40% of state public health laboratories have reported retaining DBS for at least 1 year.”); Richard S. Olney et al., *Storage and Use of Residual Dried*

Although the samples are analyzed right away for NBS, there are several reasons states might want to retain the samples for months or even years. Many of these reasons are related to the underlying purpose of NBS. For example, the retention of these samples—along with contact information—is necessary for follow-up and to ensure that there will be appropriate intervention for an affected child.¹²⁷ In addition, labs may need to perform repeat tests to make a confirmatory diagnosis or to reassure families if there is a false positive.¹²⁸ Less directly related to NBS testing per se, but still connected to the public health aspects of NBS, is the retention of blood spots for quality assurance testing and to monitor the prevalence of various conditions in the state.¹²⁹ NBS samples may also be helpful for post-mortem diagnosis; for example, when trying to establish whether a genetic condition was related to a child's death.¹³⁰

Increasingly, states are interested in long-term retention of these blood spots for purposes not directly related to NBS. Some states and/or other countries retain neonate blood spots for non-medical or non-research uses, such as identification in kidnappings or deaths.¹³¹ NBS samples have also been used for paternity testing¹³² and could potentially be used for the identification of criminals.¹³³

Blood Spots from State Newborn Screening Programs, 148 J. PEDIATRICS 618, 619 fig. (2006).

126. Carnahan, *supra* note 123, at 320 (observing that a 2002 study found that thirty-four out of thirty-six NBS program studies stored the DBS with identifying information).

127. *Id.* at 304.

128. NBSTF, *supra* note 28, at 414.

129. *Id.* at 404, 413, 415–16 (suggesting that knowing about the prevalence of various conditions is important not only for better understanding of the condition, but also for determining the optimal allocation of resources).

130. Linda Kharaboyan et al., *Storing Newborn Blood Spots: Modern Controversies*, 32 J.L. MED. & ETHICS 741, 742 (2004).

131. MICH. COMM'N ON GENETIC PRIVACY & PROGRESS, *supra* note 122, at 28.

132. In New Zealand, the High Court ordered the Auckland Health Services to provide the blood sample of a man's child for paternity testing that he sought after the baby died. *H v G* [M/1686/98] 1999, *upheld in H v G* (1999) 18 FRNZ 572 (HC).

133. Some have called for universal DNA databanking for criminal forensic purposes. See, e.g., D.H. Kaye & Michael E. Smith, *DNA Databases for Law Enforcement: The Coverage Question and the Case for a Population-Wide Database*, in *DNA AND THE CRIMINAL JUSTICE SYSTEM: THE TECHNOLOGY OF*

In addition, these blood spots, like most pathology samples, are a treasure trove for researchers because they are a valuable national repository of genetic material. As genetic technology develops,¹³⁴ the blood spots are an especially rich source of research material: they are stable over time, they constitute an unbiased collection of samples since they represent the entire population,¹³⁵ and they can potentially be linked to basic demographic information.¹³⁶ As one author notes, “[n]ewborn screening initially began as a population health endeavor but is rapidly becoming a resource for population research.”¹³⁷ Newborn blood samples have been used in research and shared with investigators since the 1980s,¹³⁸ sometimes with identifying information.¹³⁹

Only recently have professional groups begun to consider seriously how to handle the problems of storage and secondary uses of the samples.¹⁴⁰ Very few states have specific regulations

JUSTICE 247, 269–71 (David Lazer ed., 2004) (arguing that universal DNA databases would eliminate the disproportionate minority representation in forensic databases). NBS blood spots would offer an easy way to achieve this goal.

134. “Optimal storage conditions” for these samples are less crucial for genetic analysis than for other kinds of biochemical analysis. NBSTF, *supra* note 28, at 415.

135. Nanette Elster, *Future Uses of Residual Newborn Blood Spots: Legal and Ethical Considerations*, 45 JURIMETRICS 179, 180 (2005); Kharaboyan et al., *supra* note 130, at 745.

136. NBSTF, *supra* note 28, at 415 (noting, however, that because these bloodspots “will not be linked to clinical data on the children” their “potential utility . . . will need to be carefully evaluated”).

137. Elster, *supra* note 135, at 189.

138. See *Innocent Blood: Use of Newborn Heel Sticks Spurs Legal Challenges*, IRB ADVISOR (AHC Media, Atlanta, Ga.), Dec. 1, 2009 [hereinafter *Innocent Blood*] (noting that many states used them to determine things like the prevalence of HIV infections, prenatal exposure to heavy metals, frequencies of certain genes); Michelle Lore, *Is the Minnesota Department of Health Violating Privacy Laws*, MINN. LAW., Nov. 30, 2009 (stating that since the end of 2008, 52,519 NBS samples from the state of Minnesota had been used for research).

139. Elizabeth Cohen, *The Government Has Your Baby’s DNA*, CNN (Feb. 4, 2010), <http://www.cnn.com/2010/HEALTH/02/04/baby.dna.government/> (noting that a study in Minnesota found that “more than 20 scientific papers have been published in the United States since 2000 using newborn blood samples”).

140. NBSTF, *supra* note 28, at 389 (recommending that each state develop and implement policies for retention of residual DBS, educate parents regarding the storage and uses, and develop model consent forms and information materials for parents); Brad Therrell et al., *Briefing Paper: Considerations and Recommendations for a National Policy Regarding the*

governing what kind of future uses the samples may be put to or requiring that parents be notified of or give consent for such uses.¹⁴¹ North Dakota, for example, does not require specific consent, stores the samples indefinitely, and permits the use of samples for “medical, psychological or sociological research.”¹⁴² Indeed, because many parents do not realize that their child has been screened for various diseases, they are unaware of the possibility that a blood sample from their newborn may be stored in state health departments for potentially long periods of time and possibly shared with others for uses unrelated to NBS.¹⁴³

The laws in a few states are an exception to this rule. In May of 2009, while the first Texas lawsuit challenging the state’s practice of storing and using newborn samples for undisclosed research was pending,¹⁴⁴ the Texas Legislature amended its NBS laws to require parents and guardians to be informed that samples were being collected and would be stored indefinitely for potential research purposes.¹⁴⁵ Parents, or children upon reaching adulthood, can now request to have the

Retention and Use of Dried Blood Spot Specimens After Newborn Screening, RESOURCE REPOSITORY (Aug. 26, 2009), <http://resourcerepository.org/documents/1681/briefingpaper:considerationsandrecommendationsforanationalpolicyregardingtheretentionanduseofdriedbloodspotspecimensafternewborns/>; see *APHL Position/Policy Statement: Residual Newborn Screening (NBS) Specimens*, APHL (2005), http://www.aphl.org/policy/Documents/residual_newborn_screening_specimens.pdf (suggesting that retention of DBS is important for laboratory quality assurance practices and can also be useful for research among other things); see also AM. COLL. OF MED. GENETICS, STANDARDS AND GUIDELINES FOR CLINICAL GENETIC LABORATORIES (2008) (finding it critical, if states do not retain DBS, for parents to have the option to have their children’s DBS included in a national repository).

141. Lewis et al., *supra* note 125, at 703, 705, 707 (providing that “thirteen states specify the purposes for which DBS may be used,” eight states require parents to be notified of the retention of DBS, and three require “parents to be informed” so that they can request destruction of the DBS). The United States is not the only country where samples are also stored for long periods of time. See Kharaboyan et al., *supra* note 130, at 742–43 (describing practices in Australia, Canada, Denmark, France, New Zealand, and the United Kingdom).

142. Whelan, *supra* note 18, at 428.

143. See generally TEX. HEALTH & SAFETY CODE ANN. §§ 33.0111–.0112 (West 2010) (showing the ability of a state to carry out such activities with DBS).

144. See *supra* note 18 and accompanying text.

145. See *Higgins v. Tex. Dep’t of Health Servs.*, 801 F. Supp. 2d 541, 544–45 (W.D. Tex. 2011).

samples destroyed within sixty days¹⁴⁶—essentially an opt-out-of-research approach. The lawsuit was settled once the State of Texas agreed to destroy over five million coded newborn samples,¹⁴⁷ which had been stored indefinitely for possible research without parental consent.¹⁴⁸

Minnesota also has a limited opt-out provision, allowing parents to refuse NBS itself or to request the destruction of test results and samples following screening.¹⁴⁹ Even so, the Minnesota Supreme Court ruled in favor of parents who sued the state for storing and authorizing public health research on newborn samples on the grounds that these practices violated Minnesota's genetic privacy law.¹⁵⁰ Although the court construed the NBS statutes to be "an express exception to the Genetic Privacy Act," the storage, dissemination, and use of the samples were not expressly authorized and therefore violated the privacy statute.¹⁵¹ As a result of this decision, NBS samples in Minnesota were not available for research or public health studies. Recently, however, the Minnesota House of Representatives and the Minnesota Senate passed bills that would change this. If these bills become law, NBS samples would be available for research, unless parents or the child,

146. *Id.* at 545.

147. Mary Ann Roser, *Samples of Newborns' Blood to Be Destroyed*, AUSTIN AM. STATESMAN, Dec. 23, 2009, at A1 (providing that the state decided that trying to seek consent from all of those parents was a worse option than simply destroying all of the samples). The samples were not identifiable, but because they are coded, a link exists that could be used to identify the child. *Id.*

148. Cohen, *supra* note 139 (noting that in other states it may be very difficult to convince the state to destroy your baby's archived blood sample). A class action filed late 2010 in Texas, also alleging that the state had stored DBS for the purposes of undisclosed research, was dismissed as moot because there was no evidence that the parties' newborn samples were actually used or distributed for research. *Higgins*, 801 F. Supp. 2d at 545, 554.

149. MINN. STAT. § 144.125 (2012); Lore, *supra* note 138 (explaining that absent parents opting out, the NBS test results may become public health data). In Minnesota, for example, the department of health has a contract with the Mayo Clinic for analysis of NBS samples, which allows the Clinic to "keep the samples indefinitely if there is no request for their destruction." *Id.* The samples are not identifiable, although they are coded, and therefore could potentially be linked to the individual. Kharaboyan et al., *supra* note 130, at 744.

150. MINN. STAT. § 13.386 (2013); *Bearder v. Minnesota*, 806 N.W. 2d 766, 776 (Minn. 2011); Lore, *supra* note 138 (stating that Minnesota has been storing the samples since 1997); *Innocent Blood*, *supra* note 138.

151. *Bearder*, 806 N.W. 2d at 776.

over the age of eighteen, opt out, which they may do at any time.¹⁵²

Oklahoma and Michigan require more than the right to opt out. The Oklahoma Legislature recently enacted a provision that requires “express parental consent” for storage, dissemination, and use of a newborn’s DNA.¹⁵³ Michigan, after seeking input from researchers, ethicists, community groups, and the state health department’s institutional review board, created a specific repository for future research that would require affirmative, informed consent from parents.¹⁵⁴ This approach keeps the research uses of newborn samples separate and distinct from NBS itself, which remains mandatory.¹⁵⁵

As these lawsuits and this legislation suggest, many secondary uses of DBS raise ethical and even legal concerns, particularly when the uses are not related to the purposes for which the samples were originally collected.¹⁵⁶ Particularly salient are the threats to privacy and confidentiality.¹⁵⁷ In addition, questions of autonomy and research ethics come into play because the newborns potentially become research subjects via their Guthrie cards.¹⁵⁸ Contemporary practices with NBS raise pressing questions as to whether consent must be secured for storage and secondary uses of NBS samples, and if so what kind of consent—general consent for research, or specific, informed consent for a particular use.¹⁵⁹

152. *Minnesota House Passes Newborn Screening Bill*, GENOME WEB (May 2, 2014), <http://www.genomeweb.com/clinical-genomics/minnesota-house-passes-newborn-screening-bill>.

153. OKLA. STAT. ANN. tit. 21, § 1175 (West 2012).

154. *Innocent Blood*, *supra* note 138; *see also* Denise Chrysler et al., *The Michigan BioTrust for Health: Using Dried Bloodspots for Research to Benefit the Community While Respecting the Individual*, 39 J.L. MED. & ETHICS 98, 98–99 (2011) (discussing the creation of Michigan’s Neonatal Biobank).

155. MICH. COMP. LAWS ANN. § 333.5431 (West 2001).

156. *Innocent Blood*, *supra* note 138.

157. *Id.*

158. *Id.*; *see also* AGR, *supra* note 8, at 65 (discussing Guthrie cards).

159. These issues also tap into a longstanding debate about ownership and control over one’s biological material, an issue on which we still have no clear consensus. Sonia M. Suter, *Disentangling Privacy from Property: Toward a Deeper Understanding of Genetic Privacy*, 72 GEO. WASH. L. REV. 737, 803–11 (2004); *see* C. Thomas, *The Use and Control of Heel Prick Blood Samples*, 24 MED. & L. 259, 261–68 (2005) (applying various theories of property ownership to NBS samples).

An important consideration in evaluating the propriety of the long-term storage and future uses of NBS samples is whether the samples are identifiable; that is to say, whether they can be linked directly to the newborn through identifying information or indirectly through a code. NBS blood spots must, of course, be identifiable initially so labs can locate and offer follow-up testing to children with abnormal results. But researchers try to anonymize previously identifiable samples by unlinking them from their source.¹⁶⁰ While some of the possible future uses of newborn samples require the samples to be identifiable—e.g., post-mortem identification, paternity testing, forensics, and future diagnostics—many kinds of research samples might potentially be anonymized, although as I note below, people are increasingly skeptical about the effectiveness of this practice.¹⁶¹

Current regulations require informed consent for research on biospecimens that have already been archived and are identifiable or linkable.¹⁶² The Federal Protections for Human Research Subjects, sometimes called the “Common Rule,”¹⁶³ require documented informed consent for participation in research.¹⁶⁴ Research on *identifiable* DBS easily falls within the definition of human subject research under the regulations, which includes analysis of “identifiable private information.”¹⁶⁵ While state NBS programs have “not traditionally been viewed as subject” to the Common Rule given that they are regulated by state health departments,¹⁶⁶ some scholars argue convincingly that the federal regulations should apply to research on DBS.¹⁶⁷

160. NBSTF, *supra* note 28, at 416 (noting that they may have been originally collected without identifiers or with identifiers that have been removed).

161. *Id.* at 416–17; *see infra* text accompanying note 220.

162. Carnahan, *supra* note 123, at 315.

163. *Id.* Seventeen federal agencies have adopted these protections “verbatim.” *Id.* at 315 n.102.

164. 45 C.F.R. § 46.117 (2013); *see also* Health Insurance Portability and Accountability Act of 1996 (HIPAA), Pub. L. No. 104-191, 110 Stat. 1936.

165. 45 C.F.R. § 46.102(f) (2013) (defining human subject).

166. Carnahan, *supra* note 123, at 315–16.

167. *Id.* at 316–17 (arguing that federal dollars and policy guidance directly and indirectly support NBS, including the collection, analysis, and storage of “newborn bloodspots for future research purposes”); *e.g.*, Therrell et al., *supra* note 140, at 1, 3.

Under the existing regulations, however, research on de-identified biological samples is generally understood to be exempt from federal protections of human subjects research.¹⁶⁸ Indeed, the Office of Human Research Protections does not “consider research involving *only* coded private information or specimens to involve human subjects . . . if . . . the private information or specimens were not collected specifically for the currently proposed research project . . . and the investigator(s) cannot readily ascertain the identity to the individual(s) to whom the coded private information or specimens pertain.”¹⁶⁹ One scholar argues that this exemption does not apply to DBS because they were collected not only as part of a screening program, but also as part of a “*research* program.”¹⁷⁰ While sympathetic to the view that the exemption should not apply, I am not persuaded that these samples would be treated differently from any other biospecimens under the research regulations because these samples were not collected with any specific research protocol in mind.

The question of whether and how research should be allowed on NBS or other biosamples reflects tensions between public and private interests, and more specifically between norms that focus on the value of research and norms that focus on individual rights, autonomy, and privacy interests.¹⁷¹

168. 45 C.F.R. § 46.101(b)(4) (2013) (exempting from the research regulations research “involving the collection or study of existing data . . . pathological specimens, or diagnostic specimens, if these sources are publicly available or if the information is recorded by the investigator in such a manner that subjects cannot be identified, directly or through identifiers linked to the subjects”).

169. U.S. Dep’t of Health & Human Servs., OHRP - Guidance on Research Involving Coded Private Information or Biological Specimens (2008), *available at* <http://www.hhs.gov/ohrp/policy/cdebiol.html>. This interpretation clearly seems to view research on biobanks with coded samples as not involving human subjects research, even though “[t]he increase in genomic data, as well as the increase of computerization of other records about individuals, will only make identifying ‘anonymous’ biobank files easier and easier.” Henry T. Greely, *The Uneasy Ethical and Legal Underpinnings of Large-Scale Genomic Biobanks*, 8 ANN. REV. GENOMICS & HUM. GENETICS 343, 352–55 (2007).

170. Carnahan, *supra* note 123, at 320 (observing that “one purpose” of the collection and storage of the DBS “is for *future* genetic research”).

171. Storage of Genetics Materials Comm., Am. Coll. of Med. Genetics, *ACMG Statement: Statement on Storage and Use of Genetic Materials*, 57 AM. J. HUM. GENETICS 1499 (1995). This issue creates tension between the ethical principle of informed consent, which argues in favor of recontacting individuals to obtain their consent, and the serious impracticabilities of doing so.

Similar tensions about autonomy interests versus some conception of the public good arise with respect to the question of whether consent should be required for NBS itself.¹⁷² In trying to determine how best to resolve these tensions, Part III sets up a framework for balancing the conflicting interests and applies this approach to the specific questions of whether some form of consent should be required for: 1) the storage and research uses of NBS samples; and 2) some or all aspects of NBS itself.

III. BALANCING THE INTERESTS

In exploring the tensions between the public good and the individual's privacy and autonomy interests, we can see how biases can influence the weight of the interests.¹⁷³ As we shall see below, those who strongly promote research and its benefits to newborns and society tend to undervalue the privacy and autonomy interests at stake. Similarly, the strong proponents of privacy and autonomy tend to undervalue the public value of the long-term retention and research use of DBS. As a result, they reach an impasse, not only because they value things differently, but also because their approaches differ.

Many proponents of expansive access to NBS samples and other archived tissues “tend to rely on a narrow version of consequentialism” to justify a broad range of research practices, while minimizing the privacy and autonomy interests at stake.¹⁷⁴ The benefits of this approach seem “concrete and tangible”¹⁷⁵: preventing morbidity and mortality in newborns, and gaining knowledge about various inherited disorders to advance medicine and clinical care.¹⁷⁶ The risks of broader access to NBS samples—privacy intrusions and the loss of autonomy interests—“are more amorphous concerns and are therefore less viscerally compelling.”¹⁷⁷ Indeed, many of the public benefit proponents easily dismiss the value of autonomy

172. See Suter, *supra* note 50, at 246–50 (discussing value considerations in prenatal testing).

173. Sonia M. Suter, *All in the Family: Privacy and DNA Familial Searching*, 23 HARV. J.L. & TECH. 309, 375–76 (2010) [hereinafter Suter, *AITF*] (discussing a parallel trend with DNA familial searches).

174. *Id.*

175. *Id.*

176. Carnahan, *supra* note 123, at 300.

177. Suter, *AITF*, *supra* note 173, at 375.

and privacy, and informed consent.¹⁷⁸ This view argues for expansive NBS with mandatory testing, long-term retention of samples, and broad access to these samples by researchers without consent.

In contrast, a position that privileges privacy and autonomy would push toward requiring detailed informed consent for all aspects of NBS: the collection of samples, the subsequent analysis, the retention of samples, the manner in which they are stored (coded, identifiable, or anonymized), access to the samples, and uses to which the samples are put.¹⁷⁹ This approach would limit many of the potential research benefits that have come from NBS programs and use of the samples.¹⁸⁰

Clearly neither extreme fully considers all that is at stake. As a result, I recommend an approach that “does not focus exclusively on one or just a few values or desirable consequences. Instead, it recognizes the competing goods at stake.”¹⁸¹ Because I have described this approach in more detail in an earlier piece, I will only briefly outline the methodology, which borrows from philosopher W.D. Ross.¹⁸² The central premise is that we have various underlying prima facie duties, which may sometimes come into conflict.¹⁸³ We have, for example, prima facie duties to protect the public by supporting and encouraging research and identifying children with treatable conditions in a timely manner to minimize morbidity and mortality. We also have prima facie duties to protect the autonomy of the family and the future autonomy of the newborns with respect to medical decision making and participation in research, and duties to protect the privacy of newborns. None of these duties is absolute in the sense that they must always override conflicting duties.¹⁸⁴ Instead, all of these duties are “intrinsically binding”—they hold sway over us, but “they are not always determinative of how we should act in

178. *Id.* at 376.

179. Carnahan, *supra* note 123, at 322–25.

180. *Id.* at 322 (noting that informed consent is problematic because future research methods are unknowable).

181. Suter, *AITF*, *supra* note 173, at 376.

182. *Id.*

183. *Id.* at 376–77.

184. *Id.* at 377.

any given instance Instead we can only determine what our actual duty is in any circumstance by full reflection.”¹⁸⁵

This approach does not attempt to declare winners and losers when competing values come into play. Rather, it attempts to reach a resolution that may ultimately tip more in the direction of one duty than the other, but which continues to recognize the pull of the competing values.¹⁸⁶ That is to say, when we determine what the actual duty is in any particular circumstance, we should not abandon or forget about the overridden *prima facie* obligations, because they continue to “exert force on our subsequent attitudes and actions”¹⁸⁷ and leave “residual effects” or “moral traces.”¹⁸⁸ If our full reflection leads us to decide that certain research goals are particularly important to society, we may decide to limit autonomy to some extent to allow for that research. The pull of our duty to protect individual autonomy, however, continues to compel us to “approximate as closely as possible the values enshrined in the overridden duty” so that we develop measures that least infringe on parental autonomy.¹⁸⁹

Considering whether consent should be required in NBS forces us to make difficult choices between various competing values and find ways to give weight, as much as possible, to the overridden *prima facie* duties. In particular, we must apply this balancing approach to decide: 1) what kind of consent provisions, if any, we should use for NBS itself; and 2) whether

185. *Id.* I note in this piece that “this methodology does not offer conclusive answers to most moral questions.” *Id.* at 378. It is, nevertheless, not arbitrary or subjective. Rather, it requires a kind of “reflective equilibrium” where we “check decisions from general principles against more intuitive judgments about proper outcomes for particular cases.” *Id.* at 379. See generally JOHN RAWLS, A THEORY OF JUSTICE 15–19, 40–47 (rev. ed. 1999) (describing the “reflective equilibrium”).

186. Suter, *AITF*, *supra* note 173, at 378.

187. *Id.*

188. JAMES F. CHILDRESS, MORAL RESPONSIBILITY IN CONFLICTS: ESSAYS ON NONVIOLENCE, WAR, AND CONSCIENCE 69 (1982) (citing Robert Nozick, *Moral Complications and Moral Structures*, 13 NAT. L.F. 1 (1968)); RICHARD B. MILLER, CASUISTRY AND MODERN ETHICS: A POETICS OF PRACTICAL REASONING 47 (1996); Suter, *AITF*, *supra* note 173, at 376 (“[O]verridden values remain significant and continue to exert force and obligations on our actions and deliberations. In other words, the overridden values do not go away; they retain ‘moral traces.’”).

189. MILLER, *supra* note 188, at 47.

consent should be required, and if so what kind, for the storage and future uses of the samples.

I should emphasize that the issue of consent for NBS itself and consent for storage and future uses need not be treated as a package. Indeed, there are strong arguments for separating the process of screening from the process of the creation of biobanks, as I suggest below, and therefore completely disaggregating the questions of consent. At the moment, however, affirmative consent is generally removed from the entire process. When we disaggregate the two sets of decisions—whether to participate in NBS and whether to participate in the biobank—it becomes clear that the conflicting public/private values are very different. With respect to NBS itself, at least when the conditions screened for develop in infancy and are treatable or subject to amelioration, the conflict is between the state's interest in the well-being of the newborn and the autonomy of the family. With respect to questions of storage and, in particular, research uses of the samples, the public value of research comes into conflict with the private values of the families' autonomy interests and the newborn's privacy and future autonomy interests. Because each set of questions raises different tensions, I address each issue in turn. I begin with the research question because it has received the most attention recently and because it indirectly has implications for the question of consent for NBS itself.

A. RETENTION AND RESEARCH USES OF DBS

In only a few other contexts does the government take one's tissue samples without consent and retain them for extended periods of time: after conviction of certain crimes,¹⁹⁰ and in the military.¹⁹¹ In the first instance, the conviction results in the loss of certain liberty interests.¹⁹² And in the case of the military, one has a choice not to join the military. But in the context of NBS, samples are usually taken without parental consent and then stored for long periods, potentially to be used

190. Bonnie L. Taylor, Comment, *Storing DNA Samples of Non-Convicted Persons & the Debate over DNA Database Expansion*, 20 T.M. COOLEY L. REV. 509, 512–14 (2003).

191. Megan Allyse et al., *Ethics Watch: The G.I. Genome: Ethical Implications of Genome Sequencing in the Military*, 12 NATURE REVIEWS GENETICS 589 (2011).

192. Taylor, *supra* note 190, at 514.

for research, an approach that “veers from the norm.”¹⁹³ As noted, the justifications for doing so in the case of NBS are rooted in a perspective that emphasizes the value of research and that views archived samples as something akin to community property.¹⁹⁴ Some also argue that the public interest and value of research are not just communal interests, but also individual interests because everyone benefits from the research.¹⁹⁵

Even if we value research, however, we must recognize the competing interests in autonomy and privacy in being able to decide whether and to what extent to participate in research and to control access to personal information. Privacy advocates point out the dignitary interests, sometimes suggesting that biosamples belong to the individual.¹⁹⁶ Serious privacy concerns arise when others have access to our genetic material, which contains “a wealth of personal information such as predisposition to certain diseases, behaviors, physical and mental traits, parentage, and genetic relatedness to others.”¹⁹⁷ The fact that the DBS contains genetic information and is likely to be “readily identifiable” leads some to say that consent is

193. Cohen, *supra* note 139.

194. See David Korn, *Genetic Privacy, Medical Information Privacy, and the Use of Human Tissue Specimens in Research*, in GENETIC TESTING AND THE USE OF INFORMATION 16, 53 (Clarisa Long ed., 1999) (arguing that archived human tissues are “a public resource dedicated to the public good, not, like a savings bank, a depository of private property”); see also Rebecca Skloot, *Taking the Least of You*, N.Y. TIMES, Apr. 16, 2006, at M45 (“[P]eople are morally obligated to allow their bits and pieces to be used to advance knowledge to help others. Since everybody benefits, everybody can accept the small risks of having their tissue scraps used in research.” (quoting David Korn, *supra*)).

195. Korn, *supra* note 194, at 60; Karen Rothenberg, *The Social Implications of the Use of Stored Tissue Samples: Context, Control, and Community*, in GENETIC TESTING AND THE USE OF INFORMATION 84, 85–88 (Clarisa Long ed., 1999) (suggesting that both privacy and research are public and private interests); see also Lisa Feuchtbaum et al., *Questioning the Need for Informed Consent: A Case Study of California’s Experience with a Pilot Newborn Screening Research Project*, 2 J. EMPIRICAL RES. ON HUM. RES. ETHICS 3, 3 (2007) (“[T]he legitimate needs of society and the interests of newborns should not be sacrificed to respond to the autonomy interests of the few parents who did not wish their infant to participate in the study . . .”).

196. Andrews, *supra* note 2, at 63.

197. Suter, *AITF*, *supra* note 173, at 331.

required whether or not the samples are “linked or linkable.”¹⁹⁸ Because this information is “fundamental and basic to our makeup” and plays such “an important, though not monolithic, role in influencing our ‘temperament, health, capacities, and physical appearance,’”¹⁹⁹ legislators at the state and federal level have enacted various forms of genetic privacy protections in the last few decades.²⁰⁰ I, like many others, have argued that genetic information is “integral to the self,” and therefore is among the kinds of personal information in which we have strong privacy interests.²⁰¹

Proponents of consent provisions for research on biosamples are also motivated by a commitment to principles of autonomy; the notion that individuals may not be treated as merely a means to an end.²⁰² Indeed, these ethical principles have led not only to formal declarations about the various ways in which researchers have an ethical obligation to protect research subjects, but also to legal regulations protecting the way in which research may and may not be conducted in the United States.²⁰³ Among the most fundamental principles of these ethical and legal norms are informed consent and the idea that the researchers have a fiduciary obligation to protect research subjects. A decision to become a participant in research either to advance medicine or to benefit others and/or oneself is a self-defining decision. It also creates a relationship of trust because it involves sharing personal information with researchers, imposing on them “special duties of care because of the imbalance of power inherent in the relationship.”²⁰⁴

The degree to which we emphasize our duties to promote research or to protect autonomy and privacy will determine our

198. Katherine Drabiak-Syed, *Legal Regulation of Banking Newborn Blood Spots for Research: How Bearder and Beleno Resolved the Question of Consent*, 11 HOUS. J. HEALTH L. & POL'Y 1, 13 (2011).

199. Suter, *AITF*, *supra* note 173, at 332.

200. See, e.g., Genetic Information Nondiscrimination Act of 2008 (GINA), Pub. L. No. 110-233, 122 Stat. 881; *Genetic Privacy Laws*, NAT'L CONF. ST. LEGISLATURES, <http://www.ncsl.org/default.aspx?tabid=14287> (last updated Jan. 2008) (describing the full range of state genetic privacy laws).

201. Suter, *supra* note 159, at 773. I have also noted that “genetic information is not uniquely, nor is all genetic information equally, central to the conception of the self.” Suter, *AITF*, *supra* note 173, at 334.

202. FURROW ET AL., *supra* note 107, at 405.

203. See 45 C.F.R. §§ 46.301–.306 (2013) (otherwise known as the “Common Rule”).

204. Suter, *supra* note 159, at 787.

approach to research on DBS. Under the extreme pro-research position, samples should be available in any form for use by researchers for any kind of investigation. Such an approach would seriously undermine the privacy interests of the child and autonomy interests of the family. It would allow the use of the newborn samples in identifiable form, which would privilege research over privacy and autonomy. Not surprisingly, this approach is inconsistent with the well-established consensus that under the Common Rule, identifiable samples cannot be used for research without one's informed consent.²⁰⁵ The Common Rule recognizes that the value of research, while real, is not absolute and therefore cannot override autonomy at all costs.²⁰⁶

At the other, pro-privacy/autonomy extreme, any future use of the samples for research would require detailed informed consent whether the samples were identifiable, coded, or anonymized, regardless of the uses. This approach would privilege privacy and autonomy interests over the value to the public of various research studies, potentially hindering research. It would be extremely difficult (if not impossible) and expensive to implement since it would require researchers to locate families to seek their consent for virtually every future study. Moreover, meaningful informed consent is often impossible to obtain when biospecimens, whether DBS or other forms, are initially collected because the parents or sources of the samples cannot be informed of all possible research uses and outcomes. In some ways, it might even be counter-productive to privacy interests since it would require the samples to remain identifiable while in long-term storage for the purpose of contacting the families.

The current system and recommended approach of some scholars and professional groups might be considered a compromise of sorts; informed consent is required if the samples are identifiable, but otherwise consent is not required for

205. 45 C.F.R. §§ 46.101(b)(4), 46.111(a)(4) (2013).

206. There are many methodologically sound and highly valuable types of research that we do not allow because values like privacy, autonomy, and the mental and physical well-being of individuals would make such studies unethical. The unfortunate history of human subject research in Nazi Germany and even in this country has taught us important lessons about the limits to which we can endanger others and limit their autonomy simply to further science. FURROW ET AL., *supra* note 107, at 405–13.

anonymized or de-identified samples.²⁰⁷ The theory, in brief, is that the privacy risks are substantially minimized once identifiers are removed. To the extent that no samples are ever truly anonymized, however, this argument becomes less persuasive. In addition, as some have pointed out, even under this system, sometimes researchers actually use biospecimens with identifiers, rather than in anonymized form, without obtaining consent or Institutional Review Board (IRB) approval.²⁰⁸

Regardless of whether we consider the current system appropriate for biobanks in general, we must recognize that NBS biobanks are unique in implicating particularly salient privacy and autonomy interests. First, parents often have not given consent to (or are even aware of) the collection of the biospecimen and NBS in the first place, let alone the long-term storage and potential research on the specimens. Indeed, one study showed that only twelve states mention specimen storage in the informational pamphlet that parents receive for NBS.²⁰⁹ With other biobanks, it is likely that the source of the specimen consented to (and knew about) the removal of the sample from his or her body (whether or not consent was given for later uses of the sample).

Second, these samples are obtained from minors and therefore any research on these samples is research on children, who are treated under the Common Rule as a vulnerable class deserving of heightened protection.²¹⁰ While minors can participate in research, there are very limited instances in

207. Amy L. McGuire & Laura M. Beskow, *Informed Consent in Genomics and Genetic Research*, 11 ANN. REV. GENOMICS & HUM. GENETICS 361, 370 (2010).

208. Drabiak-Syed, *supra* note 198, at 43. When the plaintiff in the *Bearder* litigation requested documentation from the Minnesota Department of Health (MDH) regarding its process of de-identification of samples for research, the MDH stated that it had no such documents, suggesting that “there is no established de-identification procedure and that the process and standards vary from project to project and are subject to subjective standards.” Whelan, *supra* note 18, at 441 (internal quotation marks omitted).

209. SEC’YS ADVISORY COMM. ON HERITABLE DISORDERS IN NEWBORNS & CHILDREN, CONSIDERATIONS AND RECOMMENDATIONS FOR NATIONAL GUIDANCE REGARDING THE RETENTION AND USE OF RESIDUAL DRIED BLOOD SPOT SPECIMENS AFTER NEWBORN SCREENING 16 (2009) [hereinafter ACHDNC] (citing personal communication with Aaron Goldenberg).

210. See 45 C.F.R. §§ 46.401–.409 (2013) (describing “Additional Protections for Children Involved as Research Subjects”).

which consent for participation is not required. For example, even the least problematic category of research on children—“[r]esearch not involving greater than minimal risk”—still requires the child’s assent and parental consent,²¹¹ unless the general waiver provisions for informed consent apply.²¹² Scholars have debated whether the waiver provisions should apply in this context.²¹³ The crux of the matter turns on whether informed consent is practicable or not. As one scholar notes, even when researchers do not have to obtain informed consent under the regulations, they often do, demonstrating that it is not always impracticable.²¹⁴ When children are involved and their biospecimens are retained for long periods of time, there is a strong argument that they should have the right (upon reaching the age of majority) to decide for themselves whether they want to be research participants.²¹⁵

Third, as I shall argue in more detail below, the state, as protector of the newborn and as mandator of the collection of the DBS, has a fiduciary obligation to protect the autonomy and privacy interests of the newborn with respect to the collection, retention, and use of the samples. For all of these reasons, whatever concerns we may have about the use of biobanks without consent (informed or general) are further heightened in this context.

211. 45 C.F.R. § 46.404.

212. 45 C.F.R. § 46.116(d) (2013) (waiving informed consent requirements when the research “involves no more than minimal risk to the subjects . . . [t]he waiver or alteration will not adversely affect the rights and welfare of the subjects . . . [t]he research could not practicably be carried out without the waiver or alteration,” and when appropriate, “the subjects will be provided with additional pertinent information after participation”).

213. Compare ACHDNC, *supra* note 209, at 19 (“A balanced consideration of concerns justifies waiving informed consent for population-based newborn screening research using de-identified specimens when a clinically well-defined test and an effective therapy are present.”), with Carnahan, *supra* note 123, at 320–21 (challenging the notion that informed consent would be “impracticable” because “a physician-patient relationship already exists between the physician and the mother-to-be, and it is typically the physician that is responsible for obtaining the bloodspot for screening and research”), and Drabiak-Syed, *supra* note 198, at 38 (suggesting that waiver has “been used as a creative mechanism to overcome administrative barriers”).

214. Ellen Wright Clayton, *Patients and Biobanks*, 51 VILL. L. REV. 793, 796–97 (2006).

215. David Gurwitz et al., *Children and Population Biobanks*, 325 SCI. 818, 818 (2009).

As a result, we should not weigh the interest in favor of research as strongly in this context as we might with respect to other types of biobanks. Indeed, this strongly supports the view that we should prohibit the use of DBS for any research.²¹⁶ While this would certainly limit the privacy and autonomy risks for the newborn and his or her family, to the extent that this population offers unique possibilities for research, one might argue that such a proposal goes too far. It is undoubtedly true that much of the research done on DBS need not be done on that particular population. But some forms of research may benefit substantially by collecting data from a pool, like the NBS samples, which represents the population so well. In addition, to the extent that any clinical data are combined with research on the DBS, research from birth through later life might offer unique insights into various disease processes that would be harder to obtain with other populations. Given that research of these samples poses heightened concerns, however, if any research on DBS should be allowed, it should be limited to research that benefits the pediatric population. Michigan's approach, for example, recognizes the importance of using newborn samples only for research that is relevant to the pediatric community.²¹⁷

To the extent that any research goes forward on DBS, for all of the reasons described above, it is appropriate to give families (and the child upon reaching the age of majority) some control over whether the DBS are archived for research purposes. Consistent with current requirements for research on biospecimens, informed consent should be obtained for research on identifiable NBS samples generally (except in the rare instances where a waiver could apply).

Under the current interpretations of the Common Rule, however, affirmative consent would not be required for de-identified samples,²¹⁸ which is problematic in the NBS context. As biobanks generally become more prevalent and central to genomics research, scholars have debated whether this approach is ethically justifiable, not just with respect to NBS, but for all biobanks. Scholars have argued that "a person has an

216. Hank Greely has argued that there is simply no reason for researchers to utilize DBS when there are other biorepositories to use. Author's personal communication.

217. Chrysler et al., *supra* note 154, at 99.

218. McGuire & Beskow, *supra* note 207, at 370.

interest in consenting or not consenting to be part of research,” even if it includes analysis of biospecimens.²¹⁹ Growing concerns about the inability to truly anonymize biological samples²²⁰ have led to further calls to rethink the current approach toward research on biospecimens.²²¹ Indeed, in response to advances “in genetic and information technologies that make complete de-identification of biospecimens impossible,” the Department of Health and Human Services proposed changes to the consent requirements for research on biospecimens.²²² Specifically, the proposed changes would eliminate the ability to do research on de-identified biological samples without consent. Instead, it would require “written *general* consent” for research use of archival biospecimens, whether or not researchers ultimately decide to use identifiers.²²³ The intended general written consent would allow individuals “to say no to all future research,” and give them the option to say yes or no to “a handful of special categories of research with biospecimens” that might raise “unique concerns . . . for a significant segment of the public.”²²⁴ In addition, the proposed changes would allow

219. See, e.g., Greely, *supra* note 169, at 356.

220. See, e.g., *id.* at 351–52; Melissa Gymrek et al., *Identifying Personal Genomes by Surname Inference*, 339 SCI. 321, 321 (2013); Nils Homer et al., *Resolving Individuals Contributing Trace Amounts of DNA to Highly Complex Mixtures Using High-Density SNP Genotyping Microarrays*, 4 PUB. LIBR. SCI. GENETICS, Aug. 29, 2008, at 1–2; Zhen Lin et al., *Genomic Research and Human Subject Privacy*, 305 SCI. 183, 183 (2004); Amy L. McGuire & Richard A. Gibbs, *No Longer De-Identified*, 312 SCI. 370, 370–71 (2006); Laura L. Rodriguez et al., *The Complexities of Genomic Identifiability*, 339 SCI. 275, 275–76 (2013).

221. See, e.g., Lori B. Andrews, *Harnessing the Benefits of Biobanks*, 33 J.L. MED & ETHICS 22, 24 (2005); Carnahan, *supra* note 123, at 320.

222. Human Subjects Research Protections: Enhancing Protections for Research Subjects and Reducing Burden, Delay, and Ambiguity for Investigators, 76 Fed. Reg. 44,512, 44,525 (proposed July 26, 2011) (to be codified at 45 C.F.R. pts. 46, 160, 164). These were part of a broader proposed overhaul of the “Common Rule.” *Id.* at 44,514.

223. *Id.* at 44,519 (emphasis added). The proposed regulations would move away from the concept of “exempt research” and create a new category of “excused research” that is intended both to “increase protections”—by requiring general consent as opposed to no consent for all biospecimens (as well as for pre-existing data collected for research, whether or not the researcher uses identifiers, and for pre-existing data that were collected for purposes other than research, if the researcher uses identifiers)—“and broaden the types of studies covered,” by allowing researchers to use identified biospecimens as long as they had general consent. *Id.* at 44,518–19.

224. *Id.* at 44,519–20 (giving as examples the creation of cell lines or reproductive research).

for waivers in some (unspecified) instances.²²⁵ Although these proposed regulations have not been adopted so far, they reflect an attempt to balance the pressures to promote research and protect individual privacy and autonomy.²²⁶

Following a modified version of the proposed amendments to the regulations for human subjects research, states should ask for *general* consent for the storage of DBS for future research uses of de-identified DBS.²²⁷ Parents would be entitled to say no to all future research, yes to all future research, or no to a handful of specific categories of research that might be problematic.²²⁸ In addition, children, upon reaching the age of majority, should be able to refuse consent for research or for particular categories of research.²²⁹

The focus on general, as opposed to detailed informed, consent serves two functions. It attempts to give parents (and the future adult the newborn will become) some autonomy protections while recognizing the value of research.²³⁰ It concedes the pro-research view that fully informed consent in this context truly is problematic; at the time the samples are collected, there may not be any specific plans for research, let alone for specific research protocols.²³¹ Thus, it is simply impossible to inform parents about the details of possible future research. In addition, the circumstances in which the samples are collected—during the newborn period—do not easily lend themselves to the lengthy discussions that informed consent

225. The advance notice of proposed rulemaking (ANPRM), however, notes that the waivers “would not necessarily be the same as those for other types of research.” *Id.* at 44,520.

226. *Id.*

227. *Id.* at 44,519. I call this a modified version because the ANPRM would require general consent for both the use of identified and identifiable samples. In my view, as long as informed consent is required for identifiable samples in other contexts, there is no argument for affording NBS biobanks less protection than other biobanks. Moreover, the rationale for using samples in this form would likely be to follow clinical outcomes, which itself would require considerable efforts to contact families or physicians to obtain clinical information.

228. *Id.* at 44,518–20.

229. *Id.* at 44,524.

230. Feuchtbaum et al., *supra* note 195, at 8–9 (discussing parental autonomy protections).

231. Elster, *supra* note 135, at 187–88.

would require, even if the specific future research protocols were known.²³²

Of course, if we were to separate the NBS process from the collection of samples for research, then this removes many of the challenges of obtaining consent during the newborn period. Such an approach might be justified by the concerns that the research is not in any one newborn's best interest, but instead serves the public good.²³³ As a result, we should eliminate any pressure to consent to research that might occur during the newborn period, especially if parents do not fully understand that the question of screening is not conceptually or practically linked to whether or not research is done.

But disaggregating consent for screening from consent for research does not eliminate the general problem of obtaining fully informed consent for research on pathology samples, given the impossibility of knowing about all future research endeavors in advance. Moreover, such disaggregation potentially removes one of the benefits of collecting DBS during the newborn period—the potential of collecting samples that represent the population. The challenges of tracking down families after that period would undoubtedly diminish the yield of samples available for research, potentially even more than the process of trying to obtain more complete informed consent. A lesser, but real, concern is that families that wanted to support such research but were not tracked down would lose out on the chance to consent to research. Of course, seeking consent for retention of samples for research in the prenatal period might lessen these concerns, although this would not be helpful in cases where women do not receive prenatal care.²³⁴ Thus, while some powerful reasons argue for separating consent for research from consent for NBS, we should recognize that such an approach is not without costs.

At whatever stage the consent process occurs for research on DBS, I am advocating what is essentially an opt-in approach for future research. Undoubtedly, even this approach would be less favorable to the research community than being able to access de-identified samples without any consent requirement,

232. *Id.*

233. Drabiak-Syed, *supra* note 198, at 36–38 (focusing on the benefit of the majority).

234. Whelan, *supra* note 18, at 452 (noting that not all women receive prenatal care).

because surely the latter approach would maximize the number of available samples. As a second choice, they would likely prefer opt-out to opt-in provisions under the theory that they are likely to have a larger pool of samples if parents must act affirmatively to *prevent* the storage of the samples, as opposed to requiring parents' affirmative consent *for* storage and future research.²³⁵ One consideration in choosing opt-in versus opt-out approaches is what the legislative default goals are. If the incentives are to promote research, the "nudging" of an opt-out approach may be viewed as making it more likely that such samples are available.²³⁶ But given the many concerns surrounding research on DBS, it is hard to argue we should be trying to "nudge" families into participating in research.

In fact, the data so far suggest that it is debatable how great the risk is that people would decline participation in research. Several studies suggest that a large percentage of parents would consent to participate in research.²³⁷ A 2008 study, for example, found that 90% of mothers would agree to participate in an NBS biobank with no restrictions on the type of research performed.²³⁸ Another study found that 76.2% of parents were "very or somewhat willing" to permit storage of and research on DBS, whereas if consent were not obtained, only 28.2% would be "very or somewhat willing" to allow the use of DBS for research.²³⁹ On the other hand, Texas's limited experience with opt-out provisions suggests that it had some, though not a significant, effect on the size of the newborn pool. In a roughly six-month period, 240,000 samples were collected

235. *Innocent Blood*, *supra* note 138 (explaining how any samples moving forward require consent as part of the opt-in program).

236. See RICHARD H. THALER & CASS R. SUNSTEIN, NUDGE: IMPROVING DECISIONS ABOUT HEALTH, WEALTH, AND HAPPINESS 83–86 (2008) (noting the importance of the default position for opt-out v. opt-in rules).

237. *E.g.*, Feuchtbaum et al., *supra* note 195, at 7–8; Alon B. Neidich et al., *Empirical Data About Women's Attitudes Towards a Hypothetical Pediatric Biobank*, 146A AM. J. MED. GENETICS 297, 299 (2008); B.A. Tarini et al., *Not Without My Permission: Parents' Willingness to Permit Use of Newborn Screening Samples for Research*, 13 PUB. HEALTH GENOMICS 125, 130 (2010).

238. Neidich et al., *supra* note 237, at 302; *see also* Feuchtbaum et al., *supra* note 195, at 7 (stating that although not all parents were asked to participate in a study of NBS because of the burdens on the hospital, ninety percent of those asked consented to enroll their NBS in the study to research NBS testing methods and to identify additional genetic diseases).

239. Tarini et al., *supra* note 237, at 128–29 (finding that women had misperceptions about what participation in a biobank would entail).

and the state received 6900 requests to destroy samples—a rate of 2.8%.²⁴⁰ We do not know how these numbers would compare with an opt-in provision or what parents understood about storage and possible future uses when they opted out.

In addition, there are potentially legitimate concerns about the possibility of consent bias when parents opt in. Many argue that giving people the opportunity to say no would not only reduce the pool of biospecimens available for research because of “uninformed denial,”²⁴¹ but would also lead to consent bias in the biospecimens that are available.²⁴² Given that the pool of newborns is so vast, there may be reason to think that the effects of consent bias might be lessened, albeit not completely eliminated, by the sheer number of samples potentially available.

Even if evidence shows that the pool of research samples might be smaller with an opt-in provision or that there is a greater risk of consent bias, this alone is not a reason to reject these measures to protect autonomy.²⁴³ The entire justification for removing consent requirements from NBS generally is the notion that the screening program is intended to benefit newborns.²⁴⁴ Removing consent for participation in future research on DBS cannot be justified on the same grounds.²⁴⁵ The extent to which the research benefits newborns may vary, but even research that is primarily geared toward benefiting newborns will provide much more indirect benefits than the actual screening for treatable and serious conditions.²⁴⁶ Research that does not focus on the newborn or pediatric population offers even less benefit to newborns and cannot at all justify the lack of consent.²⁴⁷ Thus, as noted earlier, any

240. Roser, *supra* note 147, at A1.

241. Korn, *supra* note 194, at 48.

242. *E.g.*, Barbara J. Evans, *Much Ado About Data Ownership*, 25 HARV. J.L. & TECH. 69, 95–98 (2011); Kharaboyan et al., *supra* note 130, at 747.

243. Drabiak-Syed, *supra* note 198, at 36 (noting that the “benefit to the majority is not alone a sufficient interest to override individual autonomy”); Whelan, *supra* note 18, at 453 (“As a society, we cannot allow administrative costs or burdens to justify infringements on individual rights, parental rights, and genetic privacy.”).

244. Drabiak-Syed, *supra* note 198, at 36.

245. *Innocent Blood*, *supra* note 138.

246. Feuchtbaum et al., *supra* note 195, at 7–9.

247. *Id.* at 11–12.

research on DBS should ideally be limited to that which benefits the pediatric population.

One additional concern with the opt-in approach is that requiring affirmative consent for retention and research uses of DBS will lead some parents to opt out of NBS altogether in jurisdictions where that is possible.²⁴⁸ Here, the value of providing parental autonomy and the child's future autonomy is set against the potential harms to newborns if severe and treatable conditions are not identified in the newborn period.²⁴⁹ This concern might, therefore, argue for decoupling consent for NBS from the consent for research uses of DBS.

There is a strong argument to be made the other way, however. Whether or not the consent process for NBS and research are disaggregated, seeking parental consent for future research on the DBS helps establish the public's trust in the NBS process generally.²⁵⁰ Recent attention to long-term storage and research uses of these samples may lead parents to think of NBS, not so much as a program intended to protect the health of newborns, but as an effort to create a universal research pool.²⁵¹ This may create push back with respect to NBS altogether, causing parents to opt out of NBS to resist what they perceive as the heavy hand of government.²⁵² As Dr. Jeffery Botkin suggests, denying parents the chance to opt out of future research may undermine the public's trust in the entire endeavor.²⁵³ Indeed, it is precisely such suspicion and loss of trust that led to the lawsuits in Texas and Minnesota.²⁵⁴ As one parent in the Texas lawsuit explained, "To me, this whole thing is about consent If they had asked me I probably would have consented. The fact that it was a secret program really made me so suspicious of the true motives, there's no way I would consent now."²⁵⁵ Thus, as long as any research is done on the DBS, whether consent is obtained in the future or during the newborn period, the public needs to know

248. *Id.* at 11.

249. *Id.* at 8–9.

250. Drabiak-Syed, *supra* note 184, at 12–13, 23, 42.

251. *Id.* at 23, 35–36.

252. *Id.* at 35–36.

253. *Innocent Blood*, *supra* note 138 (quoting Jeffrey R. Boktin).

254. Drabiak-Syed, *supra* note 198, at 25–34.

255. Roser, *supra* note 147, at A1; *see also* Whelan, *supra* note 18, at 442 ("As one parent succinctly stated: 'I want to have the choice.'").

that any use of these samples requires affirmative consent from parents. The state should not *presume* consent.

Not only is the public's trust important to the sustainability of the NBS project as a whole, but trust is also inherent in the relationship the state creates between itself and the child in setting up NBS. The most persuasive justification for NBS is the *parens patriae* notion that the state steps in to act as parent for the child.²⁵⁶ This creates a trust-based, fiduciary relationship (which goes beyond the ordinary fiduciary obligation the state owes its citizens) given that the state takes over some aspects of the child's care for the well-being of the child.²⁵⁷ As a consequence, a strong obligation exists not only to ensure that NBS maximizes the well-being of the child, but to ensure that any ancillary uses of the samples do not in any way undermine the best interests of the child, even for the benefit of society as a whole.

Michigan's creation of the BioTrust for Health, which is intended to facilitate and promote research on the DBS of NBS, was modeled on the concept of a charitable trust.²⁵⁸ Under this model, the source of the specimen (in this case the parent acting on behalf of the child) "formally expresses" the desire to transfer the specimen into the control of the trustee (the state) who will keep the sample for the benefit of the beneficiary (the general public).²⁵⁹ Important to this approach is the notion that the transfer is *intentional and freely given*, and that the recipient of biospecimens (in this case the state) "has a responsibility to serve as a trustee, or steward, of the tissue to ensure protection of the contribution."²⁶⁰ This model suggests three things: first, that parents should consent to the use of their newborn's samples for inclusion in the research biobank; second, that the samples are to be used for the benefit of the public; and third, and most important, that the recipient has a fiduciary obligation not only to develop clear rules about the kinds of uses to which these samples can be put, but also to implement security measures to protect the confidentiality of the

256. AGR, *supra* note 8, at 261.

257. *Id.*

258. Chrysler et al., *supra* note 154, at 98 (citing David J. Winickoff & Richard Winickoff, *The Charitable Trust as a Model for Genomic Biobanks*, 349 NEW ENG. J. MED. 1180, 1180 (2003)).

259. Winickoff & Winickoff, *supra* note 258, at 1182–83.

260. *Id.* at 1182.

information in the samples.²⁶¹ Given the limits of de-identification and anonymization in protecting privacy,²⁶² it is particularly important that the state develop explicit guidelines as to the legitimate uses of the samples both in terms of the best interests of the newborns and the public and in terms of security measures.

Indeed, the charitable trust model does not require that the state hold the DBS. Instead, a non-state charitable trust could be created and charged with the obligation of holding the samples and ensuring that their use is for the benefit of the public. The fact that the state would not possess the DBS and that this approach would disentangle the NBS process from the research aspects would likely help promote public trust.

While there are legitimate concerns about the impracticabilities of obtaining informed consent about future research uses, efforts should be made to inform parents about the general nature of the permissible and impermissible uses of the samples as well as security provisions. Such efforts would not only protect the autonomy interests of the family, but might also indirectly promote research. If families believe that the government has given careful attention to the kinds of uses that it will and will not allow, and has been attentive to the security of this personal information, families may be more inclined to participate. Otherwise, the public may not trust the state, believing, at best, that it has been negligent in protecting against problematic uses of the samples or, at worst, that the state may have malignant plans for such samples, which is why it has not set limits on these future uses.

B. CONSENT FOR NBS ITSELF

A conclusion that parental consent should be required for storage and research use of a newborn's DBS does not necessarily mean that consent should also be required for NBS itself. In fact, Michigan, whose BioTrust approach for research on DBS is commendable, requires written consent for the inclusion of the samples in the biobank (and the right of a child upon age of majority to have their DBS removed), but it does

261. *See id.* at 1182–83 (describing the charitable trust model generally and emphasizing the factors asserted in the text).

262. Greely, *supra* note 169, at 352–55.

not require consent for the screening.²⁶³ Moreover, the balance of public and private interests argues less strongly for affirmative consent with respect to NBS than in the research context since achieving high rates of NBS not only benefits the newborn, but also parents and society as a whole.²⁶⁴ Even though I concede that the case for consent is less strong in this context, the recent and likely future expansions in NBS make an increasingly compelling case for rethinking parental consent in this context as well.²⁶⁵

To be sure, there are serious challenges in requiring true informed consent for the screening itself. Given the number of diseases screened for, obtaining meaningful informed consent of the sort that the law demands for a physically invasive and risky medical procedure would be virtually impossible for each and every condition in the NBS panel.²⁶⁶ The likely expansion of the panel of diseases and possibility of whole genome sequencing in the future only enhances this problem. Whatever challenges conveying this wealth of information presents in ordinary circumstances are magnified by the fact that the disclosures typically occur during the newborn period, when parents are unlikely to be able to process the details of the nature of each of these conditions, the various treatment options for affected children, and the likelihood each of the conditions will manifest symptoms.²⁶⁷ Additional concerns surrounding informed consent are the economic and logistical

263. Chrysler et al., *supra* note 154, at 100.

264. Feuchtbaum et al., *supra* note 195, at 8–12.

265. Even if consent should occur for NBS, however, it does not follow that it should occur at the same time as consent for research. Indeed, as noted above, there are some powerful reasons to separate out the two consent processes.

266. This is a problem generally with any kind of multiplex testing. See, e.g., Council on Ethical & Judicial Affairs, Am. Med. Ass'n, *Multiplex Genetic Testing*, HASTINGS CENTER REP., July–Aug. 1998, at 15, 15–18 (explaining multiplex genetic testing and informed consent within this context); Robert J. Wells, Correspondence, *Generic Consent for Genetic Screening*, 331 NEW ENG. J. MED. 1024, 1024 (1994) (“Burdening us all with a system of ‘enforceable’ standards . . . will keep us ignorant by delaying the gathering of information needed to make these kinds of determinations.”); see also Greely, *supra* note 169, at 352–55, 357–59 (discussing the hurdles in obtaining informed consent for genetic research and testing).

267. AGR, *supra* note 8, at 6 (explaining the disclosure methods during the newborn period).

burdens such a requirement would place on health care providers and the public health system.²⁶⁸

How logistically challenging it is to obtain consent for NBS, however, is debatable. One much cited pilot study for a new NBS technology confirms some of these worries. The research study, which required informed consent, found that obtaining written informed consent was a “serious logistical burden” for the hospitals involved.²⁶⁹ As a result, the researchers only achieved forty-seven percent participation in the study.²⁷⁰ On the other hand, a study in Germany suggested that much higher participation rates could be achieved when written consent was sought.²⁷¹ In that case, almost ninety-nine percent of the parents consented to NBS.²⁷² Similarly, an older study of Maryland’s previous informed consent approach to NBS found “no evidence that the parental consent regulation had a negative effect on the public’s health. . . . [or] that the [NBS] program had become less cost-effective.”²⁷³ The data seems mixed as to the burden that seeking informed or written consent imposes.

To say, however, that obtaining true informed consent is impossible, results in unacceptably low yields of parental consent, or is effective but unduly expensive, does not mean we should abandon all efforts to seek *any* form of parental consent.²⁷⁴ An approach that requires affirmative parental consent—i.e., an opt-in approach—would offer the next best form of respecting parental autonomy. Most states, however, have chosen the opt-out approach, which theoretically still offers some parental control because it creates the right for parents who greatly oppose NBS to decline screening of their

268. *Id.* at 156–57.

269. Feuchtbaum et al., *supra* note 195, at 6.

270. *See id.* at 7 (stating that only forty-seven percent of newborns participated in the MS/MS screening during the pilot study’s time frame).

271. Bernhard Liebl et al., *Very High Compliance in an Expanded MS–MS–Based Newborn Screening Program Despite Written Parental Consent*, 34 PREVENTIVE MED. 127, 127 (2002).

272. *Id.* at 127, 130–31.

273. Faden et al., *supra* note 80, at 1351.

274. *See* Ainsley Newson, *Should Parental Refusals of Newborn Screening Be Respected?*, 15 CAMBRIDGE Q. HEALTHCARE ETHICS 135, 140, 144 (2006) (“Although parental autonomy is not, of course, legally or morally limitless, parents should (and do) enjoy a degree of freedom from state interference in private and family life.”).

newborn.²⁷⁵ In order for an opt-out option to offer any true semblance of respecting parental autonomy, however, parents must understand that they have an option to opt out, which requires some awareness and general understanding of the NBS process and the option to opt out.²⁷⁶ Unfortunately, that rarely happens.²⁷⁷ This may be because providers fail to inform parents, because so much is happening during the newborn period that parents cannot absorb or process whatever information they might get, or some combination of the two. As a result, there is a strong case for NBS education to occur in the prenatal period when there is more time for reflection, discussion, and comprehension.²⁷⁸ Although, again, this is only helpful for women who receive prenatal care.

Even if education regarding NBS were enhanced by requiring NBS education during the prenatal period, there is reason to think that an opt-out approach would still be less than optimal if the goal is parental education. The incentives simply are too few to educate parents under an opt-out as compared to an opt-in approach. Under an opt-out approach, the default is to test, which creates no incentive to discuss NBS with parents.²⁷⁹ Testing will occur with or without such a discussion. A statutory requirement to discuss NBS might not be a sufficient incentive to educate the families in light of the many other demands on health care providers' time. In contrast, under an opt-in approach, the default is not to test unless parents consent, which creates strong incentives to discuss NBS with parents, even if only in general terms.²⁸⁰

An additional argument in favor of the opt-in approach, given the goal of parental education, is that it is more cost-

275. Feuchtbaum et al., *supra* note 195, at 9.

276. Moody & Choudhry, *supra* note 9, at 246–48.

277. *See id.* at 240, 244 (noting that parents “are not even aware that they have a clear choice to make” in the United Kingdom’s opt-out program, and finding in their own study that 41.7% of respondents “did not feel able to decline,” while many thought NBS was “compulsory”).

278. *See, e.g.*, MICH. DEP’T OF CMTY. HEALTH, NEWBORN SCREENING GUIDE FOR HOSPITALS 19 (2014) (“Education is ideally done during the prenatal period.”).

279. Faden et al., *supra* note 80, at 1350 (discussing how mothers believe a routine default procedure does not require consent or discussion).

280. *Id.* at 1351 (describing the procedure in Maryland, which would appear to be similar to the current suggestion).

effective than full-blown informed consent would be.²⁸¹ The study of Maryland's program established, albeit many years ago, that parents can be educated adequately about newborn screening *generally*—not with respect to the details of every condition—in no more than five minutes.²⁸² Further, there are cost-effective methods, such as decision aids, which are being developed for a range of medical decisions,²⁸³ to provide parents with an overview of NBS. Indeed, some have advocated a system that would provide basic information about NBS to parents with options for access to more detailed information should they want it.²⁸⁴ Such an approach would further promote autonomy by allowing people to decide how much information to receive.

Were there evidence to suggest that an opt-in approach would lead to a great deal of uninformed denial, this might be a powerful reason to forgo some protections of parental autonomy to prevent (the admittedly small number of) newborns from suffering from debilitating or life-threatening illnesses. But evidence suggests, as we shall see, that involving parents in the decision-making process may actually *enhance* the effectiveness of NBS, and therefore opt-in provisions may further both goals—protecting the health of the newborn population and promoting parental autonomy.²⁸⁵

A study conducted over two decades ago showed that the refusal rate for NBS is really quite low in the states where NBS is truly voluntary.²⁸⁶ It found that Maryland and New Hampshire, out of twelve states studied, had the highest percentage of NBS: ninety-eight percent of their newborns.²⁸⁷

281. *See id.* (“There was also no evidence that the program had become less cost-effective because of increased costs to the health care system.”).

282. *See id.* at 1350 (“Most nurses . . . responded that obtaining consent or refusal took from one to five minutes.”).

283. *See, e.g.,* Elie A. Akl et al., *A Decision Aid for COPD Patients Considering Inhaled Steroid Therapy: Development and Before and After Pilot Testing*, BMC MED. INFORMATICS & DECISION MAKING, May 15, 2007, at 1, 4–6 (discussing the use of decision aids for COPD).

284. Harrell, *supra* note 38, at 849–50.

285. Jean-Louis Dhondt, *Implementation of Informed Consent for a Cystic Fibrosis Newborn Screening Program in France: Low Refusal Rates for Optional Testing*, 147 J. PEDIATRICS S106, S107–08 (Supp. Sept. 2005); Liebl et al., *supra* note 271, at 130–31.

286. Andrews, *supra* note 2, at 60.

287. *Id.*

Maryland had a program that required informed consent²⁸⁸ (it now has an opt-out approach²⁸⁹), and New Hampshire allows parents to refuse NBS for any reason.²⁹⁰ In contrast, the other ten states, all with mandatory screening programs that allow parental refusal only for religious reasons, screened fewer newborns. One state managed to screen a mere fifty-eight percent of its neonates.²⁹¹ More recent studies show that parental consent is over ninety percent when parents are allowed to opt out of screening or even sometimes required to consent affirmatively.²⁹² A possible explanation for these data is that a voluntary program that informs and educates parents about NBS induces parents to ensure actively that their children will actually get screened.²⁹³ By contrast, mandatory programs—especially those in which parents are not well-educated about NBS—lack this additional “check on the procedure,” resulting in a lower yield of children screened.²⁹⁴

Interestingly, most parents do not believe that informed, or sometimes even *any*, parental consent is necessary for NBS,²⁹⁵ at least with respect to conditions that present in infancy. On first glance, these findings might cut in favor of maintaining the status quo. In one study, parents did, however, want choice.²⁹⁶ Nearly three-quarters of parents preferred opting out and a

288. PRESIDENT'S COUNCIL ON BIOETHICS, *supra* note 45.

289. MD. CODE ANN., HEALTH-GEN. § 13-109 (West 2013).

290. N.H. REV. STAT. ANN. § 132:10-c (2013).

291. Andrews, *supra* note 2, at 60–61. This study did not investigate an important question, which is how effective the education efforts are in these, as opposed to other, opt-out programs.

292. Dhondt, *supra* note 285, at S106; Liebl et al., *supra* note 271, at 130–31; Evelyn P. Parsons et al., *Mothers' Accounts of Screening Newborn Babies in Wales (UK)*, 23 MIDWIFERY 59, 62–63 (2007).

293. Liebl et al., *supra* note 271, at 130–31. One author questions whether the “consent” procedures in these voluntary programs are truly informed because consent is given at the time of screening. She suggests that parents will say yes to anything right after birth, which could result in artificially high consent rates and could explain why the voluntary programs have such high participation rates. Harrell, *supra* note 38, at 850.

294. Andrews, *supra* note 2, at 60.

295. Elizabeth D. Campbell & Lainie Friedman Ross, *Incorporating Newborn Screening into Prenatal Care*, 190 AM. J. OBSTETRICS & GYNECOLOGY 876, 876–77 (2004); Faden et al., *supra* note 80, at 1350–51 (stating that forty-six percent felt that their consent should not be sought); Moody & Choudhry, *supra* note 9, at 246–48.

296. Moody & Choudhry, *supra* note 9, at 244–46.

little over one-quarter preferred opt-in approaches.²⁹⁷ However, when asked about mandatory screening for conditions that do not present in infancy, such as Duchenne muscular dystrophy, which presents between three and ten years of age, and Alzheimer's disease, which presents in adulthood, a majority of parents opposed mandatory screening.²⁹⁸ This may reflect the fact that there is little that can be done to prevent these conditions from developing in the newborn period or at all. On the other hand, another study found that most parents support mandatory screening of diseases that present in infancy, even if no treatment is available,²⁹⁹ suggesting that for some parents elimination of the diagnostic odyssey, even if nothing can be done, is important for childhood illnesses.

The fact that parents are not clamoring to give consent for NBS or that they seem to prefer opt-out over opt-in approaches, ironically, may support an opt-in approach. The typical reason for their views is a concern that other parents would not consent. This supports the findings that when consent is required, there is actually a high level of acquiescence.³⁰⁰ In other words, the majority of parents would likely consent to NBS themselves; they do not want consent requirements because they fear that *other* parents would not consent. This reasoning alone does not, of course, necessarily overcome the concerns of cost, time, and logistical demands associated with affirmative consent.³⁰¹

What further argues in favor of the opt-in approach is the fact that parents consistently express a strong desire for *education* and *information* regarding NBS, which they are not getting.³⁰² Overall, studies suggest that parents "were more troubled over the lack of NBS education than by the lack of

297. *Id.* at 246.

298. L.E. Hasegawa et al., *Parental Attitudes Toward Ethical and Social Issues Surrounding the Expansion of Newborn Screening Using New Technologies*, 14 PUB. HEALTH GENOMICS 298, 303 (2011); *see also* Campbell & Ross, *supra* note 295, at 876–77.

299. Hasegawa et al., *supra* note 298, at 303–04.

300. *See supra* notes 286–94 and accompanying text.

301. *See* Elster, *supra* note 135, at 187–89 (discussing the ethical and legal issues regarding informed consent).

302. TIMMERMANS & BUCHBINDER, *supra* note 8, at 61 ("Public opinion research suggests that few new parents know about newborn screening."); Hasegawa et al., *supra* note 298, at 302. *But see* Whelan, *supra* note 18, at 428 ("[A] majority of parents are aware of the initial screening.").

consent.”³⁰³ Many urge that such education should happen in the less hectic prenatal, as opposed to newborn, period when they would be less preoccupied.³⁰⁴ If the goal is primarily to satisfy parental requests for information, it may be that requiring affirmative consent is the best way to do that. Studies have shown that seeking affirmative consent can increase parental knowledge in the context of research studies.³⁰⁵ In addition, as noted above, the incentives to provide some information about NBS are greater with an opt-in as compared with an opt-out approach. Thus, a powerful justification for requiring opt-in for NBS itself is to enhance the chances that parents understand something about NBS, which can satisfy their desires and likely promote the effectiveness of NBS.

If we could trust that the education would happen in the prenatal, or even newborn, period, the case for opting in would be weaker. The current inadequacy of parental education, however, not only supports the opt-in requirement as a method to try to ensure that such education occurs;³⁰⁶ it is relevant in another respect. An opt-out approach is only protective of autonomous decision making if it is *informed* refusal.³⁰⁷ If parents are not adequately educated about NBS, or even worse that NBS occurs and that they can refuse, the opt-out approach makes a mockery of the notion of autonomous decision making and informed refusal. Instead, it merely leaves parents with an empty legal right to refuse. Even if most parents, when educated about NBS, would choose not to opt out, many who do not opt out are not making an affirmative choice because they

303. Hasegawa et al., *supra* note 297, at 303; *see also* NEDRA S. WHITEHEAD ET AL., DEVELOPING A CONJOINT ANALYSIS SURVEY OF PARENTAL ATTITUDES REGARDING VOLUNTARY NEWBORN SCREENING 6 (2010), available at <http://www.rti.org/pubs/mr-0014-1003-whitehead.pdf> (“Most parents would like more information on newborn screening . . .”); Campbell & Ross, *supra* note 295, at 877 (examining the need for increased prenatal NBS education); Faden et al., *supra* note 80, at 1350 (providing that around eighty percent wanted to be informed that NBS was done).

304. WHITEHEAD ET AL., *supra* note 303, at 6; Campbell & Ross, *supra* note 295, at 877.

305. Neil A. Holtzman et al., *Effect of Parental Informed Consent on Mothers’ Knowledge of Newborn Screening*, 72 PEDIATRICS 807, 811 (1983); Parsons et al., *supra* note 292, at 63–65.

306. Campbell & Ross, *supra* note 295, at 877 (discussing how parents are strongly requesting the necessary education, especially during the prenatal period).

307. Newson, *supra* note 274, at 141 (showing how an informed decision to refuse consent does not override autonomy).

did not know about NBS or the opportunity to opt out.³⁰⁸ In short, the opt-out approach under the current circumstances is so far from true consent or informed decision making that it is hard to argue that it does anything at all to promote autonomy.³⁰⁹

If providers were to offer the kind of information about NBS that would make the opt-out approach truly informed refusal, the process would be quite close to informed *consent*. At that point, the distinctions between opt-out and opt-in are simply not that great. Indeed, studies show that if individuals are adequately informed, the number who opt in is the inverse of those who opt out.³¹⁰ One of the reasons for the opt-out is the idea of “nudging” people to make the “right” choices.³¹¹ Given that the parent community is, based both on parents’ views and surveys of parents’ choices, not a community that needs to be nudged with respect to NBS, and given the added incentives to educate parents that opt-ins provide, the case of opt-in over opt-out becomes greater.

While there has been a long tradition opposing an opt-in approach, the reasons for reconsidering this approach are quickly growing.³¹² First, the fact that the broader panel of diseases increases the risks of false positives or the possibility of incidental findings of uncertain clinical relevance means that some of the psychosocial risks of NBS are increasing.³¹³ Parental awareness of NBS may prepare parents for and therefore decrease the anxiety and confusion associated with false positives and diagnostically ambiguous results, for example.³¹⁴ Parents who understand in advance that NBS is merely a *screening*, and not a diagnostic, procedure and that a positive result is not determinative are less likely to experience

308. *Innocent Blood*, *supra* note 138 (explaining that without the proper education the parents are not truly given the option to opt out).

309. Whelan, *supra* note 18, at 448 (describing opt-out programs as “not a true model of consent” but as a mere “substitute for consent”).

310. Liebl et al., *supra* note 271, at 127 (specifying that lack of knowledge was a significant barrier to providing consent).

311. Feuchtbaum et al., *supra* note 195, at 8–10 (discussing the positive effect of having the option to opt out).

312. *Cf. id.* at 9 (most states favor the opt-out approach).

313. See Fyrö & Bodegård, *supra* note 59, at 107, 111 (noting the “persistent anxiety” associated with false positives); *supra* text accompanying notes 58–72.

314. See WHITEHEAD ET AL., *supra* note 303, at 14–19 (describing the anxiety and depression felt by parents following a false positive).

anxiety with respect to a false positive than parents who did not even know their child was screened.³¹⁵ To the extent that an opt-in approach promotes parents' awareness of NBS, this approach might function, in part, as a prophylactic to this concern.

Second, as the panel of diseases screened for expands to include diseases for which there is limited or no ameliorative treatment in the newborn period, the rationale for testing without consent disappears. The entire justification for screening without consent is the idea that the state is protecting newborns from suffering the harms of treatable conditions, which is not true with untreatable conditions.³¹⁶ In this instance, as with storing and doing research on DBS, the *parens patriae* notion used to justify screening treatable conditions without consent does not exist. As a result, the argument for affirmative consent in these cases becomes significantly stronger.

The fact that there is serious consideration of including whole genome or exome sequencing in NBS³¹⁷ should give us even more reason to be skeptical of opt-out approaches, for both of the reasons discussed above. Whatever concerns we might have about expanded panels of NBS with respect to false positives, incidental and ambiguous findings, and information about conditions for which there is no treatment are bound to be magnified considerably by the sheer amount of information that whole genome/exome sequencing (WG/ES) can generate. Indeed, for that reason, there is a very strong case to be made against nudging parents toward consent for WG/ES NBS and a very strong argument for giving parents affirmative choice—i.e., the opt-in approach.

Even if one were to argue that opt-outs are important to “nudge” parents into consenting to testing for serious, treatable conditions, as states expand their NBS panels to include conditions for which there are no treatments or WG/ES, this rationale cannot apply to the full range of screening. Rather than use an opt-out approach for all of the NBS, it would be preferable to tier the decision-making process so that there is only an option to opt out of screening for treatable conditions,

315. See *id.* at 19 (explaining that information reduced this stress).

316. See Faden et al., *supra* note 80, at 1350–51 (discussing the support of parents who believe consent is not necessary for routine testing).

317. See *supra* notes 52–54 and accompanying text.

and perhaps only for those that express in childhood. Parents, however, would have to opt in for the rest. Of course, for the reasons I gave above, I believe opt-in for all NBS is preferable. Moreover, the administrative difficulties of setting up two consent approaches for different types of diseases further argues for a single approach, in this case, opt-in.³¹⁸ But given the strong impetus in favor of opt-out for treatable conditions, it seems extremely important to ensure that consent is *affirmative*, and not presumed, when it comes to conditions for which there is no treatment, especially if they are late-onset conditions.

Finally, my arguments for seeking affirmative consent for the storage and future use of the DBS offer a final reason to advocate for opt-in approaches to NBS generally. Efforts to seek consent for research and storage of samples would effectively necessitate a discussion about NBS generally. It is only a minimal extra step to seek consent for the screening itself. Some might argue that each new decision that parents are confronted with or asked to make complicates and slows down the overall process. It seems difficult, however, to discuss the collection, storage, and research use of DBS without first explaining NBS and its purpose, at least in general terms. Given that parental awareness of NBS is likely to promote successful NBS, and given that parents want to be educated about the program, the general discussions about NBS that an affirmative consent rule would require seem very much in line with what would be required for a discussion of storage and research uses. As a result, promoting parental awareness of NBS through affirmative consent seems well worth the time. While this might not satisfy the notion of fully informed consent, it might achieve the best compromise between parental autonomy and the common good. It fulfills our *prima facie* duties to promote individual autonomy, while also honoring our *prima facie* duties as a society to protect the physical welfare of newborns by informing parents about NBS generally and seeking, rather than simply presuming, their affirmative consent.

318. Feuchtbaum et al., *supra* note 195, at 10–11.

CONCLUSION

As I have argued, the dignitary principle of respect that is central to autonomy and consent should remain central to all aspects of the NBS program from the moment the samples are collected to the moment the state considers using the samples. While autonomy should not be the overriding principle in determining what approach to take, there is a risk in deciding that the state's interest in helping newborns and advancing science will run roughshod over the family's autonomy interests and the child's privacy and future autonomy interests in determining the extent to which he or she wants to participate in research. As we have seen, many of the public goods may actually be advanced by approaches that recognize the value of autonomy and privacy, with appropriate limits, so as not to hinder the ability to protect newborns or engage in certain valuable research projects.

Underlying the goal of achieving the appropriate balance between the public good and individual interests is a third consideration: the need for transparency when the government has control over samples with highly personal information. Whatever balance of autonomy and promotion of research governments choose, they owe a fiduciary obligation to the citizenry to act not only for the benefit of the public, but to assure there is public authorization and transparency. The public's trust in the government is at stake in the development of NBS research programs.³¹⁹ This argues for educating the public not only about the existing NBS policies, but also about new approaches the state is considering so that the public may share in deliberations over the delicate balance between the public and private interests. To quote John Rawls, it is essential for a "well-ordered society" to resolve such difficult matters based on "the ideals and principles expressed by society's conception of political justice, and *conducted open to view on that basis.*"³²⁰ Until the government does a better job of educating parents about the full spectrum of issues and decisions it has made with respect to NBS, this will not be possible. This article is a call to the states to ensure that they move toward such openness.

319. See *supra* notes 249–56 and accompanying text.

320. JOHN RAWLS, POLITICAL LIBERALISM 213 (expanded ed. 2005) (emphasis added).