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Comments: Newborn Screening Programs and Privacy: Shifting Responsibility from the Parent to the Laboratory

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NEWBORN SCREENING PROGRAMS AND PRIVACY: SHIFTING RESPONSIBILITY FROM THE PARENT TO THE LABORATORY

I. INTRODUCTION

Imagine a parent takes her one-month-old child for a checkup only to be told by the pediatrician that the child carries a certain gene indicative of cystic fibrosis.¹ The parent wonders how the pediatrician knows this information since the pediatrician never took a blood sample from the infant.² The pediatrician explains that all newborns are screened at birth for a panel of genetic diseases as is mandated by the government.³ In many cases, however, this testing is done without proper informed consent of the parents, who are unaware that a DNA sample is being taken.⁴ The above story is true and resulted in multiple lawsuits in several different states claiming that the infant's privacy was violated.⁵

These cases resulted in reform regarding consent to newborn screenings,⁶ but problems still remain. The major problem facing newborn screening programs is the fact that technology has progressed tremendously since the inception of such programs.⁷ Newborn screening began in the 1960s and tested only a small number of diseases.⁸ Since then, technology has developed to the point where we cannot only test infants for a multitude of diseases, but we can sequence an entire genome and are on a path to determining the personality traits as well as physical traits of the

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1. Elizabeth Cohen, *The Government Has Your Baby's DNA*, CNN (Feb. 4, 2010), <http://www.cnn.com/2010/HEALTH/02/04/baby.dna.government/index.html>.
 2. *Id.*
 3. *Id.*
 4. *Id.*
 5. *Id.*; see, e.g., *Higgins v. Tex. Dep't of Health Servs.*, 801 F. Supp. 2d 541, 544, 546 (W.D. Tex. 2011); *Bearder v. State*, 806 N.W.2d 766, 769 (Minn. 2011).
 6. See Dena S. Davis, *Opportunistic Testing: The Death of Informed Consent?* 23 HEALTH MATRIX 35, 42 (2013); Rachel L. Schweers, Comment, *Newborn Screening Programs: How Do We Best Protect Privacy Rights While Ensuring Optimal Newborn Health?*, 61 DEPAUL L. REV. 869, 909 (2012).
 7. See Davis, *supra* note 6, 42.
 8. *Id.* at 41–42.

patient.⁹ This progression in technology raises many privacy-rights issues, not the least of which is that the essence of a person is in danger of being inappropriately accessed or disseminated.¹⁰

The solution is to, first and foremost, fundamentally change the way we view genetic information and move from our outdated way of thinking to a more modern way of thinking that is aware of the dangers new technology may pose. To do this, this Comment proposes that we implement a two-pronged approach.¹¹ First, legislation should require parental informed consent in order for the infant to participate in the newborn screening program.¹² Second, legislation should place the onus on healthcare providers, corporations, research institutions, and other entities not to violate the parents' or infant's privacy rights.¹³ This would effectively shift the burden away from the parent and would not put the infant's genetic information at risk.¹⁴ Additionally, new legislation must be implemented that would focus specifically on encompassing and protecting all genetic information, no matter its form or method of sampling.¹⁵

In Part II, this Comment will give an overview of the history of genetic testing as well as history specific to newborn screenings, including current and potential future technologies.¹⁶ The following section will discuss positive aspects of newborn screenings as well as the current newborn screening process and current legislation and governmental policies relating to newborn screening programs.¹⁷ In Part IV, this Comment will discuss negative implications associated with current newborn screening programs in the United States.¹⁸ In that same section, this Comment will discuss different theories as to how to solve the problems created by current newborn screening

9. *Frequently Asked Questions About Genetic Testing*, NAT'L HUMAN GENOME RESEARCH INST., <http://www.genome.gov/19516567#al-5> (last updated Mar. 11, 2014).

10. *See* Cohen, *supra* note 1; *see also* Stefan Timmermans, *Genetic Screening: Every Newborn a Patient*, LOS ANGELES TIMES (July 19, 2013), <http://articles.latimes.com/2013/jul/19/opinion/la-oe-timmermans-infant-genetic-screening-20130719> (discussing the harm parents suffer from false positive results).

11. *See infra* Part IV–V.

12. *See infra* Part IV.

13. *See infra* Part V.

14. *See infra* Part V.

15. *See, e.g., Health Information Privacy*, U.S. DEP'T OF HEALTH & HUMAN SERVS., <http://www.hhs.gov/ocr/privacy/> (last visited Nov. 17, 2014).

16. *See infra* Part II.

17. *See infra* Part III.

18. *See infra* Part IV.

programs.¹⁹ Finally, Part V of this Comment will set forth an overview of legislative guidelines which would solve many of the problems facing newborn screening programs.²⁰

II. OVERVIEW OF GENETIC TESTING AND HISTORY OF NEWBORN GENETIC SCREENINGS

A. Genetic Testing Overview

Genetic tests use drawn blood and other tissue to determine if an individual carries certain genetic disorders.²¹ To do this, the drawn blood is analyzed to determine if any changes in chromosomes, genes, or proteins exist,²² and “[t]he results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person’s chance of developing or passing on a genetic disorder.”²³ There are several different methods used to test the patient’s blood sample.²⁴

First, “molecular genetic tests” analyze “single genes . . . to identify variations or mutations that lead to a genetic disorder.”²⁵ Second, “chromosomal genetic tests” examine entire “chromosomes . . . to see if there are large genetic changes, such as an extra copy of a chromosome, that cause a genetic condition.”²⁶ Third, “biochemical genetic tests” look at “the amount or activity level of proteins; abnormalities in either can indicate changes to the DNA that result in a genetic disorder.”²⁷ Additionally, testing can be done using direct-to-consumer (DTC) testing kits.²⁸ These kits allow the consumer to test for certain diseases in their own home.²⁹ These DTC tests can

19. See *infra* Part IV.

20. See *infra* Part V.

21. *Genetic Testing*, MEDLINEPLUS, <http://www.nlm.nih.gov/medlineplus/genetictesting.html> (last updated July 11, 2014). The sample analyzes the deoxyribonucleic acid (DNA) in the patient’s tissue to obtain the results. *Tests and Procedures: Genetic Testing*, MAYO CLINIC (July 19, 2013), <http://www.mayoclinic.org/tests-procedures/genetic-testing/basics/definition/PRC-20014802>.

22. *What is Genetic Testing?*, GENETICS HOME REFERENCE (July 7, 2014), <http://www.ghr.nlm.nih.gov/handbook/testing/genetictesting>.

23. *Id.*

24. *Id.*

25. *Id.*

26. *Id.*

27. *Id.* Generally, newborn screenings use the biochemical test technique to determine the existence of genetic conditions in the baby’s DNA. *Frequently Asked Questions About Genetic Testing*, *supra* note 9.

28. *Frequently Asked Questions About Genetic Testing*, *supra* note 9.

29. See *id.*

identify changes in some or all of an individual's genome to detect the risk of developing particular diseases, to help develop a personalized nutrition plan, or to detect genetic markers that indicate an individual's physical traits, ancestry, or personality.³⁰ For adults, this testing is voluntary.³¹

In 2003, researchers at the Human Genome Research Institute completed their goal of mapping and sequencing the entire human genome.³² Human genome "[s]equencing involves determining the order of the chemical units of DNA" and "helps determine inherited traits, like susceptibility to some diseases."³³ Once the sequence is known, scientists can then determine "the kind of genetic information that is carried in a particular segment of DNA."³⁴ The sequencing of the genome has given researchers a "virtual blueprint of the human being," and future research will attempt to make sense of that "blueprint."³⁵ Additionally, sequencing the human genome is

30. *Id.*

31. Ariel Bleicher, *Perils of Newborn Screening*, 307 SCI. AM. 30, 30 (July 1, 2012), available at <http://www.scientificamerican.com/article/perils-of-newborn-screening/>.

32. *All About the Human Genome Project (HGP)*, NAT'L HUMAN GENOME RESEARCH INST., <http://www.genome.gov/10001772> (last updated Mar. 18, 2014). This gave researchers the ability "to read nature's complete genetic blueprint for building a human being." *Id.*

33. Andrew Pollack, *Company Unveils DNA Sequencing Device Meant to be Portable, Disposable and Cheap*, N.Y. TIMES (Feb. 17, 2012), <http://www.nytimes.com/2012/02/18/health/oxford-nanopore-unveils-tiny-dna-sequencing-device.html>.

All genes are made up of stretches of these four bases, arranged in different ways and in different lengths. HGP [Human Genome Project] researchers have deciphered the human genome in three major ways: determining the order, or "sequence," of all the bases in our genome's DNA; making maps that show the locations of genes for major sections of all our chromosomes; and producing what are called linkage maps . . . through which inherited traits (such as those for genetic disease) can be tracked over generations.

An Overview of the Human Genome Project, NAT'L HUMAN GENOME RESEARCH INST., <http://www.genome.gov/12011238> (last reviewed Nov. 8, 2012).

34. *DNA Sequencing*, NAT'L HUMAN GENOME RESEARCH INST., <http://www.genome.gov/10001177> (last reviewed Dec. 27, 2011).

35. *See id.*

However, this accomplishment should be viewed not as an end in itself, but rather as a starting point for even more exciting research. Armed with the human genome sequence, researchers are now trying to unravel some of biology's most complicated processes: how a baby develops from a single cell, how genes coordinate the functions of tissues and organs, how disease predisposition occurs and how the human brain works.

expensive, but the National Human Genome Research Institute is in the process of lowering the cost of sequencing the genome to a more reasonable price.³⁶

B. Newborn Screening History

Newborn screening began in the 1960s as techniques were developed to determine whether an infant was affected by Phenylketonuria (PKU).³⁷ In 1975, 90% of newborns were tested for PKU.³⁸ Soon thereafter, states began implementing similar tests to determine the probability of contracting other diseases for which treatments are available.³⁹ Currently, all states require newborn screening for markers of medical disorders.⁴⁰

In the 1990s, the tandem mass spectrometry tool enabled technicians to detect more disorders using only a single drop of blood⁴¹ and significantly increased the number of diseases tested by the screening panel.⁴² Further, “research has indicated that [mass spectrometry] has a false positive rate up to ten-fold lower for PKU screening than the best method previously available;” this makes it extremely effective.⁴³ In addition, genome-sequencing technologies will eventually become inexpensive enough that they can be used routinely and thereby cause another rapid expansion for newborn screenings.⁴⁴ To this point, a British company recently announced the sale of a disposable gene-sequencing device that can plug into a

Id.

36. Davis, *supra* note 6, at 42.

37. Schweers, *supra* note 6, at 875. PKU is “a rare genetic disorder that can cause mental retardation but is treatable by diet modifications if detected early.” *Id.*

38. *Id.*

39. Bleicher, *supra* note 31. Some of these diseases “includ[ed] congenital hypothyroidism and sickle cell disease.” *Id.*

40. *Id.*

41. *Id.* “A mass spectrometer sorts and counts [variously] sized molecules in the blood, somewhat like a change machine sorts coins. Unusually high levels of certain molecules indicate the enzymes that normally break down these molecules are missing or deficient, which in turn suggests a genetic disorder.” *Id.*

42. Lauren E. Fisher, Comment, *The Use of Tandem Mass Spectrometry in Newborn Screening: Australia’s Experience and Its Implications for United States Policy*, 15 PAC. RIM L. & POL’Y J. 137, 145 (2006).

43. *Id.* at 147.

44. Bleicher, *supra* note 31 (citing Lainie Friedman Ross, an ethicist and pediatrician at the University of Chicago). Ross also says “there is the potential to test for hundreds of conditions we don’t fully understand.” *Id.*

computer to deliver its results; all for a price of \$900.⁴⁵ The private sector currently uses an even more effective way to test a patient's blood and sequence their genome.⁴⁶ This new technology uses a microchip to "screen directly for the genetic basis of various disorders."⁴⁷

III. NEWBORN SCREENING PROCESS AND LEGISLATION

A. Newborn Screening Process

Typically, a blood test is performed 24 to 48 hours after the baby is born.⁴⁸ To collect a blood sample, the baby's heel is pricked to obtain several drops of blood.⁴⁹ This blood is then placed on a piece of special paper and sent for laboratory analysis.⁵⁰ If there are abnormalities, "additional testing is required to confirm the [first test's] result."⁵¹ Moreover, "[d]ifferent labs have different procedures for notifying families and pediatricians of the results."⁵² Sometimes the results are sent to the hospital where the child was born; sometimes they are sent directly to the doctor.⁵³ Once the results are determined, laboratories keep the samples as part of a databank in accordance with the laws of the state in which the sample was taken.⁵⁴

B. Newborn Screening Program Policies and Legislation

Currently, "newborn screening varies from state to state."⁵⁵ Some states test for more than twenty-nine different conditions, while other states test for fewer than fifteen conditions.⁵⁶ As technology has

45. Pollack, *supra* note 33. This is not the only attempt at a portable, inexpensive sequencing device (though this seems to be the most successful as of yet). *See id.*

46. *See* Davis, *supra* note 6, at 41–42.

47. *Id.* at 42.

48. *Newborn Screening*, GENETICS HOME REFERENCE (July 7, 2014), <http://ghr.nlm.nih.gov/nbs>.

49. *Id.*

50. *Id.*

51. *Id.*

52. *Newborn Screening Tests*, KIDSHHEALTH, http://kidshealth.org/parent/system/medical/newborn_screening_tests.html (last visited June 27, 2014).

53. *Id.*

54. *See* Davis, *supra* note 6, at 47–48.

55. *Newborn Screening*, *supra* note 48. There are programs in all 50 states and the District of Columbia. Schweers, *supra* note 6, at 879.

56. *See* Schweers, *supra* note 6, at 879 n.48. For a full list of conditions screened for by state, *see* *Conditions Screened by State*, BABY'S FIRST TEST, <http://www.babysfirsttest.org/newborn-screening/states> (last visited Nov. 17, 2014).

improved, it has become easier to test for more conditions, and each state has its own ways of determining which conditions to test for.⁵⁷ In addition, only the District of Columbia requires parental informed consent, while not one of the fifty states does.⁵⁸ Only a few states require explicit consent from the infant's parents for screening the infant's blood.⁵⁹ Alternatively, some states have a "mixed-consent program" that allows parents to opt-out for "routine" testing based on religious beliefs, while certain "optional" tests are opt-in programs.⁶⁰

States also have their own statutes and regulations regarding what is done with the infant's blood sample after the screening is complete.⁶¹ In many cases, the samples are stored indefinitely.⁶² Only a few states have regulations requiring consent in order to use the samples in future research.⁶³

IV. THE CURRENT AND FUTURE LANDSCAPE OF NEWBORN SCREENING PROGRAMS CREATE PRIVACY PROBLEMS FOR THE PARENT AND INFANT

Current legislation, procedures, and technology related to newborn screenings create both positive and negative implications that can affect the infant, the parents of the infant, and public health.⁶⁴ This section will first discuss some positive aspects of newborn screenings.⁶⁵ Next, it will discuss some current problems associated with newborn screening programs as well as future issues that may arise.⁶⁶

57. See Davis, *supra* note 6, at 42; Schweers, *supra* note 6, at 879. The determination is further complicated by the fact that there are usually a number of different advocacy groups which campaign to have certain conditions added. See *id.*

58. Timmermans, *supra* note 10. "Most states collect and test newborn samples after providing little, if any, educational primers to the parents. In fact, Michigan, Montana, Nebraska, and South Dakota do not currently provide any statutory grounds for refusal to participate." Schweers, *supra* note 6, at 880. See MICH. COMP. LAWS § 333.5431 (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).

59. See Schweers, *supra* note 6, at 880. These programs are considered "opt-in." *Id.*

60. *Id.* 880–81.

61. See Cohen, *supra* note 1.

62. *Id.* Some states allow parents to request that their infant's DNA sample be destroyed. *Id.*

63. See Davis, *supra* note 6, at 47–48. For example, in Michigan the BioTrust for Health was created which requires parental permission before stored samples are used in research. *Id.*

64. *Id.* at 43–44.

65. See *infra* Part IV.A.

66. See *infra* Part IV.A–B.

There are many reasons why newborn screenings are a great idea for both the infant and the public at large.⁶⁷ Sometimes medical conditions cannot be seen through a traditional physical exam.⁶⁸ Newborn screenings allow doctors to provide better care to the infant and can prevent serious problems, such as brain and organ damage as well as death.⁶⁹ In many cases, if it is determined that an infant has the genetic markers of a disease, then the doctor can prescribe a course of treatment or refer the parents to an appropriate specialist and prevent the infant from contracting that disease.⁷⁰ If more infants are tested and subsequently treated for these diseases, the better the health of the public at large.

A. Improper Selection Of Conditions To Be Tested For In A Newborn

1. Problems Raised by Adult-Onset And Nontreatable Disorders

While there are certainly positive aspects to newborn screening programs as they are structured today, there are many negative aspects as well.⁷¹ Ellen Wright Clayton, M.D., J.D., Professor of Genetics and Health Policy at Vanderbilt University, states that it is not a good idea to test newborns for adult-onset disorders and that informed consent should be required to screen for untreatable disorders.⁷² Clayton further questions “why this screening should be done 1) by the state and 2) in the newborn period.”⁷³ First, she argues that clinicians should be the ones to speak with parents about the positives and negatives involved in newborn screening, rather than

67. *Newborn Screening*, CTR. FOR DISEASE CONTROL & PREVENTION, <http://www.cdc.gov/newbornscreening/> (last viewed June 27, 2014).

68. *Pediatric Genetics: Newborn Screening*, CTR. FOR DISEASE CONTROL AND PREVENTION, http://www.cdc.gov/ncbddd/pediatricgenetics/newborn_screening.html (last updated May 13, 2013).

69. *Id.*

70. *Id.* For example, a doctor can prescribe a treatment plan for an infant with hypothyroidism, which is detected through newborn screening, to avoid the slowed growth and brain damage associated with hypothyroidism. *Id.*

71. *See, e.g.*, Bleicher, *supra* note 31 (stating that parents of an infant diagnosed with latent conditions tend to fret, pursue risky tests, and avoid routine treatment). An additional problem, which will not be addressed, is that in some states courts have held that parents either do not have the right to know the results of their infant’s screening, or the laboratory is not required to notify the parents of the results. *See In re Carter*, 653 S.E.2d 860, 866 (Ga. Ct. App. 2007); *Hanshaw v. River Valley Health Sys.*, 789 N.E.2d 680, 687 (Ohio Ct. App. 2003).

72. Ellen Wright Clayton, *Ten Fingers, Ten Toes: Newborn Screening for Untreatable Disorders*, 19 HEALTH MATRIX 199, 202–03 (2009).

73. *Id.*

state officials.⁷⁴ If state-run programs are tasked with obtaining parental consent, then it would “require a huge sea change since the vast majority of these programs have never sought parental permission.”⁷⁵ Second, Clayton argues that public and private clinicians continue to screen children for growth and development as well as other health problems as those children age.⁷⁶ She argues for a staged approach to testing for disorders, where different disorders are tested for at different times in a child’s life.⁷⁷

While testing newborns for adult-onset and nontreatable disorders create the problems described by Clayton, that should not mean that screening for such disorders at birth should be eliminated.⁷⁸ The real problem is not that these diseases are untreatable or don’t manifest until the child is older, but rather that parents are not educated regarding their child’s screening and are not given the chance to make an informed decision.⁷⁹ Therefore, a tiered approach, which would create an isolated tier dedicated solely to testing for adult-onset and nontreatable disorders and which requires parental informed consent, would be appropriate.

Under many circumstances, clinicians are the best choice to educate parents before the child is born as well as during the child’s developing years.⁸⁰ However, many parents either do not have access to consistent medical care for their child or choose not to seek out medical care.⁸¹ In such cases, a state program may be a more appropriate choice to educate parents who do not have regular access to a clinician’s expertise.⁸²

2. Lack Of Consensus As To Which Conditions To Test For

A glaring problem with current screening programs in the United States is that there is no consensus as to how many diseases an infant should be tested for.⁸³ In order to rectify this situation, in the 1990s the Health Resources and Services Administration commissioned a

74. *Id.* at 202.

75. *Id.*

76. *Id.* at 201.

77. *See id.* at 202.

78. *See, e.g.,* Schweers, *supra* note 6, at 875.

79. *See id.* at 902.

80. Clayton, *supra* note 72, at 201–202.

81. The Med. Child Support Working Group’s Report, *21 Million Children’s Health: Our Shared Responsibility*, PROMOTING RESPONSIBLE FATHERHOOD, <http://fatherhood.hhs.gov/medsupport00/chap1.htm> (last updated Nov. 27, 2000).

82. *See* Clayton, *supra* note 72, at 201–02.

83. Bleicher, *supra* note 31.

researcher to review the scientific literature and determine which tests would benefit newborns.⁸⁴ The report submitted recommended that all states screen for twenty-nine conditions and avoid screening for certain other conditions.⁸⁵ However, this did not stop some states from testing for additional disorders, even though the report advised against it.⁸⁶

The additional conditions those states tested for are not always those that doctors can accurately predict or treat.⁸⁷ For example, New York has been testing infants for Krabbe disease since 2006.⁸⁸ However, researchers do not currently understand Krabbe well enough to know when, or even if, the infant will develop symptoms (and, consequently, treatment is ineffective and inaccurate).⁸⁹ As a result, parents “do not know what to do with the information they receive from doctors,” and many “begin to worry.”⁹⁰

Forcing parents (and infants) to participate in such a public health program is unfair.⁹¹ Given the expansion of genetic testing technology, and as genome-sequencing technologies become less expensive, there is the potential to test for hundreds of conditions which researchers and doctors do not understand.⁹² It would seem unfair to force parents to submit their children to such a program when adults themselves can voluntarily decide not to participate in such a program on their own.⁹³ While it is exciting to have the capability to test for so many things, “[w]e shouldn’t just add these things because we can.”⁹⁴ Instead, legislation should attempt to create some uniformity by initially limiting states to testing only for

84. *Id.*; *Recommended Uniform Screening Panel*, HEALTH RESOURCES AND SERVS. ADMIN., <http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendedpanel/> (last updated Apr. 2013) (recommending that thirty-one core conditions be uniformly tested).

85. Bleicher, *supra* note 31. It was found that some tests did not cause enough good to warrant being on the list. *Id.*

86. *Id.*

87. *Id.*

88. *Id.*

89. *See id.*

90. *Id.*

91. *Id.* However, it is true that testing for untreatable disorders can avoid delays in diagnosis, “providing parents with information about their children’s health, allowing the earlier initiation of interventions, learning more about the natural history of these disorders, providing reproductive information to the family, and creating more opportunities to support these families.” Clayton, *supra* note 72, at 201.

92. Bleicher, *supra* note 31.

93. *See id.*

94. *Id.* (quoting Jeff Botkin, a medical ethicist at the University of Utah School of Medicine).

certain recommended conditions.⁹⁵ For any additional conditions to be added to the screening process those conditions should be put through rigorous tests to determine the category that the condition belongs in.⁹⁶ For example, a disorder might satisfy the same criteria as those disorders which are uniformly tested.⁹⁷ Alternatively, disorders may fall under a separate category of conditions for which testing is not as reliable and/or treatment is not as successful.⁹⁸ This categorization makes sure that conditions are not being tested for just because we can test for them.⁹⁹ Rather, parents can become educated regarding the different categories and can make an informed decision as to whether they consent to having their child tested for those disorders.¹⁰⁰

B. A Lack Of Informed Consent Places The Burden On Parents

Another negative aspect of current newborn screening programs has to do with inadequate parental informed consent.¹⁰¹ Currently, informed consent to participate in a newborn screening program is only required in the District of Columbia and not in any other state.¹⁰² In many states, even though the parents are allowed to refuse, this right is meaningless, because parents are frequently not told about the test before the sample is taken.¹⁰³

Dena Davis,¹⁰⁴ in her article *Opportunistic Testing: The Death of Informed Consent?*, focuses attention on the problems associated with a lack of informed consent in newborn screening programs.¹⁰⁵

95. See *Recommended Uniform Screening Panel*, *supra* note 84; Schweers, *supra* note 6, at 903.

96. See AM. ACAD. FAMILY PHYSICIANS, ISSUE BRIEF: NEWBORN SCREENING, (January 2006), <http://www.aafp.org/dam/AAFP/documents/advocacy/workforce/scope/ES-NewbornScreening-0106.pdf> (recommending possible testing procedures for states including: (1) “that their states give consideration to mandatory newborn screening for those [diseases] for which the evidence is most rigorously supportive”; (2) “that families be appropriately informed for consent”; and (3) that “[f]amily physicians and their office staffs should prepare to educate families concerning newborn screening, and to respond to questions from families concerning positive tests”).

97. See Schweers, *supra* note 6, at 903.

98. See *id.*

99. See *id.*

100. See *id.*

101. See Timmermans, *supra* note 10.

102. *Id.*

103. Davis, *supra* note 6, at 42.

104. Davis is the Presidential Endowed Chair in Health, Humanities/Social Sciences at Lehigh University. *Id.* at 35.

105. *Id.*

First, Davis explains that there is a debate regarding how to choose which conditions are added to existing newborn screening tests.¹⁰⁶ Underlying this debate is the concept of informed consent (or lack thereof)¹⁰⁷ and that “[a] test given without parental consent can only ethically be defended on the grounds of potential benefit to children, backed up by strong evidence.”¹⁰⁸ Davis warns that while some conditions may pass this test, others may not.¹⁰⁹

Additionally, Davis discusses how false positives can be a major problem, especially if no informed consent is required.¹¹⁰ In order to avoid these problems, Davis suggests that parents need to become more involved in newborn screening programs.¹¹¹ To do this, parents need to receive as much information as possible during the prenatal period in order for them to acclimate to the information and ask questions.¹¹²

Davis is absolutely correct, and the idea that parents must be involved and educated regarding newborn screenings is the exact reason why the burden for protecting the infant’s privacy must be shifted away from the parents.¹¹³ For example, society automatically assumes that patients should be informed about the procedure, risks involved, and prognosis relating to surgery and other procedures performed by physicians.¹¹⁴ Society needs to begin thinking the same way regarding newborn screening programs in order to properly protect the parent’s and, more importantly, the infant’s privacy as screening technologies become more advanced.

Further exacerbating the problem associated with this lack of proper informed consent, hospitals use a tactic known as

106. *Id.* at 42.

107. *Id.* The author writes that “the ease with which new screens can be added threatens parental rights to be aware of and consent to the medical tests conducted on their children . . .” *Id.* at 48.

108. *Id.* at 42-43.

109. *Id.* at 43. PKU probably passes this test, but “[s]creening for cystic fibrosis, for example, has been controversial because not everyone agrees that there is a medical advantage to early, presymptomatic diagnosis. However, studies show that early diagnosis of cystic fibrosis prevents malnutrition and improves children’s growth and cognitive function.” *Id.* at 43. In addition, we need to question what counts as a “benefit.” *See id.*

110. *Id.* at 44.

111. *Id.* at 45.

112. *Id.* The author suggests several ways of doing this, including information from the internet, brochures, etc. *Id.* at 45-46.

113. *See infra* Part V.

114. *See* Owen A. Anderson & I Mike J. Wearne, *Informed Consent for Elective Surgery—What is Best Practice?*, 100 J. OF ROYAL SOC’Y OF MED. 97, 97-99 (2007), available at <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC1791005/>.

opportunistic testing.¹¹⁵ Put simply, “Opportunistic testing or screening is medical testing that makes use of an ‘opportunity’ engendered by some other test . . . to which the patient is accustomed or has already given consent.”¹¹⁶ In a situation in which a blood sample is used, opportunistic testing means that the “sample is drawn for an established purpose and then one or more extra tests are added” to that initial test.¹¹⁷ This creates obvious problems, because parents may give consent to one test, but without their knowledge be subjected to additional tests.¹¹⁸ How can this be acceptable, when it would not be acceptable to obtain consent for a particular heart procedure, for example, and then allow the surgeon to perform an additional procedure for which proper, explicit consent was not obtained?

Newborn screening program advocates have also tried to add more conditions to the testing list as well as incorporate genetic sequencing into the program.¹¹⁹ The National Institute of Child Health and Human Development and the National Human Genome Research Institute are currently funding research proposals to integrate gene sequencing technologies into newborn screening programs which may allow us to provide every newborn with a full genetic printout at birth.¹²⁰ As noted above, however, sometimes these tests can yield false positives or can test for diseases that are untreatable.¹²¹ A lack of informed consent means that parents will be notified that their child has a condition they did not know was even tested for during the initial screening.¹²² This can devastate parents and cause them to worry.¹²³

Furthermore, if parents do not know exactly what is being done to their child, the information being collected (in this case the infant’s DNA) can be used for nefarious purposes.¹²⁴ These purposes can

115. See Davis, *supra* note 6, at 36.

116. *Id.* The term “implies ‘piggybacking’ one intervention onto another and thus exploiting an opportunity.” *Id.*

117. *Id.*

118. *Id.*

119. Timmermans, *supra* note 10.

120. *Id.*

121. *Id.*

122. *Id.*

123. *Id.*

124. See, e.g., Becca Aaronson, *Lawsuit Alleges DSHS Sold Baby DNA Samples*, TEX. TRIBUNE (Dec. 8, 2010), <http://www.texastribune.org/2010/12/08/lawsuit-alleges-dshs-sold-baby-dna-samples/> (indicating that the Texas Department of State Health

include unlawfully distributing infant blood samples or giving samples to outside researchers without consent.¹²⁵

Rachel L. Schweers points out correctly that since each state has its own rules, there is no true uniformity regarding these programs, and as a result privacy rights are eroded and in some cases violated.¹²⁶ Schweers suggests that there should be a model newborn screening statute among all programs throughout the country.¹²⁷ This model should require explicit informed consent from the parents.¹²⁸ In addition, discussions with healthcare providers should include (1) the conditions tested, (2) “the disclosure of the results,” (3) “the retention time or . . . destruction of the sample,” and (4) “the permissible research uses of the sample.”¹²⁹ Schweers suggests that there should be a tiered system regarding which conditions are tested for and what amount of consent is required.¹³⁰ Schweers writes that the only standard conditions which should be tested for are those that are “known to benefit from diagnosis and treatment at an early stage in life.”¹³¹ With regard to permissible research uses, Schweers suggests “parents should be given the option of electing sample retention for no longer than eighteen years”¹³²

C. Genome Sequencing As The Future of Newborn Screenings

In the field of genomics, the goal of technology moving forward is to have the ability to sequence an entire genome quickly, accurately, and for low cost.¹³³ Cost is one of the most significant barriers to creating widely available sequencing opportunities, but next-generation technologies indicate that low-cost genome sequencing is on the way.¹³⁴ Further, genome sequencing has tremendous value:

Services sold and distributed infant blood samples to outside laboratories without parental consent).

125. *See id.*; Cohen, *supra* note 1.

126. Schweers, *supra* note 6, at 879.

127. *Id.* at 900.

128. *Id.* 900–01.

129. *Id.* at 900–901.

130. *See id.* at 903.

131. *Id.*

132. *Id.* at 904.

133. Mark A. Rothstein, Column, *Currents in Contemporary Bioethics: The Case Against Precipitous, Population-Wide, Whole-Genome Sequencing*, 40 J.L. MED. & ETHICS 682, 682 (2012). Mr. Rothstein “is the Herbert F. Boehl Chair of Law and Medicine and Director, Institute for Bioethics, Health Policy and Law, University of Louisville School of Medicine.” *Id.*

134. *See id.*

[I]n elucidating the genetic etiology of rare disorders, in identifying atypical variants in common diseases, in determining pharmacogenomically appropriate drugs and dosages, in performing tumor genome sequencing, and in aiding other clinical applications for the diagnosis and treatment of individuals who are *symptomatic* or whose family health history places them at substantial risk.¹³⁵

Moreover, the development, promotion, and use of new sequencing technologies will not slow down as a result of societal opinions, and as soon as it is technically and financially feasible, population-wide genome sequencing will occur.¹³⁶

Mark Rothstein critically evaluates population-wide, whole-genome sequencing and concludes that the costs associated with such sequencing outweigh any potential benefits.¹³⁷ Rothstein's concerns with population-wide, whole-genome sequencing are numerous (only a few of which are discussed here).¹³⁸

First, “[i]t is not clear how sequence data [is] interpreted.”¹³⁹ Currently, no standardized software exists for performing sequencing, and it is not clear whether the FDA would have to get involved in such a process.¹⁴⁰ Second, Rothstein questions whether health care providers have the expertise and time to translate the genome sequence data and develop prevention and treatment plans for each patient.¹⁴¹

Rothstein further argues that informed consent should be conducted in the clinical setting, but the sheer magnitude of information obtained from sequencing the entire genome would create numerous

135. *Id.* (emphasis in original) (footnotes omitted).

136. *Id.*

137. *Id.*

138. *Id.* at 683–87.

139. *Id.* at 684.

140. *See id.*

141. *Id.* at 684–85.

Physicians who routinely deal with genetics include obstetricians, pediatricians, neurologists, and oncologists. Primary care physicians generally do not have significant training or experience in traditional clinical genetics, let alone the increasingly arcane world of molecular genetics and genomics. Furthermore, third-party payers may not pay for genetic counseling, and in all but 5 states genetic counselors cannot bill for their services independent of an affiliated clinical geneticist.

Id. at 685.

problems in obtaining proper informed consent.¹⁴² Rothstein finds that this flaw is almost impossible to overcome, unless there is a major revolution in the healthcare field.¹⁴³

Finally, Rothstein argues that there is an absence of health privacy and discrimination protections.¹⁴⁴ Mainly, Rothstein points to the Health Insurance Portability and Accountability Act (HIPAA) and the Genetic Information Nondiscrimination Act (GINA) as flawed legislation whose flaws are compounded when a large volume of information is generated, as is the case with whole-genome sequencing.¹⁴⁵ Without solutions for the issues with this legislation, a patient's privacy and nondiscrimination rights are in jeopardy.¹⁴⁶

As discussed above, a tiered approach is an appropriate way to solve some of the above-mentioned negative aspects of newborn screening programs.¹⁴⁷ This is especially true when we consider that the future of genetic testing—whole-genome sequencing—is becoming less expensive and more accurate.¹⁴⁸ As sequencing begins to enter the newborn screening arena, we must be cognizant of the risks which surface if there is a severe lack of uniformity and education among parents.¹⁴⁹ A tiered system which creates a separate tier for genome sequencing would help society to adjust and face the reality that genome sequencing presents.¹⁵⁰

142. *Id.* “[A]s a result of sequencing, an average person will have about 100 genetic risks that will need to be discussed. If each risk were covered in 3 minutes, it would take 5 hours of patient contact to explain the risks.” *Id.*

143. *Id.*

144. *Id.* at 686–87.

145. *Id.* at 686.

146. *See id.*

147. *See supra* Part IV.

148. *See* Bleicher, *supra* note 31; Pollack, *supra* note 33.

149. Audrey Tluczek et al., *Newborn Screening: An Appeal for Improved Parent Education*, 23 J. PERINATAL & NEONATAL NURSING 326, 333 (2009), available at <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2947955/>.

150. *See* Schweers, *supra* note 6, at 903-04; Rothstein, *supra* note 133, at 682–83.

V. AS A SOCIETY WE NEED TO CHANGE THE WAY WE LOOK AT GENETIC INFORMATION AND SHIFT THE BURDEN OF PRIVACY TO LABORATORIES AND HEALTHCARE PROVIDERS

A. Society Needs To Fundamentally Change The Way We Look At Genetic Information

The United States has an outdated view regarding the privacy of newborn screenings.¹⁵¹ When newborn screening programs were first implemented and related legislation was created, genetic testing technology was not as advanced as it is today¹⁵² (we could only test for a few diseases and not much else).¹⁵³ What has changed since then is that we have now unlocked the human genome and have begun reading and analyzing the sequence to learn as much as we can about human DNA.¹⁵⁴ What this means is that we can now not only determine an individual's predisposition for a multitude of diseases, but we can identify personality traits and physical traits as well.¹⁵⁵ It is true that we have not progressed too far into deciphering personality and physical traits, but that ability is not so far in the distant future.¹⁵⁶ The power that this technology holds creates many problems that our old way of thinking is woefully incapable of solving.¹⁵⁷

Society needs to change its thinking regarding genetic information. Genetic information is the most personal information a person can have.¹⁵⁸ An individual's genetic code can be considered a "coded future diary"¹⁵⁹ and "the ultimate answer to the commandment

151. See Jonathan Hsu, Note, *Genetic Testing: Balancing Preventative Medicine with Privacy and Nondiscrimination*, 6 J.L. & POL'Y FOR INFO. SOC'Y 557, 558–59 (2011).

152. See Diane B. Paul, *Promoting Safe and Effective Genetic Testing in the United States*, App. 5: *The History of Newborn Phenylketonuria Screening in the U.S.*, LAW, SCIENCE & PUBLIC HEALTH PROGRAM SITE OF LA. STATE UNIV. (Sept. 1997), <http://biotech.law.lsu.edu/research/fed/tfgt/appendix5.htm>; *All About the Human Genome Project (HGP)*, *supra* note 32.

153. See Paul, *supra* note 152.

154. See Paul Bisceglia, *Cracking the Code of the Human Genome*, SMITHSONIAN (June 14, 2013), <http://www.smithsonianmag.com/smithsonian-institution/unlock-the-science-and-ethics-of-the-human-genome-in-a-new-exhibit-at-the-natural-history-museum-559673/?no-ist>.

155. See *supra* note 9 and accompanying text.

156. See Davis, *supra* note 6, at 41–42.

157. See *supra* Part IV.

158. See *All About the Human Genome Project (HGP)*, *supra* note 32.

159. Hsu, *supra* note 151, at 573.

'[k]now thyself.'"¹⁶⁰ It can be argued that it is even more personal and private than an individual's social security number.¹⁶¹ Such personal information must be protected to the greatest extent possible, and while new parents have neither the appropriate information nor the ability to refuse newborn screenings, the burden is on them to affirmatively protect their and their child's privacy.¹⁶²

This Comment proposes that we implement a two-pronged approach to solve this issue.¹⁶³ First, legislation should require parental informed consent in order for the infant to participate in the newborn screening program.¹⁶⁴ This would mean a new generation of parents would be better informed about what genetic information is, what current (and potential future) technology exists, and the positives and negatives of genetic testing.¹⁶⁵ Second, legislation should place the onus on healthcare providers, corporations, research institutions, and other entities not to violate the parents' or infant's privacy rights.¹⁶⁶ This would effectively shift the burden away from the parent and would not put the infant's genetic information at as great a risk.¹⁶⁷

B. Restructuring Current Newborn Screening Programs

One major issue to consider regarding newborn screening programs is which conditions to test for.¹⁶⁸ Obviously, there are certain diseases that are treatable if detected early,¹⁶⁹ and testing and treating those conditions can make a previously difficult or shortened life more normal.¹⁷⁰ It is less clear whether adult onset or untreatable disorders should be included as well.¹⁷¹ Clayton suggests that clinicians should be the only ones who should obtain informed

160. *Id.* (quoting JERRY E. BISHOP & MICHAEL WALDHOLZ, *GENOME: THE STORY OF THE MOST ASTONISHING SCIENTIFIC ADVENTURE OF OUR TIME* 218 (Touchstone Books 1991)).

161. Genetic information should be held to a higher standard than a social security number. *See The Privacy Act and the Freedom of Information Act*, SOCIAL SEC. ADMIN., <http://www.ssa.gov/privacyact.htm> (last visited Nov. 17, 2014).

162. *See Davis, supra* note 6, at 42.

163. *See supra* Part IV.B–C.

164. *See supra* Part IV.B.

165. *See supra* Part IV.B.

166. *See supra* Part IV.C.

167. *See supra* Part IV.B.

168. *See Bleicher, supra* note 31.

169. *See Recommended Uniform Screening Panel, supra* note 84.

170. *See Davis, supra* note 6, at 42–43.

171. Clayton, *supra* note 72, at 201.

consent from parents and questions whether nontreatable disorders should be included in the testing panel to begin with.¹⁷²

Requiring that consent only be obtained by clinicians is important in proper education of the parents.¹⁷³ Clinicians typically have close relationships with their patients and patients have a certain level of respect for their clinicians.¹⁷⁴ As a result, a better dialogue would occur if parents are informed of all aspects of the screening process and the clinician obtains consent.¹⁷⁵

However, just because a condition is nontreatable or will not show itself until the infant is an adult should not mean that it is appropriate to deny testing for those conditions.¹⁷⁶ Clayton argues that what would be more appropriate is to test for such conditions on a continuous basis as the child gets older.¹⁷⁷ Part of the purpose of informed consent is to educate parents and allow them to make an informed decision.¹⁷⁸ As such, some parents may want to test the child for these conditions all at once, and it should be their choice whether or not to do so.¹⁷⁹

As stated above, the foundation for good policy regarding newborn screening programs absolutely needs to start with parental informed consent.¹⁸⁰ Davis's suggestion that parents should have access to a lot of information during the prenatal period is a good one.¹⁸¹ However, it would not work well enough. Informed consent may solve the problem that a false positive presents,¹⁸² but it does not solve the problem of researchers and policy-makers wanting to continue to add conditions to be tested for through a screening program.¹⁸³ Therefore, in order for a new condition to be added to the panel, it would have to pass several tests, including, but not limited to, whether it is treatable, the severity of the effects of the disorder, the reliability of testing, and the ease of testing.

In addition, from a theoretical standpoint, the model guidelines that Schweers proposes are a good start toward adequate privacy

172. *Id.* at 201–02.

173. *See id.*

174. *See id.*

175. *See id.*

176. *See id.* at 203.

177. *See id.* at 202–03.

178. *See Davis, supra* note 6, at 45–46.

179. Clayton, *supra* note 72, at 203.

180. *See id.* at 202.

181. Davis, *supra* note 6, at 45–46.

182. *See id.*

183. *See id.* at 42.

protection.¹⁸⁴ Many of the problems addressed by Schweers would be solved if states uniformly implemented her model guidelines. A tiered approach which requires informed consent at every level is a clever and potentially effective way of assuring that parents are not bombarded with too much information all at once.¹⁸⁵ Each tier would consist of different tests and would require informed consent. The first level would consist of a panel of diseases which are uniformly tested by all programs.¹⁸⁶ The second level should consist of tests which are less reliable and do not have as great of a success rate after treatment. The third level should consist of nontreatable disorders and adult-onset disorders. Finally, the fourth level should consist of whole-genome sequencing and related testing.

Contrary to what Rothstein argues, it is possible to overcome the costs of incorporating whole-genome sequencing into newborn screening programs.¹⁸⁷ Rothstein argues that clinicians do not have the time or expertise to translate the genome sequence data and develop prevention and treatment plans for each patient.¹⁸⁸ However, while it is difficult for clinicians to do this now, as technology advances, it will get easier.¹⁸⁹

C. Fixing Issues Presented by HIPAA's Framework

HIPAA is a statute that sets out rules and regulations, which ultimately create an affirmative duty on the part of the healthcare provider not to disseminate or in any other way use an individual's personal health information without the explicit consent of the individual.¹⁹⁰ If an entity falls under the scope of HIPAA, "it may not disclose 'protected health information' without the patient's authorization"¹⁹¹ To protect health information "[such information] must be individually identifiable and maintained by a covered [entity]."¹⁹² The U.S. Department of Health and Human

184. See *supra* text accompanying notes 128–34.

185. See Schweers, *supra* note 6, at 906.

186. The panel proposed by the Health Resources and Services Administration might be a reasonable option. See *Recommended Uniform Screening Panel*, *supra* note 84.

187. See Pollack, *supra* note 33.

188. Rothstein, *supra* note 133, at 685.

189. See *DNA Sequencing*, *supra* note 34.

190. See *Health Information Privacy*, *supra* note 15.

191. Hsu, *supra* note 151, at 578.

192. *Does the HIPAA Privacy Rule Protect Genetic Information?*, U.S. DEP'T OF HEALTH & HUMAN SERVS.,

http://www.hhs.gov/ocr/privacy/hipaa/faq/protected_health_information/354.html (last updated March 14, 2006); 45 C.F.R. §§ 160.103, 164.501 (2013). This definition might not apply to un-identified samples. See *id.*

Services considers genetic information to be included in this definition.¹⁹³ However, HIPAA does not apply to all genetic screenings, because the “default guardian[s] of personal genetic information” are not “covered entities” under HIPAA.¹⁹⁴ Furthermore, HIPAA “does little to limit secondary uses of health information”¹⁹⁵

In addition to legislation that would create uniformity and require informed consent,¹⁹⁶ legislation should include requirements similar to HIPAA but tailored specifically toward an expanded definition of the term “genetic information” and which apply to a wider range of entities. First, in such legislation, the term “genetic information” should be defined to include any information about a genetic test, including analyses of “human DNA, RNA, chromosomes, proteins, or metabolites,” as long as the analysis “detects genotypes, mutations, or chromosomal changes.”¹⁹⁷ Further, “genetic information” should include any information relating to an individual’s genetic counseling or genetic education.¹⁹⁸ Finally, “genetic information” should include any method of obtaining the sample, such as via blood, skin, or hair.¹⁹⁹

Second, legislation would seek to encompass entities which are not included in HIPAA.²⁰⁰ The term “covered entities” would be defined as any entity that obtains the genetic information of an individual through a newborn screening program or voluntary genetic test.²⁰¹ These entities would consist of covered entities, as described in HIPAA, as well as corporations, research institutions, public and

193. *Health Information Privacy*, *supra* note 15. But it is not specified what “genetic information” includes. See 45 C.F.R § 160.103.

194. *Privacy & Ownership of an Individual’s Personal Genetic Information*, GENOMICS LAW REPORT (Oct. 29, 2009), <http://www.genomicslawreport.com/index.php/2009/10/29/privacy-ownership-of-an-individuals-personal-genetic-information/>. Corporations such as direct-to-consumer genetic testing companies are not considered “covered entities.” *Id.* “[HIPAA] only applies to the following three classes of covered entities involved in health claims: health care providers, health plans, and health clearinghouses.” Rothstein, *supra* note 133, at 686.

195. Rothstein, *supra* note 133, at 686.

196. See *supra* Part IV.B.

197. *FAQs on the Genetic Information Nondiscrimination Act*, U.S. DEP’T OF LABOR, <http://www.dol.gov/ebsa/faqs/faq-GINA.html> (last visited Nov. 17, 2014).

198. *Id.*

199. See *id.*

200. Rothstein, *supra* note 133, at 686.

201. See Ellen Wright Clayton, *Screening and Treatment of Newborns*, 29 HOUS. L. REV. 85, 144-45 (1992); *supra* Part I.

private universities, and testing laboratories.²⁰² Increasing the scope of covered entities would assure that an individual's privacy rights are not in jeopardy simply because an entity does not fit into the statute's definition.²⁰³ Overall, if the above definitions are incorporated into legislation, the main issues presented by HIPAA are eliminated and the burden on protecting the infant's privacy is shifted away from parents and onto the entities conducting the analyses.

VI. CONCLUSION

While there are many obvious positives to newborn genetic screenings, we must realize that some problems with current programs create privacy issues for the infant.²⁰⁴ Current and future genetic testing technology holds tremendous power which can affect the lives of parents and their children.²⁰⁵ In order to fully protect an infant's privacy rights relating to their genetic information, society must change from its old way of thinking about what genetic information is.²⁰⁶ To do this, this Comment proposes that not only should we require parents to be informed of newborn screening technologies, processes, and policies, but a statute should exist which is similar in effect to HIPAA that will create an affirmative duty of confidentiality on any entity or person who becomes associated with the infant's genetic information.²⁰⁷

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202. See 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, 9 (2011).

203. See Rothstein, *supra* note 135, at 686 (acknowledging the limited protection provided by HIPAA due to the limited scope of covered entities).

204. See *supra* Part IV.B.

205. See *supra* Part IV.

206. See *supra* Part V.A.

207. See *supra* Part V.