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# Newborn Screening Saves Lives: A Comparative Analysis Of New Jersey And New York State Law And Why Early-Onset Alzheimer's Disease Should Be Included On The Recommended Newborn Screening Panel

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**NEWBORN SCREENING SAVES LIVES: A COMPARATIVE ANALYSIS OF NEW JERSEY AND NEW YORK STATE LAW AND WHY EARLY-ONSET ALZHEIMER’S DISEASE SHOULD BE INCLUDED ON THE RECOMMENDED NEWBORN SCREENING PANEL**

By: Christina Velazquez

INTRODUCTION ..... 2

I. THE PROCESS AND HISTORY OF NEWBORN SCREENING ..... 4

    A. *The Expansion of Diseases Tested in Newborn Screening Programs* ..... 8

II. NEWBORN SCREENING IN THE PATIENT PROTECTION AND AFFORDABLE CARE ACT ..... 9

III. NEWBORN SCREENING IN NEW JERSEY AND NEW YORK ..... 13

    A. *Newborn Screening in New Jersey* ..... 13

    B. *New York’s Newborn Screening Law* ..... 20

IV. INCLUSION OF ALZHEIMER’S DISEASE TO NEWBORN SCREENING PANELS ..... 22

    A. *Overview of Early- Onset Alzheimer’s Disease* ..... 23

    B. *Therapy, Prevention and Intervention* ..... 25

    C. *Ethical Considerations and Policy Recommendations* ..... 27

    D. *Application of the Wilson and Jungner Test Supports Adding Early-onset Alzheimer’s Disease to Newborn Screening Panels* ..... 29

V. CONCLUSION ..... 32

## INTRODUCTION

The development of newborn screening is one of the most significant advancements in public health genetics. It is the practice of testing all babies for certain disorders, diseases and conditions that may hinder their normal development. In the United States alone, more than four million newborns are screened shortly after birth for certain genetic diseases, in addition for screening for non-genetic conditions.<sup>1</sup> While early detection and treatment can help prevent physical and intellectual disabilities, as well as life threatening illnesses, the early detection of some disorders that are required to be tested, cannot be cured, but rather, through therapies and intervention may be treatable or preventable.

This concept of ‘treatability’ is interesting considering that several diseases not required to be screened as recommended by the Department of Health and Senior Services (DHS) and state law, through intervention and therapies, are treatable. This perhaps should reveal the most obvious aspect of newborn screening in the United States- that the diseases, disorders and conditions screened vary from state to state.

The Supreme Court’s recent decision to uphold the Patient Protection and Affordable Care Act (ACA) supports the importance of recommending the nation-wide insurance coverage of diseases and disorders that are required to be tested through newborn screening. It also brings us closer to the institution of these federally recommended diseases in state newborn screening panels. Importantly, the law also stipulates that insurance companies must pay for the newborn screening of diseases recommended by DHS. This is a great achievement in the effort to assure

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<sup>1</sup> See NIH Program Explores the Use of Genomic Sequencing in Newborn Healthcare, <http://www.nih.gov/news/health/sep2013/nhgri-04.htm>, (last visited Dec. 8, 2013).

that all newborn babies are tested for these diseases and disorders that may save or improve the quality of their young lives.<sup>2</sup>.

Despite this laudable achievement in the law, it is important to note that not all of the disorders, conditions and diseases generally required to be tested under the federal panel can be cured. Rather, many of these conditions, can be treated or their onset delayed through the early detection of the presence of the gene. This presents several ethical considerations and questions as to why certain diseases in this non-treatable category are not tested through the newborn screening process.

In recent years, in particular, there has been a public push to add neurological diseases to newborn screening panels across the country. Such diseases include Alzheimer's disease and Krabbe disease, also referred to as globoid cell leukodystrophy. Alzheimer's disease is an irrevererable, progressive brain disease that slowly deteriorates memory and thinking abilities.<sup>3</sup> At its most debilitating stages, individuals with Alzheimer's disease must rely on others to complete the simplest of tasks.<sup>4</sup> Krabbe disease is an untreatable neurological disorder that leads to extreme muscle weakness and slowed mental and physical development.<sup>5</sup> It eventually leads to death.<sup>6</sup> New Jersey and New York are among the few states to test for Krabbe's disease.

This paper will the will present a comparative analysis as to the diseases tested under newborn screening under the ACA, New Jersey and New York state law. It will discuss the significance of the ACA to newborn screening in the United States. This paper will also focus on the inclusion of Krabbe's disease to New York and New Jersey's Newborn screening panels

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<sup>2</sup>See Health Care Reform Law Benefits Children with Genetic Diseases: Despite the law's benefits, many questions about coverage remain, <http://onlinelibrary.wiley.com/doi/10.1002/ajmg.a.35676/full>, (Last visited Dec. 8, 2013).

<sup>3</sup> See National Institute off Aging, Alzheimer's disease Fact; Sheet <http://www.nia.nih.gov/alzheimers/publication/alzheimers-disease-fact-sheet> (Last visited Dec. 8, 2013).

<sup>4</sup> See *id.*

<sup>5</sup> See Krabbe Disease, <http://ghr.nlm.nih.gov/condition/krabbe-disease>, (Last visited Dec. 8, 2013).

<sup>6</sup> See *id.*

and why its inclusion represents a milestone so that other neurological disorders, such as Early-onset Alzheimer's disease can be included. It will conclude with why Early-Onset Alzheimer's should be added to the federally recommended Uniform Screening Panel.

Part I of this paper will focus on the process and history of newborn screening in the United States. Part II of this paper will explore the changes made to newborn screening under the ACA and what is required of insurance companies under the Act. It will also break down the diseases approved by the Secretary of DHS to be included in the Recommended Newborn Screening Panel (RNSP) and what diseases are curable, treatable through intervention, and not treatable. Part III will explore the diseases required to be screened under New Jersey and New York's newborn screening laws. Part IV will explore what Early Alzheimer's disease is and will argue that it should be added to the list of diseases screened at the federal level. It will also discuss therapies and interventions that can reduce the risk of the disease and urge that the availability of these interventions supports its inclusion on the federal RNSP. This writing will conclude with Part V which will close with why Early-Onset Alzheimer's disease should be added to newborn screening panels, both in New York and New Jersey in addition to the federally recommended list of diseases screened for.

#### IV. THE PROCESS AND HISTORY OF NEWBORN SCREENING

Newborn screening is a medical test that is performed on infants shortly after birth that occurs by taking a sample of an infant's blood and analyzing it for abnormal genes types,

enzymes, metabolites and other chemicals.<sup>7</sup> This test was established to diagnosis and detect the presence of life-threatening disorders in infants.<sup>8</sup> The newborn screening procedure is a non-invasive blood test in which a small sample of blood is collected by simply taking a sample of blood from an infant's heel.<sup>9</sup> It is then analyzed for abnormal gene types, enzymes, metabolic and other chemicals that may lead to the diagnosis of a disorder.<sup>10</sup> This is usually performed in a laboratory and the results are provided to medical professionals usually within twenty-four (24) hours.<sup>11</sup>

Dr. Robert Guthrie developed the newborn screening test in the 1960s. Dr. Guthrie initially began his career as a principal cancer scientist that the Roswell Park Cancer Institute in Buffalo, New York.<sup>12</sup> After the birth of his second son John, who was born with phenylketonuria (PKU), he then altered his career and dedicated his research to altering and measuring phenylalanine levels in the blood of infants who acquired PKU in order to find a cure and treat his son.<sup>13</sup> Through his research, he developed the newborn screening test, a relatively easy procedure. Basically, within the first twenty-four hours of life, three to four droplets of an infant's blood are collected and placed on special filter paper.<sup>14</sup> The sample is then dried and sent out to a special laboratory for testing. The results are received usually within a day. The most important result was that through the Guthrie method of newborn screening, a definitive therapy for treating PKU was able to be administered to infants before the neurotoxicity that

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<sup>7</sup> See Association of Public Health Laboratories, What is newborn Screening, <http://www.aphl.org/aphlprograms/newborn-screening-and-genetics/50th-Anniversary-of-Newborn-Screening/Pages/What-is-Newborn-Screening.aspx>, (Last visited Dec. 8, 2013).

<sup>8</sup> See *id.*

<sup>9</sup> See *id.*

<sup>10</sup> See *id.*

<sup>11</sup> See *id.*

<sup>12</sup> See Jason Gonzalez & Monte S. Willis, *Robert Guthrie MD, PhD, Clinical Chemistry/Microbiology*, *LABMEDICINE*, 40, 748-749 (2009).

<sup>13</sup> See *id.*

<sup>14</sup> See *id.*

resulted from PKU became fatal.<sup>15</sup> Thus, it helped to save the lives of the infants affected by this debilitating disease.

In the United States, newborn screening began with the testing for genetic disease PKU. PKU is a genetic disorder the results in an individual's inability to produce the specific enzyme that causes a buildup of the amino acid phenylalanine.<sup>16</sup> An accumulation of the amino acid phenylalanine produces several physical and mental symptoms such as stunted growth, seizures, hyperactivity, sudden uncontrolled movements of the arms and legs, skin rashes and mental retardation.<sup>17</sup> In addition, phenylalanine affects the body's production of melanin, the pigment that determines the hair and skin color of an individual.<sup>18</sup> Therefore, infants with this condition often have lighter skin, hair and eyes than their brothers and sisters.<sup>19</sup> Fortunately, if PKU is diagnosed within the first few days of life, it is treatable and the majority of its symptoms can be avoided or reversed.<sup>20</sup> This is accomplished by constricting the amount of phenylalanine consumed in the infant's daily diet.<sup>21</sup> It also requires that a doctor or registered dietician closely monitor the individuals diet.<sup>22</sup> Infants, who are treated for PKU within the first few weeks of life, are able to lead normal, healthy lives.<sup>23</sup>

The original purpose of newborn screening had two main goals.<sup>24</sup> First, the programs were created to the monitor the health of the infant population in the United States.<sup>25</sup> Second,

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<sup>15</sup> *See id.*

<sup>16</sup> *See id.*

<sup>17</sup> *See id.*

<sup>18</sup> *See* A.D.A.M. Medical Encyclopedia, Phenylketonuria, <http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0002150/>, (Last visited Dec. 6, 2013)

<sup>19</sup> *See id.*

<sup>20</sup> *See id.*

<sup>21</sup> *See id.*

<sup>22</sup> *See id.*

<sup>23</sup> *See id.*

<sup>24</sup> *See* Rachel L. Schweers, Ph.D, *Newborn Screening Programs: How Do We Best Protect Privacy Rights While Ensuring Optimal Newborn Health?*, 61 DEPAUL L. REV. 869, 875 (2012).

the programs were intended to detect metabolic abnormalities in infants that were known, at the time, to have severe consequences such as death, that were easily discoverable through a simple blood test.<sup>26</sup>

With this scientific advancement, and in response to advocacy of geneticists, the medical community and parents, states began establishing newborn screening programs. The first state to mandate a newborn screening program was Massachusetts.<sup>27</sup> They started testing for PKU and added other diseases as they were discovered.<sup>28</sup> By the early 1970's all fifty states created programs with the support of the medical community.<sup>29</sup> They followed the Massachusetts lead, which resulted in approximately ninety percent of all newborns in the United States being tested for PKU.<sup>30</sup>

For the next ten years, states continued to add disorders to their newborn screening program. One important aspect of these new additions to newborn screening programs was that only disorders were included if treatment could avert serious harm to the affected child.<sup>31</sup> From then on, newborn screening has become a permanent part of infant health care in the United States. Today, all states, including the District of Columbia have implemented successful newborn screening programs to test for numerous metabolic and genetic disorders.<sup>32</sup> Although, it is important to note that not all of the diseases recommended by the RNSP are tested for in each state.

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<sup>25</sup> *See id.*

<sup>26</sup> *See id.*

<sup>27</sup> *See id.* Diane B. Paul, *The History of Newborn Phenylketonuria Screening in the U.S.*, in *Final Report of the Task Force on Genetic Testing*, Appendix 5 (Neil A. Holtzman & Michael S. Watson eds., 1997).

<sup>28</sup> *See id.*

<sup>29</sup> *See* Ellen Wright Clayton, *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 (2010).

<sup>30</sup> *See id.*

<sup>31</sup> *See id.*

<sup>32</sup> *See id.*

A. The Expansion of Diseases Tested in Newborn Screening Programs

As a result of the ever-increasing advancements in science through technology and genetics, states have continued to expand the disorders, diseases and conditions tested for in their newborn screening programs. Many of these diseases included not only diseases that benefit from intervention at infancy, such as PKU, but also for diseases for which there is no known treatment, cure or intervention available. Such diseases include Huntington's disease and some forms of Krabbe Disease.<sup>33</sup> In addition, diseases for which a prediction of diagnosis is given based upon an individual's genetics are also included.<sup>34</sup>

The accuracy and validity of predicting a diagnosis of a certain disease or disorder is, to date, one of the most controversial aspects of newborn screening programs. In fact, some medical professional organizations do not fully support predictive testing to reveal the propensity of diseases with no exiting treatment or for which the disease does not physically present itself until post-childhood.<sup>35</sup> However, despite this controversy, in recent years there has been a significant drive from advocacy, interest groups, and lobbyists to add diseases such as Krabbe's disease, also known as Globoid Cell Leukodystroph, to the list of conditions screened.<sup>36</sup> This is significant because the onset of Krabbe disease varies.<sup>37</sup>

In 1968, Maxwell Grove Wilson, the then Principal Medical Officer at the Ministry of Health in London, England, and Gunner Jungner, the then Chief of the Clinical Chemistry

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<sup>33</sup> See About Newborn Screening, Conditions Screened by State, <http://www.babysfirsttest.org/newborn-screening/states>, (last visited Dec. 8, 2013).

<sup>34</sup> See Beth A. Tarini et al., *Not Without My Permission: Parents' Willingness to Permit Use of Newborn Screening Samples for Research*, 13 PUB. HEALTH GENOMICS 125,127 (2010).

<sup>35</sup> See Beth A. Tarini et al., *Parents' Interest in Predictive Genetic Testing for Their Children when a Disease Has No Treatment*, 124 PEDIATRICS 432, 435 (2010).

<sup>36</sup> See Krabbe Newborn Screening, Information for Parents whose Baby Screened Positive for Krabbe Disease, [http://www.huntershope.org/site/PageServer?pagename=unbs\\_krabbe\\_newborn\\_screening](http://www.huntershope.org/site/PageServer?pagename=unbs_krabbe_newborn_screening), (Last visited Dec. 7, 2013).

<sup>37</sup> Please refer to Part 3, section A of this paper for a more detailed explanation on the onset of Krabbe disease.

Department of Sahlgren's Hospital in Gothenburg, Sweden, published a report that established a set of classic criteria, which still remain influential in defining what disorders and diseases should be deemed suitable for inclusion in newborn screening and genetic screening both on the state and federal level.<sup>38</sup> The report was entitled, *Principles and Practice of Screening for Disease*.<sup>39</sup>

Despite being established many years ago, these core principles remain relevant today.<sup>40</sup> The principles are generally as follows. First, the disease must be well defined and serious.<sup>41</sup> Second, there must be an accurate testing method available.<sup>42</sup> Third, the costs of the test must be reasonable. Fourth, there must be available treatment for the disorder.<sup>43</sup> And last, there must be adequate medical management facilities to refer infants for confirmatory diagnosis and treatment.<sup>44</sup>

With this framework in mind, the next part of this paper will explore the significance of the ACA followed by an analysis of newborn screening laws in New Jersey and New York.

## V. NEWBORN SCREENING IN THE PATIENT PROTECTION AND AFFORDABLE CARE ACT

The single most significant development in healthcare law was the Supreme Court's decision to uphold the Patient Protection and Affordable Care Act (ACA). In the landmark case

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<sup>38</sup> See Anne Andermann, Ingeborg Blancquaert, Sylvie Beauchamp & Véronique Déry, *Revisiting Wilson And Jungner In The Genomic Age: A Review Of Screening Criteria Over The Past 40 Years*, BULLWORLD HEALTH ORG, 2008 Apr;86(4):317-9.

<sup>39</sup> See *id.*

<sup>40</sup> See *id.*

<sup>41</sup> See Maxwell Grove Wilson & Gunner Jungner, *Principles and Practice of Screening for Disease*, 27-8, WORLD HEALTH, PUBLIC HEALTH PAPERS NO. 34 (1968).

<sup>42</sup> See *id.*

<sup>43</sup> See *id.*

<sup>44</sup> See Eileen M. McKenna, *The Mandatory Testing of Newborns for HIV: Too Much, Too Little, Too Late*, 13 N.Y.L. SCH. HUM. RTS. 307, 327-28 (1997).

*National Federation of Independent Business v. Sebelius*, several states filed suit against Secretary Sibelius based upon the constitutionality of the Patient Protection and Affordable Care Act signed into law by President Barak Obama on March 23, 2010.

The Court found that the Medicaid expansion mandate under the Affordable Care Act was unconstitutional, because states did not have adequate notice to voluntarily consent to the new Medicaid expansion provision. In addition, the Court held that if a state did not comply with the new law, the Secretary could withhold all of a state's existing federal Medicaid funds.<sup>45</sup> Despite this, the majority of the Supreme Court still found that this issue was appropriately remedied through the ACA. Thus, the ACA expansion was upheld.

As a result of this ruling by the Supreme Court, all provisions and mandates in the ACA remain effective. This signifies an important landmark for newborn screening. According to the ACA, insurance companies are now required to provide coverage for newborn screening of diseases that are federally recommended<sup>46</sup>.

Section 2713, Coverage for Preventable Health Services of the ACA states:

- (a) IN GENERAL- a group health plan and a health insurance issuer offering group or individual health insurance coverage *shall, at a minimum provide* coverage for and shall not impose any cost sharing requirements for-
  - (3) With respect to infants, children, adolescents, evidence-informed preventative care and *screenings provided for in the comprehensive guidelines supported by the Health Resources and Services Administration.*<sup>47</sup>

This provides that all the conditions added to the panel of diseases recommended to be screened by the Secretary's Discretionary Advisory Committee on Heritable Diseases in Newborns and Children must be covered by insurance, regardless of whether or not a state requires screening for that specific newborn disease. This is significant because not all states test for the diseases

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<sup>45</sup> *See id.*

<sup>46</sup> The Patient Protection and Affordable Care Act, 42 U.S.C. § 2713 (2010).

<sup>47</sup> *See id.*

recommended by the federal government. Therefore, mandating that insurance pay for all the diseases recommended by the federal government provides newborns with more thorough screening exams and can save more infant lives.

The Discretionary Advisory Committee on Heritable Disorders in Newborns and Children was established under the Public Health Service Act (PHS), 42 U.S.C. 217, Advisory Councils or Committees.<sup>48</sup> The Committee on Heritable Diseases in Newborns and Children recommends that every newborn screening panel in the country screen for the thirty-one core disorders and twenty-six secondary disorders recommended by the Uniform Screening Panel.<sup>49</sup> The Uniform Screening Panel defines primary disorders as those disorders for which medical evidence suggests that there is a medical benefit for detecting the disorder in infants.<sup>50</sup> The Panel defines secondary disorders as those disorders that can be detected while screening for the primary disorders.<sup>51</sup> These disorders include metabolic disorders<sup>52</sup>, Endocrine Disorders<sup>53</sup>, Hemoglobin Disorders<sup>54</sup> and other disorders such as Biotinidase deficiency, Critical Congenital Heart Disease, Cystic Fibrosis, Classic Galactosemia, Hearing loss and severe combined

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<sup>48</sup>See Discretionary Advisory Committee on Heritable Disorders In Newborns and Children, <http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/>, (Last visited Dec. 6, 2013).

<sup>49</sup> See *id.*

<sup>50</sup> See Virginia A. Moyer, Ned Calonge, Steven M. Teutsch, and Jeffrey R. Botkin, on behalf of the United States Preventive Services Task Force, *Expanding Newborn Screening: Process, Policy, and Priorities*, HASTINGS CENTER REPORT 38, no. 3, 32-39 (2008).

<sup>51</sup> See *id.*

<sup>52</sup> See Recommended Uniform Screening Panel Core Conditions, <http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendedpanel/uniformscreeningpanel.pdf> (as of April 2013). Metabolic Disorders occur when abnormal chemical reactions in your body disrupt the process your body to make energy from the food you consume. They include Organic Acid Conditions, Fatty Acid Oxidation Disorders and Amino Acid Disorders.

<sup>53</sup> See *id.* Endocrine disorders occur when your hormone levels are too high or too low. Hormone diseases also occur if your body does not respond to hormones the way it is supposed to.

<sup>54</sup> See *id.* Hemoglobin disorders is produced by genes that control the expression of the hemoglobin protein. Defects in these genes can produce abnormal hemoglobins and anemia

immunodeficiencies<sup>55</sup>. Currently, no state's newborn screening panel requires all thirty-one primary disorders to be screened. However, several states screen for all but one or two disorders. According to the Genetic Alliance, insurance companies have one year from when a condition is recommended by the Department of Health and Human Services to provide coverage for the newly tested conditions.<sup>56</sup> This signifies a remarkable victory for parents, because the new diseases added by the federal panel will be paid for through insurance because of the implementation of the ACA.

The Advisory Committee on Heritable Disorders in Newborns and Children also has the authority to review diseases, disorders and conditions that individuals seek to include on the Recommended Newborn Screening Panel (RNSP).<sup>57</sup> The RNSP is a comprehensive list approved and recommended by the Secretary of DHS for states to screen as part of their individual state newborn screening programs.<sup>58</sup> In order to include a condition on the RNSP, individuals and organizations must partake in an application process wherein they nominate the condition they desire to be included on the panel.<sup>59</sup> The application includes multiple letters in support of the condition being added, supporting data and scientific/clinical references to support its inclusion.<sup>60</sup> In addition, conflict of interest disclosures on behalf of the individuals and organizations must also be submitted.<sup>61</sup> Once the application is complete, it is then reviewed by the Advisory Committee's Nomination and Prioritization Workgroup who compiles a summary

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<sup>55</sup> *See id.*

<sup>56</sup> *See* Genetic Alliance Applauds Supreme Court Decision on Affordable Care Act, <http://www.geneticalliance.org/genetic-alliance-applauds-supreme-court-decision-affordable-care-act>, (Last visited Dec. 8, 2013).

<sup>57</sup> *See* Discretionary Advisory Committee on Heritable Disorders in Newborns and Children, Nominate a Condition, <http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/nominatecondition/index.html>, (last visited Dec. 8, 2013)

<sup>58</sup> *See id.*

<sup>59</sup> *See id.*

<sup>60</sup> *See id.*

<sup>61</sup> *See id.*

of the information submitted that is then provided to the Secretary of DHS who makes the final decision as to the disease's inclusion on the RNSP.<sup>62</sup>

## VI. NEWBORN SCREENING IN NEW JERSEY AND NEW YORK

As stated above, currently, the federal government recommends diseases, disorders and conditions to be screened through state Newborn Screening programs. As such, each state adopts its own newborn screening laws, develops its policies and procedures related to the newborn screening and operates their own individual newborn screening. Thus, the diseases, disorders and conditions screened for vary from state to state.

In New Jersey and New York, over forty diseases, conditions and disorders are screened for under their newborn screening laws.<sup>63</sup> This section will compare the diseases screened for by the newborn screening panels in New Jersey and New York.

### A. Newborn Screening in New Jersey

Currently, New Jersey has expanded its statewide system of screening to include a total of fifty-four (54) disorders, diseases and conditions, both primary and secondary as defined by the RNSP, which, if not detected early, can lead to severe health problems and interfere with the

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<sup>62</sup> See id.

<sup>63</sup> See What is Newborn Screening, <http://www.state.nj.us/health/fhs/nbs/faq.shtml> (Last visited Dec. 8 2013); Wadsworth Center, New York State Department of Health <http://www.wadsworth.org/newborn/> (Last visited Dec. 8, 2013).

physical and mental development of the newborn.<sup>64</sup> Together, these fifty-four diseases and disorders can be characterized into eleven disease groups for further clarification.

The first group of disorders tested under the New Jersey law are Biotinidase Deficiency disorders. Biotinidase deficiency results from defective activity of an enzyme that links biotin to other essential substances in the body.<sup>65</sup> A failure to diagnose and treat the deficiency causes permanent neurological damage and mental retardation in an infant. However, early diagnosis and treatment with pharmacological doses of biotin produce “marked improvement.”<sup>66</sup> In fact, pre-symptomatic treatment can result in the normal development of the infant.<sup>67</sup> It will also enable the infant to lead a healthy life. Testing for these deficiencies are also recommended by the Secretary of DHS.<sup>68</sup>

The second group of diseases are characterized as Congenital Adrenal Hyperplasia (CAH). CAH is a family of disorders whose common feature is an enzymatic defect in the steroidogenic pathway leading to the biosynthesis of cortisol.<sup>69</sup> The 21-hydroxylase enzyme deficiency accounts for 90-95% of CAH cases, resulting in ambiguous genitalia in females and salt-wasting crises in either males or females.<sup>70</sup> When infants suffer from Congenital Adrenal Hyperplasia, their bodies cannot retain salt, which is often referred to as salt-wasting.<sup>71</sup> Treatment for this disorder consists of hormone medications, along with monitoring and lifelong

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<sup>64</sup> See Disorders Screened in New Jersey, <http://www.nj.gov/health/fhs/nbs/disorders.shtml>, (Last visited Dec. 8, 2013).

<sup>65</sup> See Biontinidase Deficiency, <http://www.state.nj.us/health/fhs/nbs/biotinidase.shtml>, (Last visited Dec. 8, 2013).

<sup>66</sup> See Sanford J. Fox & Diony Young, *International Protection Of Children's Right To Health: The Medical Screening Of Newborns*, B.C. THIRD WORLD L. J.1, 9 (1991) (discussing biotinidase deficiency).

<sup>67</sup> See *id.*

<sup>68</sup> See *id.*

<sup>69</sup> See Congenital Adrenal Hyperplasia, <http://www.state.nj.us/health/fhs/nbs/cah.shtml>, (Last visited Dec. 8, 2013).

<sup>70</sup> See *id.*

<sup>71</sup> See *id.*

treatments.<sup>72</sup> Detecting this disorder through newborn screening allows an infant to have a normal life without intellectual and physical disabilities.<sup>73</sup>

Another group of diseases that are tested for include Congenital Hypothyroidism. Infants that are born with congenital hypothyroidism are unable to produce adequate amounts of the thyroid hormone called thyroxine.<sup>74</sup> This hormone is integral for the normal function and development of all of the body's organs and is essential for the normal development of the brain.<sup>75</sup> Without treatment, this disorder leads to cognitive and intellectual developmental delays.<sup>76</sup> It can also be acquired later on life.<sup>77</sup> Treatment for Congenital Hypothyroidism includes thyroid hormone therapy.<sup>78</sup> In addition, infants cannot be fed soy, fiber and iron.<sup>79</sup>

Under the New Jersey law, cystic fibrosis is another disease that is required to be tested. According to the NJ State Department of Health, it is the most common recessive genetic disorder in Caucasians in the U.S., Cystic Fibrosis occurs as a result of a defective Cystic Fibrosis Transmembrane regulator that results in thick mucus membranes, chronic obstructive lung disease and recurring pulmonary infections that can lead to death.<sup>80</sup> Cystic Fibrosis can be treated through nutritional care, such as a high calorie diet.<sup>81</sup> There is no known cure for cystic fibrosis and antibiotics and medical aggressive airway clearance techniques are often part of the treatment of this disease.<sup>82</sup>

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<sup>72</sup> *See id.*

<sup>73</sup> *See id.*

<sup>74</sup> *See* Congenital Hypothyroidism, <http://www.state.nj.us/health/fhs/nbs/congenital.shtml>, (Last visited Dec. 8, 2013).

<sup>75</sup> *See id.*

<sup>76</sup> *See id.*

<sup>77</sup> *See id.*

<sup>78</sup> *See* Liz Smith, *Updated AAP Guidelines on Newborn Screening and Therapy for Congenital Hypothyroidism*, 1 AM FAM PHYSICIAN, 439, 441 (2007).

<sup>79</sup> *See id.*

<sup>80</sup> *See* Cystic Fibrosis, <http://www.state.nj.us/health/fhs/nbs/fibrosis.shtml>, (Last visited Dec. 8, 2013).

<sup>81</sup> *See id.*

<sup>82</sup> *See id.*

Fatty acid oxidation disorders are also tested under the Newborn Screen Program in New Jersey.<sup>83</sup> Fatty acid oxidation disorders are inherited metabolic conditions that impair fatty acid metabolism. Each fatty acid oxidation disorder is associated with a specific enzyme shortcoming.<sup>84</sup> The most serious problem associated with fatty acids is that without them, the body can run out of energy and will often cease to function.<sup>85</sup> Without treatment, fatty acids that cannot be broken down will result in damage to an infant's heart and liver.<sup>86</sup> Through proper diet, exercise and monitoring, fatty acid oxidation disorders can be controlled, although not cured.<sup>87</sup>

Galactosemia disorders are another condition that is screened for under the New Jersey newborn screening program. It is an autosomal recessive disorder of carbohydrate metabolism.<sup>88</sup> This deficiency results in the build up of galactose in the body, which causes physical symptoms and discomfort following lactose ingestion. If an infant is found to have this condition, they are immediately placed on a soy diet and cannot be breastfed.<sup>89</sup> If Galactosemia disorders are untreated or not discovered, they can result in the death of the infant.<sup>90</sup>

Genetic disorders characterized as Sickle Cell Diseases are another group of disorders that are tested through the newborn screening program in New Jersey. This disorder affects hemoglobin, which is the molecule in red blood cells used to deliver oxygen throughout the

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<sup>83</sup> See Fatty Acid Oxidation Disorders, <http://www.state.nj.us/health/fhs/nbs/fatty.shtml>, (Last visited Dec. 8, 2013).

<sup>84</sup> See *id.*

<sup>85</sup> See Fatty Acid Oxidation Disorders, [http://mydoctor.kaiserpermanente.org/ncal/specialty/genetics/resources/conditions/fatty\\_acid\\_oxidation\\_disorders.jsp](http://mydoctor.kaiserpermanente.org/ncal/specialty/genetics/resources/conditions/fatty_acid_oxidation_disorders.jsp), (Last visited Dec. 8, 2013).

<sup>86</sup> See *id.*

<sup>87</sup> See *id.*

<sup>88</sup> See Galactosemia, <http://www.state.nj.us/health/fhs/nbs/galactosemia.shtml>, (Last visited Dec. 8, 2013).

<sup>89</sup> See *id.*

<sup>90</sup> See *id.*

body.<sup>91</sup> These diseases are tested in newborns; however, the classic signs and symptoms do not develop until later on in life.<sup>92</sup> In addition, bone marrow transplants often are the only cure for this disease. However, treatment can be administered through prophylactic antibiotics and appropriate immunizations to prevent infection.<sup>93</sup>

Another group of disorders screened for in New Jersey are Maple Syrup Urine Diseases. These are another autosomal recessive disorder.<sup>94</sup> They are associated with progressive neurological damage that can lead to death.<sup>95</sup> Treatment for these diseases must be continued throughout life and consist of a strict diet of amino acids.<sup>96</sup> Through newborn screening, early detection of this disease is possible. In fact, normal development and positive neurologic outcomes have been observed in infants who started treatment prior to developing this disease.<sup>97</sup>

Organic Acidemia Disorders are another group of conditions tested under New Jersey's newborn screening laws. Organic Acidemias disorders are inherited metabolic disorders that result in the accumulation of organic acids in blood and urine.<sup>98</sup> Treatment options for this disorder include low protein diets, carnitine and vitamin supplements.<sup>99</sup> Monitoring of the infants development is an important part of the treatment for those infants with Organic Acidemia Disorders.<sup>100</sup>

The most famous disease tested is Phenylketonuria (PKU). PKU was the first disorder to be tested through the administration of the newborn screening exam. It is a rare genetic

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<sup>91</sup> See Hemoglobinopathies Including Sickle Cell Disease, <http://www.state.nj.us/health/fhs/nbs/hemo.shtml>, (Last visited Dec. 8, 2013),

<sup>92</sup> See Sickle Cell Disease, <http://ghr.nlm.nih.gov/condition/sickle-cell-disease>, (Last visited, Dec. 8, 2013).

<sup>93</sup> See *id.*

<sup>94</sup> See Maple Syrup Urine Disease, <http://www.state.nj.us/health/fhs/nbs/maple.shtml>, (Last visited, Dec. 8, 2013).

<sup>95</sup> See *id.*

<sup>96</sup> See *id.*

<sup>97</sup> See *id.*

<sup>98</sup> See Organic Acidemia Disorders, <http://www.state.nj.us/health/fhs/nbs/acidemia.shtml>, (Last visited Dec. 8, 2013).

<sup>99</sup> See *id.*

<sup>100</sup> See *id.*

condition in which an infant is unable to breakdown the amino acid phenylalanine in the blood, which results in mental retardation and developmental delays.<sup>101</sup> This disorder is often inherited and occurs when the body cannot use the amino acid phenylalanine properly.<sup>102</sup> Treatment for PKU must begin as early as possible. It consists of selectively restricting dietary phenylalanine except for the precise amount needed for the infant's healthy growth and development.<sup>103</sup> This is preformed by providing infants with a special formula and as they grow and develop, placing them on a diet that is low in phenylalanine but high in other essential nutrients.<sup>104</sup> If treatment is successful, the infant will be able to have normal physical and mental development.<sup>105</sup>

The last group of disorders are called Urea Cycle Disorders. Urea cycle disorders are characterized by an accumulation of ammonia and its precursor amino acids that result in the defect in the urea cycle.<sup>106</sup> Clinical symptoms for this disorder usually appear when the infant is one to three days old.<sup>107</sup> When the disorder develops, the newborn undergoes rapid neurological deterioration.<sup>108</sup> As the ammonia levels in the child's body increases, the infant develops anorexia, hypothermia, irritability and seizures.<sup>109</sup> Without intervention, infants often become comatose and die.<sup>110</sup> Treatment for this disease includes providing the correct amount of protein in the infant's diet so that they can grow and develop.<sup>111</sup>

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<sup>101</sup> See Vani Kilakkathi, *Newborn Screening in America: Problems and Politics*, COUNCIL FOR RESPONSIBLE GENETICS, 2, 2-12 (2012) <http://www.councilforresponsiblegenetics.org/pageDocuments/WNMAKEPP1P.pdf>.

<sup>102</sup> See Phenylketonuria, <http://www.state.nj.us/health/fhs/nbs/pku.shtml>, (last visited Dec. 8, 2013).

<sup>103</sup> See *id.*

<sup>104</sup> See *id.*

<sup>105</sup> See *id.*

<sup>106</sup> See Urea Cycle Disorders, <http://www.state.nj.us/health/fhs/nbs/urea.shtml> (Last visited Dec. 8, 2013).

<sup>107</sup> See *id.*

<sup>108</sup> See *id.*

<sup>109</sup> See *id.*

<sup>110</sup> See *id.*

<sup>111</sup> See *id.*

Most recently, New Jersey became one of the few states to include Krabbe Leukodystrophy to the New Jersey newborn screening panel.<sup>112</sup> Krabbe disease is an autosomal recessive lysosomal storage disorder that affects the central nervous system.<sup>113</sup> The significance of testing for Krabbe disease in New Jersey represents a huge step towards newborn screening of neurological disorders.

In early 2013, New Jersey became only the fifth state to add Krabbe's Disease to their state newborn screening panel.<sup>114</sup> This represented a huge victory for advocates of this deadly neurological disorder as well as the infants born with this disorder. Similar to the other diseases screened for in New Jersey, Krabbe Disease is an autosomal recessive disorder that is associated with mutations in the galactosylceramidase gene (GALC).<sup>115</sup> When the GALC gene is mutated, it produces toxic substances in the brain, causing myelin loss, changes in the brain cells, and severe neurological damage.<sup>116</sup>

Unlike some of the diseases tested by newborn screening panels, Krabbe Disease has variable ages of onset and forms of progression. It is characterized into four types- early infantile, late infantile, juvenile and adult.<sup>117</sup> Children with early-infantile Krabbe disease show symptoms of developmental delays before six months of age.<sup>118</sup> It typically leads to death before

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<sup>112</sup> See Janet E. Deane, Stephen C. Graham, Nee Na Kim, Penelope E. Stein, Rosemund McNair, M. Begona Cahon-Gonzalez, Timothy M. Cox, & Randy J. Read, *Insights into Krabbe Disease from Structures of Galactocerebrosidase*, INSIGHTS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, September 13, 2011, at 59.

<sup>113</sup> See *id.*

<sup>114</sup> See New Jersey Becomes the Fifth State to Add Krabbe Disease to Newborn Screening Panel with Passage of Emma's Law, [http://www.huntershope.org/site/DocServer/Press\\_Release\\_New\\_Jersey\\_Becomes\\_5th\\_State\\_to\\_Add\\_Krabbe.pdf?docID=9301](http://www.huntershope.org/site/DocServer/Press_Release_New_Jersey_Becomes_5th_State_to_Add_Krabbe.pdf?docID=9301), (Last visited Dec. 8, 2013).

<sup>115</sup> See Suzuki Kunihiro, *Globoid cell leukodystrophy (Krabbe's disease): update*, JOURNAL OF CHILD NEUROLOGY, 595, 595-603 (2013).

<sup>116</sup> See *id.* at 596.

<sup>117</sup> See *id.*

<sup>118</sup> See *id.*

the age of two.<sup>119</sup> Late-infantile Krabbe disease is also deadly and has an onset between six and three months of age.<sup>120</sup> Children with juvenile Krabbe disease become symptomatic between three and eight years of age and may have a longer survival time.<sup>121</sup> Adult-onset Krabbe disease is even more inconstant. Affected individuals may have a normal life span.<sup>122</sup>

The addition of Krabbe disease to the newborn screening panel in New Jersey signifies a change in the philosophy that newborns should only be screened for treatable or preventable conditions. Presently, there is no known cure for Krabbe's Disease. For those infants and individuals who acquire this disease, treatment is limited to supportive care, palliative care and drug therapy to control irritability.<sup>123</sup> Currently, the only temporary curable treatments available are the transplantation of umbilical cord blood, hematopoietic stem cell transplantation and allogeneic bone marrow transplantation.<sup>124</sup> These treatments have resulted in less severe signs and symptoms and significant delays in the onset of symptoms.<sup>125</sup>

In general, newborn screening in New Jersey is paid for through insurance.<sup>126</sup>

In addition, initial newborn screening collection forms are purchased by hospitals at a cost of ninety dollars per form.<sup>127</sup> This cost is incorporated into labor and delivery charge.<sup>128</sup>

#### *B. New York's Newborn Screening Law*

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<sup>119</sup> *See id.*

<sup>120</sup> *See id.*

<sup>121</sup> *See id.*

<sup>122</sup> *See id.*

<sup>123</sup> *See id.*

<sup>124</sup> *See* Krabbe Disease, <http://ghr.nlm.nih.gov/condition/krabbe-disease>, (Last visited Dec. 8, 2013).

<sup>125</sup> *See id.*

<sup>126</sup> *See* Conditions Screened by State, <http://www.babysfirsttest.org/newborn-screening/states/new-jersey>, (Dec. 8, 2013).

<sup>127</sup> *See id.*

<sup>128</sup> *See id.*

Similar to the diseases screened by the Newborn Screening Panels in New Jersey, New York also screens for many of the same diseases, conditions and disorders. Such groups of diseases and disorders include Endocrine Disorders, which include Congenital Adrenal Hyperplasia and Congenital Hypothyroidism, various Hemoglobinopathies such as Sickle Cell disease, Amino Acid disorders such as PKU, Fatty Acid Oxidation Disorders, Organic Acid Disorders and Urea Cycle Disorders.<sup>129</sup> In addition, like New Jersey, New York screens for Biotindase Deficiency, Cystic Fibrosis, and Galactosemia.<sup>130</sup>

Unlike New Jersey, the major difference between the diseases screened by both states newborn screening panels is that New York requires the mandatory screening of the Human Immunodeficiency Virus (HIV). Newborn screening tests in New York examine the infant's blood for the presence of HIV antibodies.<sup>131</sup> If HIV antibodies are detected in the infant, the mother is also infected with the virus.<sup>132</sup> Under the newborn screening laws in New Jersey, HIV testing is not mandatory. However, since 1995, regulations in New Jersey have required all pregnant women to be offered HIV counseling and *voluntary* HIV tests.<sup>133</sup>

New York was the first state in the country to include Krabbe Disease to its screening program in August 2006.<sup>134</sup> The impetus for the inclusion of this test by the New York newborn screening panel came from the advocacy group, Hunter's Hope who is currently trying to lobby

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<sup>129</sup> See Disorders Identified, <http://www.wadsworth.org/newborn/babhealth.htm>, (Dec. 8, 2013).

<sup>130</sup> See *id.*

<sup>131</sup> See *id.*

<sup>132</sup> See *id.*

<sup>133</sup> See Sindy Paul & Charles E Denk, *Screening for HIV in Pregnant Women in New Jersey*, PREGNANCY RISK ASSESSMENT MONITORING SYSTEM, (2008).

[http://www.nj.gov/health/fhs/professional/documents/brief\\_hiv\\_screening.pdf](http://www.nj.gov/health/fhs/professional/documents/brief_hiv_screening.pdf).

<sup>134</sup> See Roberta Salveson, *Expansion of the New York State Newborn Screening Panel and Krabbe Disease: A Systematic Program Evaluation*, (2011), (unpublished thesis, Columbia University) (on file with Columbia University library system.)

all states for the inclusion of this disease in their newborn screening programs.<sup>135</sup> They argued that Krabbe disease should be added to New York's newborn screening panel because of the possibility of preventing the disease's development through chord blood transplants.<sup>136</sup> Newborn screening for Krabbe disease could allow doctors to pre-symptomatically identify if an infant is susceptible to develop the disease, and treat it.<sup>137</sup> Soon after New York began testing for this disease, other states, such as New Jersey followed.

## VII. INCLUSION OF ALZHEIMER'S DISEASE TO NEWBORN SCREENING PANELS

There is a valid argument for including Early-onset Alzheimer's disease on the list of diseases tested on newborn screening panels in New Jersey, New York and most importantly on Federally recommended list. Through scientific advancements and technology, it is easy to predict if one is a carrier of this degenerative, neurological disorder. In addition, since this type of Alzheimer's is the most rare form, individuals susceptible to this disorder should have the right to know that they may acquire it.

Furthermore, unlike Krabbe's disease, which is already included in some newborn screening panels, a neurological disorder for which there is no cure, science has demonstrated that through intervention and therapy, the risk of developing Early-onset Alzheimer's disease may be reduced by living a particular type of lifestyle<sup>138</sup>. This is something that can be taught to children who have an increased risk of developing this disease. Therefore, if states such as New Jersey and New York are at the forefront in beginning to screen newborns for neurological

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<sup>135</sup> See Hunters Hope Foundation, [www.huntershope.org](http://www.huntershope.org), (Last visited Dec. 8, 2013).

<sup>136</sup> See *id.*

<sup>137</sup> See *id.*

<sup>138</sup> See David S. Geldmacher, *Alzheimer disease prevention: Focus on cardiovascular risk, not amyloid?* 10 CLEVE. CLINIC J. OF MED., 689, 689-690(2010)(discussing lifestyle choices).

disorders that are deadly, they should consider screening for neurological disorders whose onsets can be delayed.

A. Overview of Early- Alzheimer's Disease

Alzheimer's disease is a degenerative disease of the brain that results in dementia.<sup>139</sup> According to the National Institute of Dementia and Stroke, dementia occurs when brain cells die.<sup>140</sup> Brain cell death is not an immediate occurrence.<sup>141</sup> Rather, it is progressive and occurs over time.<sup>142</sup> Symptoms of dementia include the gradual loss of memory, judgment and the ability to process information so that an individual may function.<sup>143</sup>

There are two types of Alzheimer's disease: early onset or familial and late-onset. Early on-set Alzheimer's disease occurs in those individuals who acquire the disease at age sixty-five (65) or under.<sup>144</sup> This article solely focuses on the genetic testing of children for Early-onset Alzheimer's disease as it is the type of Alzheimer's that is the rarest and most easily detected through genetic testing.

The genetic origin of Early-onset Alzheimer's disease is easily understood. Most cases of Early-onset Alzheimer disease are caused by gene mutations that can be passed from parent to child.<sup>145</sup> The Early-onset familial form of the disease can arise in individuals who are in their

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<sup>139</sup> See Edmund G. Howe, *Ethical Issues in Diagnosing and Treating Alzheimer's Disease*, *Genetics and the Moral Future of Dementia Care*, 3 PSYCHIATRY (EDGMONT), 44, 43–53 (2006)(discussing dementia).

<sup>140</sup> See *Id.*

<sup>141</sup> See *Id.*

<sup>142</sup> See *Id.*

<sup>143</sup> See Early-Onset Alzheimer's Disease, <http://www.medicalnewstoday.com/articles/142214.php>, (Last updated Oct. 31, 2013)

<sup>144</sup> See *Id.*

<sup>145</sup> See *Id.*

forties and fifties, and in very rare cases can develop in people who are thirty years of age.<sup>146</sup> However, it is generally characterized as occurring in individuals age sixty-five (65) and under.<sup>147</sup> With this familial form of Early-onset Alzheimer's, there usually will be many parents, siblings and cousins who acquire the disease and also have dementia.<sup>148</sup> When an individual is a carrier of one of the gene mutations that cause Early-onset Alzheimer's disease to develop, it is a good predictor that they are susceptible to develop this debilitating disease.<sup>149</sup>

As the progression of the disease worsens, people with Early-onset Alzheimer's disease experience personality and behavioral changes that make social integrations difficult for them.<sup>150</sup> As a result, individuals suffering from this disease experience frequent agitation, restlessness, withdrawal and even the loss of language skills.<sup>151</sup>

The genes associated with Alzheimer's disease are amyloid precursor protein (APP), Presenilin (PSEN-1 and PSEN-2) and apolipoprotein (APOE).<sup>152</sup> Mutations in APP result in an incorrect amount of the protein, which produces a version of amyloid  $\beta$  that is more likely to form plaques. Mutations in APP account for ten to fifteen percent of familial Early-onset Alzheimer's disease cases.<sup>153</sup> The PSEN genes encode proteins that function in the cleavage of Amyloid Precursor Protein.<sup>154</sup> Mutations in both PSEN1 and PSEN2 result in incorrect cleavage of APP, and are associated with the development of familial Early-onset Alzheimer's disease.<sup>155</sup> Mutations in PSEN1 are thought to account for 30%-70% of familial Early-onset Alzheimer

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<sup>146</sup> See Id.

<sup>147</sup> See Id.

<sup>148</sup> See Id.

<sup>149</sup> See Mark A. Rothstein, *Predictive Genetic Testing For Alzheimer's disease In Long-Term Care Insurance*, 5 GA. L. REV., 709, 709-711 (2001).

<sup>150</sup> See Early Detection Matters, [http://www.alz.org/national/documents/checklist\\_10signs.pdf](http://www.alz.org/national/documents/checklist_10signs.pdf), (Last visited Dec. 8, 2013).

<sup>151</sup> See id.

<sup>152</sup> See id.

<sup>153</sup> See ROTHSTEIN, *supra* note 147, at 720.

<sup>154</sup> See id.

<sup>155</sup> See id.

disease developments, while mutations in PSEN2 are thought to account for less than 5%.<sup>156</sup>

Children of an affected parent have a 50% chance of inheriting the mutation and developing Early-onset Alzheimer's Disease. It is important to note that mutations in APP, PSEN1 and PSEN2 do not account for all of the genes that result in familial early-onset Alzheimer's disease.<sup>157</sup> In fact, scientists believe that there are other genes that attribute to this disease that are not known at this time and likely to be discovered in the future.<sup>158</sup> However, mutations in these genes will likely lead to the development of Early-onset Alzheimer's disease.

### *B. Therapy, Prevention and Intervention*

In addition to genetics in predicting the possibility of acquiring Early-onset Alzheimer's Disease, environmental factors, lifestyle choices, among many other factors can also attribute to the development of the disease.<sup>159</sup> Thus, with this known, it is important to know whether an infant is a carrier of the genes in order to delay the progression and onset of the Early-onset Alzheimer's disease occurring later on in life. Therefore, the testing for the genes associated with Early-onset Alzheimer's disease is important to include in newborn screening programs so that the onset of the disease is delayed and individuals can have a longer and healthier quality of life.

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<sup>156</sup> See id.

<sup>157</sup> See id.

<sup>158</sup> See id.

<sup>159</sup> See The Search for Alzheimer Prevention Strategies, <http://www.nia.nih.gov/alzheimers/publication/preventing-alzheimers-disease/search-alzheimers-prevention-strategies> (Last updated, Sep.12, 2012.)

Over the last three decades, researchers have found that parents who warn their children about the risk of developing Alzheimer’s Disease has lead to its delayed development.<sup>160</sup> Knowledge is power. Knowing this from a very young age, leads to, what scientists refers to as “building your cognitive reserve.” According to this theory, by exercising and developing the mind as early as possible, later on in life, when Early-onset Alzheimer’s Disease begins to deteriorate the brain, people with the largest and most comprehensive cognitive reserves would experience less brain deterioration.<sup>161</sup> Cognitive reserve has also been used to explain the neuropathologic changes associated with coping with Alzheimer’s disease.<sup>162</sup> According to the American Academy of Neurology, cognitive reserve capacity is set early on in life and gradually changes as the nervous system changes.<sup>163</sup> As individuals age, those with a higher reserve capacity will have a lower risk of developing dementia.<sup>164</sup> This is significant, because, since Early-onset Alzheimer’s disease is very predicable through genetic testing, families should have the right to know that their children are carriers of genes so that they can start cognitive therapy early on in life.

Another treatment recommended for the prevention of Alzheimer’s is physical, cardiovascular exercise.<sup>165</sup> The scientific and medical communities have established that evidence suggests that cholesterol plays a role in the development of Alzheimer disease in general.<sup>166</sup> Therefore, individuals who build in regular cardiovascular exercise into their daily

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<sup>160</sup> See Adrienne M. Tcuker & Yaakov Stern, *Cognitive Reserve in Aging*, Columbia University, 76 (2011) <http://www.cumc.columbia.edu/dept/sergievsky/pdfs/cognitivereserveinaging.pdf>.

<sup>161</sup> See *id.*

<sup>162</sup> See *id.*

<sup>163</sup> See *id.*

<sup>164</sup> See *id.*

<sup>165</sup> See *id.*

<sup>166</sup> See Benjamin Wolozin, MD, PhD; Wendy Kellman; Paul Ruosseau, MD; Gastone G. Celesia, MD & George Siegel, *Decreased Prevalence of Alzheimer Disease Associated With 3-Hydroxy-3-Methylglutaryl Coenzyme A Reductase Inhibitors*, 57 ARCH NEUR., 1439, 1443 (2000) (discussing cholesterol’s role in Alzheimer’s disease).

routine are less likely to develop Early-onset Alzheimer's disease.<sup>167</sup> In fact, individuals who exercised regularly have about half the risk of developing dementia than those who do not exercise. Therefore, it is important for children to be brought up knowing the importance of exercising and engaging in actual exercise so that those, who are carriers of the genes associated with Early-onset Alzheimer's disease, can reduce their risk of acquiring the disease later on in life.<sup>168</sup>

Finally, a change in diet may reduce the risk of developing Early-onset Alzheimer's Disease.<sup>169</sup> According to the Physician's Committee for Responsible Medicine, there are seven dietary principles that are proven to reduce the risk of Alzheimer's disease.<sup>170</sup> These principles were revealed during the International Conference on Nutrition and the Brain.<sup>171</sup> They include reducing your intake of saturated and trans fats and cooperating vegetables, fruits and whole grains into your diet.<sup>172</sup> Those individuals who are susceptible to developing Early-onset Alzheimer's disease should incorporate Vitamin E and B12 into their daily vitamins. When selecting multiple vitamins, those at risk for Early-onset Alzheimer's disease should avoid iron and copper.<sup>173</sup> Providing this information to parents enables them to educate their children and, through physician supervision, integrate these vitamins into their children's daily diet.

### *C. Ethical Considerations and Policy Recommendations*

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<sup>167</sup> *See id.*

<sup>168</sup> *See id.*

<sup>169</sup> *See id.*

<sup>170</sup> *See id.*

<sup>171</sup> *See id.*

<sup>172</sup> *See id.*

<sup>173</sup> *See id.*

While the technology is available for Early-onset Alzheimer's disease to be screened in newborns, there are still several ethical questions that arise, especially since even the earliest indications of this disease occur once a child has matured into an adult. In particular, it is important for parents to proceed with caution in acquiring this information and utilizing it in their child's daily life.

The genetic testing for Early-onset Alzheimer's disease in newborns can present some ethical problems in regard to the child's autonomy. This discussion ultimately focuses on whose right to autonomy is jeopardized; the parents or the child, when there is no immediate identified medical benefit. If parents chose to have their child tested to their susceptibility for obtaining Early-onset Alzheimer's disease, the child's right to autonomy is violated. However, with the technology available to predict this disease in infants, there is an argument to be made that denying this test to parents deprives their children of health care that could provide them with a better quality of life.

In addition, the physiological effects of telling a child that they are susceptible to developing Early-onset Alzheimer's disease still needs to be determined. Children are vulnerable and basically telling a child that they will one day lose their ability to remember memories and information can have detrimental effects on the child's development. The scientific and medical communities are yet to determine the implications that result from when a child learns this information. In addition, there still needs to be more information available as to what the appropriate age is that a child should learn that they are likely to develop Early-onset Alzheimer's disease in the future.

Furthermore, if infants and children are treated for their susceptibility in developing Early-onset Alzheimer's disease, there needs to be strict physician guidance in overseeing these

treatments. Also, guidelines need to be implemented as to what types of treatments can be used in children and the safety of these treatments. This guidance should be established on the federal level so that a least, minimum standards are developed through state implementation.

Infants with negative or unclear newborn screening test results also need have the newborn screening test re-administered to them. This re-administration should be performed at no additional cost to parents. Rather, should either be paid for through insurance or by the state.

As to the unification of newborn screening panels nationwide, there needs to be an incentive for states to adopt all the diseases recommended by the Uniform Screening Panel. By doing so, this will ensure that all newborns are screened for diseases that can save their lives. This should not be viewed as a mandate or a political agenda. Rather, it should be considered a public health initiative. The Secretary of DHS should evaluate the current list of diseases screened, together with all the other diseases that states screen for to make “master” list of diseases that should be screened. This process would appear to be faster than the current process of medical professionals and individuals submitting an application to the Department for review. Together, it is important that these ethical considerations and policy recommendations are considered when adding Early-onset Alzheimer’s disease to newborn screening panels.

*D. Application of the Wilson and Jungner Test Supports Adding Early-onset Alzheimer’s Disease to Newborn Screening Panels*

In addition to these treatments proven to delay the onset of Early-onset Alzheimer’s disease, it also passes the Wilson and Jungner classic screening criteria for inclusion on newborn screening panels discussed in Part I, Section A.

According to Wilson and Jungner, there are five criteria that must be met for a genetic disorder to be screened.<sup>174</sup> According to them, in order for a disease to be screened, it must be well defined and serious.<sup>175</sup> Early-onset Alzheimer's disease is a defined neurological disorder that results in clinical symptoms resulting in the progression of cognitive and behavioral impairment.<sup>176</sup> Therefore, the disease is defined as serious as required under the Wilson and Jungner criteria.

Second, Wilson and Jungner state that there must be an accurate testing method available for the genetic disease.<sup>177</sup> The Newborn Screening Quality Assurance Program (NSQAP) is a voluntary, non-regulatory program established by the federal government to aid state health departments and their laboratories in maintaining accurate newborn screening test results.<sup>178</sup> The NSQAP provides proficiency-testing services, which give state laboratories, coded specimens of the diseases screened to help scientists and doctors determine the results of the administered newborn screening test.<sup>179</sup> Therefore, there is accurate testing available for Early-onset Alzheimer's disease.

Third, the costs of the test must be reasonable. According to the New Jersey State Department of Health, the cost of newborn screening is covered through insurance so it is reasonable.<sup>180</sup> In New York, the cost of newborn screening is absorbed by the state. Therefore,

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<sup>174</sup> See ANDERMANN, BLANCQUAERT, BEAUCHAMP & DÉRY, *supra* note 37, at 720.

<sup>175</sup> See *id.*

<sup>176</sup> See Reisa A. Sperling, Paul S. Aisen, Laurel A. Beckett, David A. Bennett, Suzanne Craft, Anne M. Fagan, Takeshi Iwatsubo, Clifford R. Jack Jr., Jeffrey Kaye, Thomas J. Montine, Denise C. Park, Eric M. Reiman, Christopher C. Rowe, Eric Siemers, Yaakov Stern, Kristine Yaffe, Maria C. Carrillo, Bill Thies, Marcelle Morrison-Bogorad, Molly V. Wagster, Creighton H. Phelps, *Toward defining the preclinical states of Alzheimer's disease: Recommendations from the National Institute of Aging-Alzheimer's Association workgroup on diagnostic guidelines for Alzheimer's disease, Alzheimer's and Dementia*, 7 THE JOURNAL OF THE ALZHEIMER'S ASSOCIATION, 280, 282-85, (2011) (discussing definition of Alzheimer's disease)

<sup>177</sup> See *id.*

<sup>178</sup> See Newborn Screening Quality Assurance Program, <http://www.cdc.gov/labstandards/nsqap.html> (Last visited Dec. 8, 2013).

<sup>179</sup> See *id.*

<sup>180</sup> See How much does it cost, <http://nj.gov/health/fhs/nbs/faq.shtml>, (last visited Dec. 8, 2013).

the cost of newborn screening to detect Early-onset Alzheimer's disease is reasonable as there are already numerous diseases that states and insurance companies pay for that are tested.

The fourth criteria established by Wilson and Jungner is that there must be available treatment for the disorder.<sup>181</sup> Recent medical studies have indicated that there is treatment available to help delay the onset of Alzheimer's disease in general.<sup>182</sup> Such therapies and treatments include exercising and developing the mind as early as possible. Therefore, this criterion is also satisfied.

The last Wilson and Jungner criterion is that there must be adequate medical management facilities to refer infants for confirmatory diagnosis and treatment.<sup>183</sup> This is satisfied through the sophistication of hospitals and medical facilities. In addition, health care systems across the county are developing specific treatment centers for Alzheimer's disease treatment. Most recently, Capitol Health's Institute for Neuroscience announced that it will be opening a specialized program focusing on the treatment of Alzheimer's disease in New Jersey.<sup>184</sup>

Based upon the above analysis, it is evident that Early-onset Alzheimer's disease passes renowned criteria set forth by Wilson and Jungner needed for diseases to be included in newborn screening panels. If we were to apply this same test to Krabbe's disease, in which there is no cost-effective treatment for and no known cure, it appears that it would not pass the Wilson and Jungner test. Despite this, New Jersey and New York have already added this disorder to their state newborn screening panel. Therefore, this provides a strong argument for the inclusion of

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<sup>181</sup> See MAXWELL GROVE WILSON & GUNNER JUNGNER, *supra* note 40, at 27.

<sup>182</sup> See Preventing Alzheimer's Disease, What We Know, <http://www.nia.nih.gov/alzheimers/publication/preventing-alzheimers-disease/search-alzheimers-prevention-strategies> (last updated, Sep.12, 2012).

<sup>183</sup> See EILEEN M. MCKENNA, *supra* note 43, at 307.

<sup>184</sup> See Susan K. Livio, *Capitol Health Opens Alzheimer's Treatment Center*, STAR LEDGER, October 23, 2013.

Early-onset Alzheimer's disease to newborn screening panels at least on the federal Uniform Newborn Screening Panel.

## V. CONCLUSION

This paper discussed the importance of states adopting all of the diseases recommended by the RNSP to their individual newborn screening programs. If this is not accomplished, there is a possibility that some infants will die as a result of their state not choosing to screen for a particular disease recommended by the RNSP. Evidence of this has already occurred from some states not choosing to screen for Krabbe disease as discussed above. If the testing and treatment is available to save the lives of infants, not utilizing it is a foolish.

In addition, this paper also discussed the importance of including Early-onset Alzheimer's disease to the list of federally recommended screened diseases, conditions and disorders approved by the Secretary of DHS. Based upon the information known about Early-onset Alzheimer's disease and the medical research available that confirms possible treatments and interventions for the disease, it is clear that this disease should be added to the federal list of diseases that are recommended to be screened for in newborns. In addition, Early-onset Alzheimer's disease passes the criteria for screening diseases established by Wilson and Jungner. As evidenced in this paper, Krabbe disease, which is already screened for by some states does not pass this well-established criteria. It is important to note that there are treatments and interventions available to delay the onset of Early-onset Alzheimer's disease. Therefore, this presents a strong argument that favors adding Early-onset Alzheimer's disease to the federal panel.

Furthermore, with the ACA requirement that all diseases recommended by the federal panel be paid for through insurance, more infant lives will be saved, regardless of whether or not their state screens for the disease. This is a significant advancement in newborn screening and demonstrates the importance of screening infants for all the diseases federally recommended. Therefore, Early-onset Alzheimer's disease should be added to the Recommended Newborn Screening Panel. If this cannot be accomplished, Early-onset Alzheimer's should be included as a disease tested in New York and New Jersey's newborn screening panels.

With the ability to test if an infant is a carrier of the genes associated with Early-onset Alzheimer's disease, medical research suggests that through therapies and interventions, its onset can be delayed. Therefore, the testing for these genes is important so that those individuals who are susceptible for acquiring Early-onset Alzheimer's disease can have a healthier and better quality of life and slow the progression of this devastating illness.