Citizens' Council on Health Care

CCHC Report
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A Critical Analysis of the Implications for Genetic Privacy and Consent Rights in Congress' Proposed 'Newborn Screening Saves Lives Act of 2007'

S.1858 & H.R. 3825

Proposed federal legislation to establish a national system for government genetic testing, surveillance and research on citizens using blood taken at birth

by Twila Brase

Genetic privacy, parent consent and individual self-determination rights are at stake with the U.S. Senate's recent passage of S. 1858, the 'Newborn Screening Saves Lives Act of 2007.' This paper outlines five key issues of concern.

Overview

If enacted, S.1858 will nationalize the genetic testing of all newborn children—and indirectly their families. The bill and its U.S. House companion bill, H.R. 3825, will:

- Establish a national list of genetic conditions for which newborns and children are to be tested.
- Establish protocols for the linking and sharing of genetic test results nationwide.
- Build surveillance systems for tracking the health status and health outcomes of individuals diagnosed at birth with a genetic defect or trait.
- Use the newborn screening program as an opportunity for government agencies to identify, list, and study "secondary conditions" of individuals and their families.
- Subject citizens to genetic research without their knowledge or consent.

Newborn screening is "the first program of populationwide genetic testing."[1] Yet this legislation does not require informed parent consent for testing, surveillance, or research.

Babies are newborn citizens. Each one of the 4,000,000 children born each year in the United States has all the constitutional rights of fully-grown citizens. Eventually, these children become voting adults.

The U.S. Senate passed S. 1858 on December 13, 2007. An anonymous—said to be unanimous—voice vote was taken. The Senate bill awaits action in the U.S. House, along with the House bill, H.R. 3825. (CCHC Appendix A and B) *This analysis examines H.R. 3825 IH (as Introduced in the House), the House version of the Senate bill.*

[1] "Newborn Screening: Complexities in Universal Genetic Testing" Nancy S. Green M.D. [March of Dimes] et al. American Journal of Public Health. (96) No. 11, Nov. 2006.

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FIVE KEY CONCERNS

A careful examination of H.R 3825 elicits the following five concerns regarding genetic privacy and consent rights:

1. **No consent; Expanded testing**
2. **Nationalized government databases and registries**
3. **Tracking individuals with "disorders"**
4. **Research on citizens without consent**
5. **Coercive funding**

No Consent; Expanded Testing

H.R. 3825 will expand government genetic testing of citizens—newborns and children, and indirectly their families. It calls on the Advisory Committee on Heritable Disorders in Newborns and Children to "develop a model decision-matrix for newborn screening program expansion" and to "consider ways to ensure that States attain the capacity to screen for the conditions" the Committee recommends. Furthermore, the bill appears to call on the Committee to recommend testing *beyond* the actual genetic conditions for which children are to be tested. (p. 12) The Committee is asked to:

> *make recommendations that include the heritable disorders for which all newborns should be screened, including secondary conditions that may be identified as a result of the laboratory methods used for screening.* [my emphasis]

The term "secondary conditions" is left undefined, but the Centers for Disease Control and Prevention say "efforts are underway" to do predictive screening for "such chronic conditions as asthma and diabetes."[2] This level of testing and analysis of a child's blood and DNA are not likely known or contemplated by parents when the hospital pricks the baby's heel. Nor do many parents know that their baby's blood is sent to a State government agency for testing.

The Institute of Medicine has recommended, *"all screening, including for newborns, be voluntary"*[3] and that PKU testing not be mandatory.[4] Yet H.R. 3825 includes no parent consent requirements, suggesting only that grantees use federal funds to communicate to parents, "the right of refusal of newborn screening, if applicable." (p. 7)

Nationalized Government Databases and Registries

Almost every child born today is entered into a State government newborn genetic screening database without parent consent or knowledge. While most States mandate genetic testing for newborns, a few do not and some have a mandatory set and an optional set of conditions for which children must be tested. States that allow parent dissent for testing or retention of newborn blood usually require the hospital to send the State health department a form with information on the child and the signature of a parent. Thus, every child tested or not tested is entered into a State *database of children screened* usually operated within the state public health laboratory. Children who test positive for a genetic or heritable condition, whether a symptomatic condition or an asymptomatic trait, are often placed into a State *tracking registry of cases* for management and follow-up by the state health department staff. Parents have virtually no say in the matter.

H.R. 3825 will unify and nationalize these state newborn screening programs and data

[2] "A New Era in Newborn Screening: Saving Lives, Improving Outcomes." CDC Satellite Broadcast September 19, 2002. Accessed February 6, 2008.

[3] "Genetic testing policies must stress informed consent." Institute of Medicine, Division of Health Sciences Policy Committee on Assessing Genetic Risks. November 4, 1993.

[4] "Roundtable: The politics of genetic testing," Finneran, Kevin. Report from discussion at "The Genetics Revolution: A Catalyst for Education and Public Policy" (3/2006, Dallas) Statement by Robert M. Cook-Deegan, senior program officer, National Academy of Sciences. Accessed 2/11/08.

collection systems. On page 14, the bill calls for "coordination of surveillance activities, including standardized data collection and reporting," and "harmonization of laboratory definitions…in order to assess and enhance monitoring of newborn diseases." It requires the Secretary of the U.S. Department of Health and Human Services (HHS) to also coordinate with programs currently collecting data on persons with birth defects for HHS (Sec. 317C, p. 25), and on page 21 to "collect and analyze data on the incidence and prevalence of genetic and heritable disorders, as well as the poor health outcomes resulting from these disorders."

With these harmonized data on individuals, the Secretary will "operate regional centers" (p. 21) to do "epidemiological research" and "research on the prevention of poor health outcomes resulting from such disorders and secondary health conditions among individuals with such disorders." (pp. 21-22) These government-funded surveillance and research activities will encourage sharing of private data between State governments and ongoing government surveillance of individuals who have been placed in State registries after the discovery of a genetic, metabolic, or heritable defect. (p. 24, ln. 3-12)

H.R. 3825 provides false assurances on privacy: "All Federal laws relating to the privacy of information shall apply to the data and information that is collected under this section."(p. 25) However, government agencies are not under the HIPAA privacy Rule. Nor does the Rule require patient consent for government access to data for "public health activities" (§ 164.512(b)) In addition, the federal rule on research allows research to be conducted without patient consent if a review board at the government agency or other research institution allows the individual's consent rights to be *waived*. The only real protection for individual and family privacy is *opt-in* parent consent. Opt-out, refusal, objection and dissent rights are not consent rights and do not guarantee that parents are fully informed prior to the collection and testing of blood.

Tracking Individuals with "Disorders"

Federal grants in H.R. 3825 will go to entities that "establish, maintain, and operate a system to assess and coordinate treatment relating to congenital, genetic and metabolic disorders." (p. 26) These funds can be used to "coordinate ongoing followup treatment with individuals, families, primary care physicians, and appropriate subspecialists… after a newborn receives an indication of the presence or increased risk of a disorder on a screening test." (p. 30) Thus, infants and their families will be placed into tracking and registries just because the child has an "*increased risk of a disorder*." This could mean the presence of a genetic trait without a corresponding genetic condition.

What kind of disorders, traits, and risks could be registered and tracked? As the search for a gene for obesity, Alzheimer's disease and alcoholism progresses, the government list of trackable disorders may expand. As genetic research advances and data technology allows government agencies to share data online directly from hospitals and health plans, more children and families could find themselves entered into genetic tracking registries for ongoing monitoring and assessment. As noted above, data on the health outcomes of these individuals can be collected and analyzed.

Research on Citizens Without Consent

H.R. 3825 exposes every newborn tested—and their families—to becoming subjects of government research or government-funded research and analysis. The entire population tested by the newborn genetic screening program could find themselves analyzed far into the future. On page 31, those receiving government grants are encouraged to analyze data collected from newborn screenings to "*identify populations at risk* for disorders affecting newborns, examine and respond to health concerns, recognize and address relevant *environmental,*

behavioral, socioeconomic, demographic, and other relevant risk factors." [my emphasis]

On February 20, 2007, the Centers for Disease Control and Prevention (CDC) and the National Institutes of Health took part in a meeting held at the University of North Carolina in Chapel Hill. The following statement about newborn dried blood spots was made in a report on the meeting:

> *More states are keeping residual spots and making them available for additional research purposes. Methods to extract DNA from the DBS [dried blood spots] have been developed and have been used for genotyping in a variety of studies.*[5]

The CDC contemplates creating a databank of the DNA-filled newborn blood spots and using them—seemingly without parent consent—for additional purposes beyond the newborn genetic screening program, specifically genetic and other research:

> *Newborn screening (NBS) programs collect dried blood spots (DBS) in every state for the approximately 4 million children born each year. Leftover DBSs are available after routine metabolic, endocrine, hematologic and other screening tests so re-testing can be performed, if needed. Over 95% of newborns have leftover DBS retained by state programs for some time period. <u>These leftover DBS specimens are a unique, valuable population-based source for important public health surveillance and potential epidemiologic research, including population-based data on prevalence of genetic variants, markers of environmental exposure and infectious disease and constitute a specimen bank of a large cohort of state populations</u>…Assessing the impact of genetic variation on the health of populations will be critical to guide public health research, policy, and practice on using genetic information to prevent disease. In collaboration with state health departments and other partners, <u>we would like to examine the feasibility of establishing a bank of leftover newborn dried blood spot (DBS) specimens and assess the logistical structure for controlled access to a multi-state spot bank or a central spot bank</u>. The purpose of the banks would be to provide a unique resource for obtaining population-based data on prevalence of gene variants of public health significance, and the association of gene variants with disease and risk factors, including measuring markers of environmental exposure, infectious disease, or risk factors associated with developmental disabilities and chronic disease.*[6] [my emphasis]

In 2003, the Association of Public Health Laboratories conducted a study of the use of dried newborn blood spots in 49 state health departments.[7] Several of the study's results are listed below in the CCHC table.

<u>Note:</u> In the table below the term "unlinked" does not necessarily mean anonymous. It is not clear that "unlinked" samples cannot be *re-linked*. And regarding the sending of "anonymous" blood spots, consider a point made by Ann Cavoukian, Ph.D., Information & Privacy Commissioner in Canada in a 2005 presentation at the University of Toronto Law School:

> *It is impossible to completely anonymize DNA since there is always a means to identifying the tissue or the sample.*[8]

[5] "Meeting Report: The Use of Newborn Blood Spots in Environmental Research: Opportunities and Challenges." Andrew F. Olshan. Environmental Health Perspectives 2007 December; 115(12): 1767-1779. (published online August 30, 2007)

[6] "Banking Newborn Blood Spots for Public Health," a workshop convened by the CDC. 9/23-24/02 in Atlanta, GA. http://www.cdc.gov/genomics/training/conference/Spotbank.htm.

[7] "2003 APHL State Survey on Storage and use of Residual Dried Blood Spots," Centers for Disease Control and Prevention. n.d (presented at May 2004 Newborn Screening and Genetic Testing Symposium).

[8] "Privacy of Genetic Information," Ann Cavoukian, Ph.D. Information & Privacy Commissioner/Ontario and Ken Anderson, Assistant Commissioner. Powerpoint presentation at University of Toronto Law School, March 15, 2005.

CCHC TABLE: Data from APHL Study of 49 State Newborn Screening Programs

State Activity	Yes	No	Don't Know/No Response/Unsure
Use dried blood spots (DBS) to conduct research to evaluate existing newborn screening test or new screening tests	**34**	9	6
Use *identifiable* dried blood spots for evaluation of new and existing screening tests	20 *(of 34)* = 9 use identifiable and 11 use both identifiable and "unlinked" spots	12 *(of 34)* use only "unlinked" blood spots	8
Use dried blood spots (DBS) to conduct epidemiological research	**13**	30	6
Use *identifiable* DBS to conduct epidemiological research	5 *(of 13)*		6
Send DBS out of state for epidemiological research	5 *(of 13)*		7
Favor storage of *identifiable* dried newborn blood spots	37	8 – favor "unlinked"	4
Willing to send "anonymous" dried newborn blood spots for multi-state epidemiological study	20 *(1.74 million annual births)*	6 *(0.24 million annual births)*	23 *(1.99 million annual births)*

As Stanford professor Henry T. Greely notes in his paper on 'genomic biobanks,'

> *With a rich data set, [anonymization] will not work—some, and potentially all, of the donors could be re-identified.*[9]

On pages 35 - 38, H.R. 3825 authorizes additional research through a proposed *Hunter Kelly Research Program*. The program will receive funding from the National Institutes of Health to "expand the number of conditions for which screening tests are available" (p. 36). The program will also fund "experimental treatments and disease management strategies for *additional newborn conditions* and other genetic, metabolic, hormonal and/or functional conditions that can be detected through newborn screening..." (p. 36).

The Hunter Kelly research provision provides an open door to using the blood of around 4.2 million children born each year for various genetic research studies. The term '*additional newborn condition'* means "any condition that is not one of the core conditions designated by the Advisory Committee"(p. 36). In other words, these conditions may not be on the established committee list of conditions for testing. This provision could thus support the pilot projects—research and development of new tests—health departments sometimes now conduct on children without specific legal authority, parent consent or public notice.

Coercive Funding

H.R. 3825 and S. 1858 appropriations (CCHC Appendix B) are tied to compliance with the Advisory Committee's recommendations. For instance, on page 10, the grantee must

> *use grant amounts received…to adopt and implement the guidelines and recommendations of the Advisory Committee on Heritable Disorders in Newborns and Children…and shall include the screening of each newborn for the heritable disorders recommended by the Advisory Committee.*

[9] "The Uneasy Ethical and Legal Underpinnings of Large-Scale Genomic Biobanks." Henry T. Greely [Stanford University]. Annual Reviews. May 29, 2007.

State health departments and advocates of newborn screening will likely use the new federal grant dollars to push for expansion of newborn genetic testing programs—and to argue against parent consent even though *"no physician would dream of doing a test like this in the clinic without talking with parents."*[10]

CONSIDER THIS

How many different medical, or perhaps even behavioral,[11] conditions could a newborn child be screened for? While the American College of Medical Genetics recommends only screening children for 29 conditions (p. 2), some states like Minnesota and Indiana already test for more than 50 conditions, including the controversial test for cystic fibrosis, which is incurable. The potential for expansive government genetic testing is clear in the vision statement of the Midwest's 8-state Heartland Genetics and Newborn Screening Collaborative:

> *Do you see…every Heartland newborn screened for 200 conditions where early recognition makes a difference in their life and health?* [my emphasis]
>
> *Do you see…every new student in the Heartland with an individual program for education based on confidential interpretation of their family medical history, their brain imaging, their genetic predictors of best learning methods?*[12]

There are serious legal, ethical and self-determination implications to government-imposed genetic testing:

- The parent/patient right of consent for medical testing and procedures is a long-established legal right.
- Genetic tests are only predictive, but many erroneously consider them presumptive.
- States automatically send genetic test results to the child's doctor and permanent medical record where they could be used in the future for discriminatory purposes.
- Government data collection turns private data into a public record.
- Blood and test results held by the State are considered *government property*, available for whatever purposes future legislatures might choose, including genetic research or genetic fishing expeditions.
- Studies find false positives *(e.g. only 1 out of every 10 babies tested "positive" for Cystic Fibrosis has the disease* [13]*)* cause long-term anxiety in parents.
- Not everyone wants to know or think about his or her own health future—or let others know about it.

CONCLUSION

H.R. 3825 will expand newborn genetic testing, initiate identification of secondary conditions found in newborn DNA, expand government tracking systems, build intrusive government treatment monitoring and follow-up programs, strengthen government claims of ownership to the citizen DNA and genetic information, make citizens into unwitting research subjects, and violate the individual's right to "not know/not tell" their genetic predispositions.

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[10] "Talking with Parents – What are the Issues?" Ellen Wright Clayton, MD, JD, Vanderbilt University, Center for Genetics and Health Policy. Dated June 30, 2004. Given at May 2004 APHL conference.

[11] "Behavioral Genetics Research and Criminal DNA Databases," D.H. Kaye. Law and Contemporary Problems (Duke Law). Winter-Spring 2006.

[12] Heartland Genetics and Newborn Screening Collaborative: Strategic Plan 2006-2009. Heartland Regional Collaborative. http://heartland.ouhsc.edu. Accessed February 5, 2008.

[13] "Newborn Screening for Cystic Fibrosis, Implementation: October 1, 2007." (Letter to Practitioners) William Young, PhD. State of Michigan Department of Community Health, September 2007.

The proposed legislation does not acknowledge or uphold the rights of citizens to be free from government genetic testing, government surveillance and government-conducted/supported genetic research.

If enacted, H.R. 3825 and S. 1858 will expose citizens, young and old, to the genetic testing, surveillance and research agendas of government officials, genetic researchers, and other for-profit and non-profit advocates of comprehensive and predictive government genetic testing of the American public.

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Appendix A

History of the Newborn Screening Saves Lives Act of 2007

U.S. Senate

S 1858 passed the Senate on December 14, 2007. *Sen. Christopher Dodd* introduced it in Senate on July 27, 2007. There are 21 cosponsors.

The bill has been sent to the House where it was referred to the House Committee on Energy and Commerce.

On June 14, 2002, Sen. Dodd held a hearing in the Subcommittee on Children and Families: "Newborn Screening: Increasing Options and Awareness." Testifiers appeared to only include proponents: Peter van Dyck MS, MD, MPH at the Maternal and Child Health Bureau of the Health Resources and Services Administration in the U.S. Department of Health and Human Services; Jill Wood, parent from Alexandria, VA; Jeffrey Botkin, M.D., MPH, University of Utah School of Medicine; Scott Rivkees, MD, Connecticut Genetics Advisory Committee; and Brad Therrell, MS, PhD, Director National Newborn Screening and Genetics Resource Center.

U.S. House of Representatives

HR 3825 was introduced in the House on October 15, 2007 by *Rep. Lucille Roybal-Allard* (73 cosponsors) and referred to the Subcommittee on Health.

HR 3825 was preceded by **HR 2889**, the SHINE bill (Screening for Health of Infants and Newborns Act) sponsored by *Rep. Thomas M. Reynolds* (no cosponsors) introduced June 27, 2007, and referred to the Subcommittee on Health. *Sen. Hillary Clinton* is the author of SHINE (**S 1712**). It has no co-sponsors.

Appendix B

Appropriations in House and Senate bills* (as of March 3, 2008)

Purpose	House Appropriation - H.R. 3835	Senate Appropriation – S. 1858
Grants to expand and enhance testing, counseling, and health care services for "newborns and children having or at risk for heritable disorders"		**$76,875,000** (2008 – 2012)
Newborn Screening Grant Programs (Sec.399Z-1) to work with public and private entities to provide education and training for health care providers and newborn screening laboratory personnel education, screening.	**$5,000,000 for FY 2008** *and* "such sums as may be necessary for each of fiscal years 2009 through 2012"	
Grants to "establish, maintain, and operate a system to assess and coordinate treatment relating to congenital, genetic, and metabolic disorders;" training and education about newborn screening, counseling, testing, follow-up, treatment, specialty services and new technologies for doctors, laboratories, parents, families and patient advocacy and support groups.		**$76,875,000** (2008 – 2012)
Grants to establish, expand, improve newborn screening programs; establish or expand programs to reduce mortality or morbidity from heritable disorders; provide information and counseling for newborns and children with heritable disorders; improve access to screening…	**$15,000,000 for FY 2008** *and* "such sums as may be necessary for each of fiscal years 2009 through 2012"	
Grants to States to evaluate the effectiveness of newborn and child screening programs	**$5,000,000 for FY 2008** *and* "such sums as may be necessary for each of fiscal years 2009 through 2012"	**$25,625,000** (2008 – 2012)
Advisory Committee on Heritable Disorders in Newborns and Children whose duties shall include making recommendations that include "the heritable disorders for which all newborns should be screened, including secondary conditions that may be identified as a result of the laboratory methods used for screening" and recommendations for "surveillance activities" and "quality assurance, oversight, and evaluation of State newborn screening programs"	**$1,000,000 for FY 2008** *and* "such sums as may be necessary for each of fiscal years 2009 through 2012"	**$5,125,000** (2008 – 2012)
Clearinghouse of Newborn Screening Information	**$2,500,000 for FY 2008** *and* "such sums as may be necessary for each of fiscal years 2009 through 2012"	**$12,812,500** (2008 – 2012)
Laboratory quality, including "population-based pilot testing for new screening tools for evaluating use on a mass scale." (H.R. 3825)	**$5,000,000 for FY 2008** *and* "such sums as may be necessary for each of fiscal years 2009 through 2012"	**$25,625,000** (2008 – 2012)
Grants to public and non-profit groups for surveillance programs for heritable disorders screening, including the collection and analysis of "data on the incidence and prevalence of, as well as poor health outcomes resulting from 'heritable disorders'" and the coordination of activities with "existing birth defects surveillance activities."	**$15,000,000 for FY 2008** *and* "such sums as may be necessary for each of fiscal years 2009 through 2012"	
Establishment and maintenance of an Interagency Coordinating Committee on Newborn and Child Screening to assess activities and infrastructure, including activities on birth defects and developmental disabilities, and to "make recommendations for the establishment of regional centers for the conduct of applied epidemiological research to promote the prevention of poor health outcomes…"		**$5,125,000** (2008 – 2012)
Grants to deliver educational programs, establish a system to assess and coordinate treatment relating to congenital, genetic, and metabolic disorders, and "coordinate ongoing followup treatment with individuals, families, primary care physicians and appropriate subspecialists…after a newborn receives an indication of the presence or increased risk of a disorder on a screening test" and "analyze data collected from newborn screenings to identify populations at risk for disorders affecting newborns" and "recognize and address relevant environmental, behavioral, socioeconomic, demographic, and other relevant risk factors" (Sec 1115)	**$10,000,000 for FY 2008** *and* "such sums as may be necessary for each of fiscal years 2009 through 2012"	
Newborn screening research, including new screening technologies, expansion of number of conditions for which newborns are tested, experimental treatments and disease management strategies for "additional newborn conditions and other genetic, metabolic, hormonal and or functional conditions that can be detected through newborn screening for which treatment is not yet available"	Funding from the National Institutes of Health (NIH)	Funding from the National Institutes of Health (NIH)
Total Federal Funding 2008-2012	**>$58,500,000 + NIH funding**	**$228,062,500 + NIH funding**

* The bills vary in length and content. The House bill is 38 pages; the Senate bill is 20 pages. Where the appropriations are tied to similar language, the stated appropriations are listed in both the House and Senate column. Where the bills have different sections or significantly different language, the appropriation listed is only for the House or Senate bill in which the section or language appears.