

Shared Principles for 2019 Reauthorization of the Newborn Screening Saves Lives Act (P.L. 113-240)

Endorsing Organizations: American Academy of Pediatrics, American Association for Clinical Chemistry, American College of Medical Genetics and Genomics, American College of Obstetricians and Gynecologists, Association of Maternal & Child Health Programs, Association of Public Health Laboratories, Cure SMA, Cystic Fibrosis Foundation, EveryLife Foundation for Rare Diseases, Expecting Health at Genetic Alliance, March of Dimes, Muscular Dystrophy Association, National Organization for Rare Disorders (NORD), Parent Project Muscular Dystrophy (PPMD)

1. Increase authorized funding for newborn screening programs at the Centers for Disease Control and Prevention (CDC) and the Health Resources and Services Administration (HRSA)

Stakeholders have advocated in recent years for increased funding for newborn screening programs at CDC and HRSA. Additional federal funds are needed to ensure that states are prepared to implement new conditions added to the Recommended Uniform Screening Panel (RUSP) and that states continue to screen effectively for conditions already on state panels and ensure follow-up of identified infants. Our organizations have requested an additional \$20 million for programs at CDC and an additional \$8 million for programs at HRSA over FY 2017 levels.

Recognizing the significant need to support the US newborn screening system, Congressional appropriators increased funding for both CDC and HRSA programs in both FY 2018 and FY 2019. When reauthorizing the Newborn Screening Saves Lives Act, Congress should increase the authorized funding for programs at CDC and HRSA to levels outlined below.

Program	Authorized Level	FY17	FY18	FY19	Request for Authorization FY20 – FY24
HRSA – Heritable Disorders	\$11,900,000	\$13,883,000	\$15,883,000	\$16,383,000	\$21,883,000
CDC – NBS Quality Assurance Program	\$8,000,000	\$8,400,000	\$13,400,000	\$16,000,000	\$29,650,000
CDC – NBS for SCID	n/a	\$1,250,000	\$1,250,000	\$1,250,000	included in NBSQAP request

2. Ensure continued funding for newborn screening research at the National Institutes of Health (NIH)

NIH, through the Hunter Kelly Newborn Screening Research Program, is essential to advancing the field of newborn screening research. Congress should make the Hunter Kelly Newborn Screening Research Program a mandatory activity to ensure NIH’s essential research in this area continues.

3. Refine permitted activities of the Hunter Kelly Research Program to focus on promising new research

Congress should update NIH’s existing authorities to ensure the Hunter Kelly Research Program continues to focus research activities on conditions with a strong likelihood of recommendation for the RUSP.

4. Enhance the Hunter Kelly Research Program to Better enable private grants for newborn screening pilot studies

The Hunter Kelly Grant program is a vital source of funding for research into new screening technologies, including the piloting of screening tests for diseases not yet on the RUSP. By encouraging the Department of Health and Human Services (HHS) to use existing mechanisms that allow private entities to support grants within the Hunter Kelly program, states will have more opportunities to screen for new conditions while also collecting data that can later be used to nominate new conditions to the RUSP.

5. Update language on informed consent to reflect final revisions to the Federal Policy for the Protection of Human Subjects (the Common Rule)

In mid-2018, final revisions to the Federal Policy for the Protection of Human Subject, also known as the Common Rule, became effective. Pursuant to language in the Newborn Screening Saves Lives Act of 2014, the changes made to the use of nonidentified newborn screening dried blood spots for research outlined in Section 12 sunset with promulgation of the Common Rule. Congress should update the statute to affirm that the revised Common Rule should be followed in federally-funded research using nonidentified newborn screening dried blood spots.

6. Revise CDC's authority to enhance disease detection in newborns through improved data analysis, test result interpretation, data harmonization and dissemination of laboratory best practices

For over 40 years, CDC's scientists and public health experts have provided leadership and technical laboratory expertise to newborn screening programs which have expanded to include new conditions, the evaluation of more complex biomarker profiles and the introduction of new testing platforms, including DNA-based tests. CDC is actively developing newborn screening tests with features that address evolving state needs for timely state implementation of new conditions added to the RUSP, together with enhanced detection of existing conditions. Congress should revise CDC's existing authorities to reflect the essential work its Newborn Screening Quality Assurance Program is doing to advance newborn screening throughout the United States.

7. Expand HRSA's authority to support educational programming to address emerging challenges for newborn screening

Congress should permit expanded educational programming to improve public understanding of newborn screening based on health promotion and communication science principles. While the Clearinghouse of Newborn Screening Information provides a central location for newborn screening information, based on the media reports and continued legal challenges, a more proactive approach to understanding the public's knowledge and perception of newborn screening is needed in order to implement proven communications strategies and to monitor for improvements in public perception of screening within the context of an evolving public health program.

8. Modify nonduplication requirement for clearinghouse of newborn screening information

The nonduplication requirement for the clearinghouse of newborn screening information has led to limitations in expansion of education efforts in response to new newborn screening conditions and emerging topics. For instance, HRSA has limited the ability of some grantees to develop and maintain a website to support outreach efforts, citing this provision. Congress should modify the existing requirement to clarify the clearinghouse should enhance other federal newborn screening information-sharing activities.

9. Improve public and stakeholder understanding of the RUSP nomination process

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) is committed to developing resources for stakeholders so that they better understand the nominations and

evidence review process. Unfortunately, many stakeholders remain uncertain about data requirements for nominations and types of technical assistance available for entities seeking to add new conditions to the RUSP, including how international data can be used. Congress should direct the ACHDNC to develop consumer-friendly materials outlining the RUSP nomination process; evidence requirements and standards, including incorporating international data in support of adding a new condition to the RUSP; and information on receiving technical assistance for a RUSP nomination packet from ACHDNC without introducing conflict of interest. This information should be made available on a publicly accessible website and updated as necessary.

10. Commission an expert report on the modernization of newborn screening

For the past 50 years, newborn screening has saved and improved the lives of children across the United States. It is one of the greatest public health success stories of our time. However, new technologies, the development of treatments for rare disorders, the growing availability of early intervention treatments for individuals with late onset symptoms, and ever increasing rates of data acquisition have put pressure on the newborn screening system to modernize. Congress should commission a National Academy of Medicine (NAM) report to make consensus recommendations to shift to a 21st century newborn screening system. This report should include, but not be limited to the following:

- An examination of the RUSP review and recommendation processes with goals to identify factors that impact decisions on adding new conditions to the RUSP; to describe challenges posed by newly nominated conditions, including low-incidence diseases, late onset variants and new treatments without long-term efficacy data; and to outline cogent recommendations to modernize the screening process.
- A review of the barriers that preclude states from adding new RUSP conditions to their state screening panels with recommendations about the necessary resources for states to implement RUSP recommendations.
- An analysis of current federal- and privately-funded newborn screening research with recommendations to optimize the capacity of this research, including piloting multiple prospective conditions at once and to address rare disease questions.
- An examination of new and emerging technologies that would permit screening for new categories of disorders, or would make current screening more effective, more efficient or less expensive.
- An overview of technological and other infrastructure needs that will improve timeliness of diagnosis and short- and long-term follow-up for infants identified through newborn screening and hence improve public health surveillance.
- An examination of current and future communication and educational needs for priority stakeholders and the public to promote understanding and knowledge of a modernized newborn screening system with an emphasis on evolving communication channels and messaging.
- An analysis of the extent to which newborn screening yields better data on the disease prevalence for screened-conditions and improves long-term outcomes for those identified through newborn screening, including existing systems supporting such data collection and recommendations for systems that would allow for improved data collection.

The expert report should be completed within 1.5 years from the time HHS enters into a contract with NAM. Congress should appropriate \$2 million for this activity.

Reauthorization of the Newborn Screening Saves Lives Act (P.L. 113-240) – Changes to Statutory Language

1. Increase authorized funding for newborn screening programs at CDC and HRSA

42 USC 300b-16: Authorization of appropriations for newborn screening programs and activities

There are authorized to be appropriated-

(1) to carry out sections 300b–8, 300b–9, 300b–10, and 300b–11 of this title, ~~\$11,900,000~~ \$21,883,000 for each of fiscal years ~~2015~~ 2020 through ~~2019~~ 2024; and

(2) to carry out section 300b–12 of this title, ~~\$8,000,000~~ \$29,650,000 for each of fiscal years ~~2015~~ 2020 through ~~2019~~ 2024.

2. Ensure continued funding for newborn screening research at NIH

42 USC 300b-15: Hunter Kelly Research Program

The Secretary, in conjunction with the Director of the National Institutes of Health and taking into consideration the recommendations of the Advisory Committee, ~~may~~ shall continue carrying out, coordinating, and expanding research in newborn screening

3. Refine permitted activities of the Hunter Kelly Research Program to focus on promising new research

42 USC 300b-15: Hunter Kelly Research Program

(D) conducting pilot studies on conditions recommended by ~~or~~ with a high probability of being recommended by the Advisory Committee on Heritable Disorders in Newborns and Children to ensure that reliable newborn screening technologies are evaluated and ready for use ~~they are ready for implementation in newborn screening programs~~; and

4. Enhance the Hunter Kelly Research Program to better enable private grants for newborn screening pilot studies

42 SC 300b-15: Hunter Kelly Research Program

In carrying out the research program under this section, the Secretary and the Director-

(1) shall ensure that entities receiving funding through the program will provide assurances, as practicable, that such entities will work in consultation with the appropriate State departments of health, and, as practicable, focus their research on screening technology not currently performed in the States in which the entities are located, and the conditions on the uniform screening panel (or the standard test existing on the uniform screening panel); ~~and-~~

(2) may accept donations from private, including not-for-profit entities, in accordance with federal law.

5. Update language on informed consent to reflect final revisions to the Federal Policy for the Protection of Human Subjects (the Common Rule)

§289. Institutional review boards; ethics guidance program

(a) In General.—Research on nonidentified newborn dried blood spots shall be considered secondary research with nonidentified biospecimens ~~carried out on human subjects meeting the definition of section 46.102(f)(2) of title 45, Code of Federal Regulations,~~ for purposes of Federally funded research conducted pursuant to the Public Health Service Act [42 U.S.C. 201 et seq.]. ~~until such time as updates to the Federal Policy for the Protection of Human Subjects (the Common Rule) are promulgated pursuant to subsection (c).~~ For purposes of this subsection, sections 46.116(c) and 46.116(d) of title 45, Code of Federal Regulations, shall not apply.

~~———— (b) Effective Date.—Subsection (a) shall apply only to newborn dried blood spots used for purposes of Federally funded research that were collected not earlier than 90 days after the date of enactment of this Act [Dec. 18, 2014].~~

~~———— (c) Regulations.—Not later than 6 months after the date of enactment of this Act, the Secretary of Health and Human Services shall promulgate proposed regulations related to the updating of the Federal Policy for the Protection of Human Subjects (the Common Rule), particularly with respect to informed consent. Not later than 2 years after such date of enactment, the Secretary shall promulgate final regulations based on such proposed regulations.~~

6. Revise CDC’s authority to enhance disease detection in newborns through improved data analysis, test result interpretation, data harmonization and dissemination of laboratory best practices

§300b–12. Laboratory quality and surveillance

(1) quality assurance for laboratories involved in screening newborns and children for heritable disorders, including quality assurance for newborn-screening tests, timeliness for processing such tests, development of new screening tests, ~~performance evaluation services,~~ and technical assistance and technology transfer to newborn screening laboratories to ensure analytic validity and utility of screening tests; ~~and~~

(2) appropriate quality control and other test performance ~~test~~ materials to evaluate the performance of new screening tools; and

(3) performance evaluation services to enhance disease detection, including the development of tools, resources and infrastructure to improve data analysis, test result interpretation, data harmonization and dissemination of laboratory best practices.

7. Expand HRSA’s authority to support educational programming to address emerging challenges for newborn screening

§300b–8. Improved newborn and child screening for heritable disorders

(3) to develop and deliver educational programs (at appropriate literacy levels) about newborn screening counseling, testing, follow-up, treatment, ~~and~~ specialty services, and long-term care to parents, families, and patient advocacy and support groups; that assess the target audience’s current knowledge, incorporate health communications strategies and measure impact;

8. Modify nonduplication requirement for clearinghouse of newborn screening information

§300b–11. Clearinghouse of newborn screening information

(c) Nonduplication: In carrying out activities under this section, the Secretary shall ensure that such activities minimize duplication ~~and supplement, not supplant, existing information sharing efforts~~ and complement other federal newborn screening information-sharing activities.

9. Improve public and stakeholder understanding of the RUSP nomination process

§300b–10. Advisory Committee on Heritable Disorders in Newborns and Children

(b) Duties

The Advisory Committee shall-

[...]

(7) consider ways to ensure that all States attain the capacity to screen for the conditions described in paragraph (3), and include in such consideration the results of grant funding under section 300b–8 of this title; ~~and~~

(8) develop, maintain, and publish on a publicly-accessible website consumer-friendly materials detailing -

(A) the uniform screening panel nomination process, including data requirements, standards, and the use of international data in nomination submissions; and

(B) the process for obtaining technical assistance for submitting nominations to the uniform screening panel and detailing the instances in which the provision of technical assistance would introduce a conflict of interest for members of the advisory committee; and

~~(8)~~(9) provide such recommendations [...]

10. Commission an expert report on the modernization of newborn screening

New legislative language modeled off of requirements for an NAM (formerly IOM) report found in [P.L. 110-275](#).

NAM REPORT ON THE MODERNIZATION OF NEWBORN SCREENING

(a) Study and Report by the NAM.--

(1) Study.-- Not later than 60 days after the date of the enactment of this Act, the Secretary of Health and Human Services shall enter into a contract with the National Academy of Medicine of the National Academies (in this section referred to as “NAM”) under which NAM shall conduct a study on the following:

(A) the uniform screening panel review and recommendation process to identify factors that impact the decision to add new conditions to the uniform screening panel, reflect challenges posed by newly nominated conditions, including low-incidence diseases, late onset variants and new treatments without long-term efficacy data, and make recommendations to modernize the process.

(B) the barriers that preclude states from adding new uniform screening panel conditions to their state screening panels and recommendations on resources needed to help states implement uniform screening panel recommendations.

(C) the current state of federal- and privately-funded newborn screening research and recommendations for optimizing the capacity of this research, including piloting multiple prospective conditions at once and addressing rare disease questions.

(D) new and emerging technologies that would permit screening for new categories of disorders, or would make current screening more effective, more efficient or less expensive.

(E) technological and other infrastructure needs to improve timeliness of diagnosis and short- and long-term follow-up for infants identified through newborn screening and improve public health surveillance.

(F) current and future communication and educational needs for priority stakeholders and the public to promote understanding and knowledge of a modernized newborn screening system with an emphasis on evolving communication channels and messaging.

(G) the extent to which newborn screening yields better data on the disease prevalence for screened-conditions and improves long-term outcomes for those identified through newborn screening, including existing systems supporting such data collection and recommendations for systems that would allow for improved data collection.

(2) Report.—Not later than 18 months after the effective date of the contract under paragraph (1), the NAM, as part of such contract, shall submit to the Secretary of Health and Human Services and the appropriate committees of jurisdiction of Congress a report containing the results of the study conducted under paragraph (1), together with recommendations for such legislation and administrative action as the Institute determines appropriate.

(b) Funding.--Out of any funds in the Treasury not otherwise appropriated, there are appropriated for the period of fiscal years 2020 and 2021, \$2,000,000 to carry out this section.