Newborn Screening Informed Consent Amendment

In 2008, Congress passed the original Newborn Screening Saves Lives Act (P.L. 110-204), which established national newborn screening guidelines and helped facilitate comprehensive newborn screening in every state. The Act was first reauthorized in 2014. The Newborn Screening Saves Lives Reauthorization Act of 2019 passed the House in July, but the Senate Health, Education, Labor, and Pensions (HELP) Committee has not yet held a markup of the bill. This is because Senator Rand Paul (KY) is pushing for a change to the bill that would require parents to opt-in to allow their newborn’s unidentified dried blood spots (DBS) to be used for research which would create a detrimental burden for rare disease research. This change is similar to an amendment by Senator Paul included in the 2014 reauthorization that threatened the development of newborn screening. Following a public comment period in 2017, the Department of Health and Human Services (HHS) recognized the harm this policy could do to rare disease research and reversed the decision.

Key Bill Provisions Currently Held Up
Reauthorization of the Health Resources and Services Administration (HRSA) grants to states to expand and improve their screening programs, educate parents and health care providers, and improve follow-up care for infants with a detected condition.

Reauthorization of the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC), which provides states with a Recommended Uniform Screening Panel (RUSP) to help ensure every infant is screened for conditions which have a known treatment. All official business of the committee has been halted due to the September 30 expiration.

Direction for the National Academy of Medicine to conduct a study on how to modernize the newborn screening program.

Increases to authorized funding levels for these programs.

Newborn Screening Research Effected
The DBS used for newborn screening provide benefits long after the NBS panel for the individual child. Public health laboratories and scientific researchers need them to conduct life-saving research to improve the current tests and work to develop new treatments for the thousands of rare diseases still without a cure. 19.8 percent of published studies using DBS centered around quality improvement for NBS tests to improve the speed and success rates of the current testing technology. In addition, 72.4 percent of published studies used DBS to develop new tools and treatments for diseases not yet on the RUSP so that NBS panels can continue to grow and provide early diagnosis that will save countless lives. To conduct either form of rare disease research, the scientists require 3,000 to 10,000 unique samples to properly test the range of the tests. The storage of DBS is essential to provide the samples needed to create and improve the tests responsible for one of the most successful public health programs in the country.

Previous Government Action on Informed Consent for Dried Blood Spot Research
In 2015, the HHS Secretary’s Advisory Committee on Human Research Protection noted that requiring “…informed consent for research use of [DBS] will present a significant challenge…” and that research ethics boards “…have traditionally considered research on such samples to involve minimal risk and impracticable without a waiver of informed consent.”

Concerns with re-identifiable information of the DBS were remedied through the 21st Century Cures Act in 2016. The law states that if there is “…at least a very small risk…” that a de-identified biospecimen, such as DBS, may be re-identified, any federally-funded researcher must obtain a certificate of confidentiality to engage in the research. This ensures the protection of people’s privacy while allowing the vital rare disease research can continue to function.

Effect of Informed Consent on the State Level
During a California NBS technology pilot study, the researchers were required to obtain informed consent. Only 52 percent of newborns were invited to participate in the study. This was a result of hospital burden, not a lack of consent. 90 percent of parents consented to participate, indicating a willingness of parents to allow researchers to use children’s DBS for research. However, 20 percent of hospitals refused to participate in the study, with increased burden on hospital staff the most cited reason for not participating. In addition, for the hospitals that did participate, only 23 percent offered screening to more than 75 percent of newborns.

In Michigan, more than half of the parents who declined the use of the DBS for research did not fill in the informed consent form properly, resulting in a passive decline of consent.

In a Massachusetts pilot study for two diseases on the RUSP, more than 99 percent of parents consented to the use of DBS for research.