

Newborn Screening Portfolio Evaluation

Advisory Committee on Heritable Disorders in
Newborns and Children

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Purpose of the Portfolio Evaluation

- The purpose of the evaluation was to understand
 - the needs of the newborn screening system and its stakeholders,
 - the role of HRSA programs in addressing those needs, and
 - the unmet needs to inform future programs

Goals of the Newborn Screening System

- Enhance, improve, or expand the ability of state and local public health agencies to provide screening, counseling, and health care services to newborns and children having heritable disorders.
- Provide education, training, and technical assistance (TA) to lab personnel and other genetics/health care professionals on the implementation of state-based public health newborn screening programs.
- Establish, maintain, and operate a system to assess and coordinate follow-up and treatment related to congenital, genetic, and metabolic conditions.
- Improve the timeliness of newborn screening from specimen collection through diagnosis.
- Develop and provide education to, and engage with, consumers (i.e., parents, families, patient advocacy groups) about screening, counseling, follow-up, and treatment to increase awareness, knowledge, and understanding of newborn screening and genetic conditions.
- Improve health equity and health outcomes of individuals with genetic conditions, reduce morbidity and mortality caused by genetic conditions (including congenital and metabolic disorders), and improve the quality of coordinated and comprehensive genetic services to children and their families.

HRSA-Funded Newborn Screening Programs

- Newborn Screening Data Repository and Technical Assistance Program
- Quality Improvement in Newborn Screening
- Newborn Screening Family Education Program
- Newborn Screening State Evaluation Program
- Newborn Screening Implementation Program Regarding Conditions Added to the Recommended Uniform Screening Panel
- Improving Timeliness of Newborn Screening Diagnosis

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Evaluation Methods

Evaluation Questions

- To what extent has the portfolio of programs contributed to achieving HRSA's overall NBS program goals?
- What are the current needs of the NBS system?
- What are the unmet needs or gaps that currently exist in the portfolio?
- What are the expected needs of the NBS system in the future?

Data Collection

- Primary data collection with 52 stakeholders
 - Stakeholder groups included grantees, NBS staff, parents, patient advocacy representatives, clinicians, and subject matter experts
 - Individual interviews (n=31), each lasting about 60 minutes
 - Six focus groups (n=21), each lasting about 90 minutes
- Secondary data collection
 - Data gathered from grantee-reported materials or other sources (e.g., NewSTEPs data repository)
 - A review of the published NBS literature
 - An environmental scan of NBS websites and partner organizations

Analysis Approach

- Interviews and focus groups were recorded and later transcribed for analysis.
- Rapid turnaround analysis techniques were used to distill findings
 - A template was used to organize and condense the qualitative data based on key topics of interest
 - A tagging procedure was used to label data from the interview notes by NBS goal and evaluation question
- Summaries for each stakeholder group were created for each evaluation question within each goal.
- Concurrently, the team aligned results from the literature review and the environmental scan to support the primary data collected.

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Evaluation Findings

Goal 1: Enhance, improve, or expand the ability of state and local public health agencies to provide screening, counseling, and healthcare services to newborns and children having heritable disorders.

- HRSA's portfolio of programs has made progress over the past decade in creating a more efficient and proficient NBS system with an emphasis on labs and screening.
- HRSA funding has supported states' ability to expand screening of new RUSP disorders.

"I don't think we could do what we do [without HRSA programs]. They're the ones that help us move it forward. They connect us with other states. They provide us with screening algorithms...I would say these programs are our go-to on screening."

Goal 1: Current and future needs of NBS system

- **Additional federal guidance:** Stronger federal guidance is needed to improve the “patchwork” system of the NBS system.
 - Concern about efforts being duplicated across states and about equity due to differences in state funding of NBS.
- **NBS system capacity because of new treatments and screening technology:** There are concerns related to treatment availability, sample sizes for clinical trials, and feasibility and readiness of states to implement screening for new conditions
 - Whole exome and genome sequencing are becoming more readily available.
- **Enhanced data interoperability:** Better infrastructure is needed to enable data sharing, data standards, and harmonization across the NBS system.
 - Benefits of interoperability include increasing efficiency in the system, increasing testing accuracy, and improving outcomes for identified infants.

Goal 1: Potential solutions to address gaps and needs

- **Collaborate among federal agencies and funding programs**, including integrating NBS with other programs and developing cohesive approaches to NBS among stakeholder groups.
- **Increased federal guidance on NBS**, including developing standards by condition to help systematize the implementation of the RUSP at the state level and guidance on how quickly states should begin screening for new conditions.
- **Invest in expanding the NBS workforce** through additional funding to states, training programs for medical students/genetic counselors, fellowships/certification programs for primary care providers, or creating incentives for working in NBS similar to the National Institutes of Health's (NIH's) loan repayment program.
- **Conduct an evaluation of NBS system** to assess the effectiveness and outcomes of NBS.
- **Evaluation of RUSP evidence review process** to address current and future needs.
- **State funding to support and enhance NBS** to limit fee increases, buy new equipment, pay for new staff, and support TA.

Goal 2: Provide education, training, and technical assistance (TA) to lab personnel and other genetics/health care professionals on the implementation of state-based public health newborn screening programs.

- HRSA's strong support for education and training opportunities for lab, follow-up, and health care professionals, particularly around timeliness, adding new conditions, and the data repository, has successfully engaged state NBS programs of all sizes
- However, there is not a widespread focus on educating healthcare providers.

“HRSA's [NBS programs] are making huge efforts to provide education and training and TA; it's continuous and every month. It's unprecedented the amount of time and energy spent, even during this past challenging year, to provide opportunities for people to enhance their skills and move forward with different projects, whether in the laboratory or with education and training.”

Goal 2: Current and future needs of NBS system

- **Additional training and TA for state lab and follow-up staff**
 - Topics such as data analytics, adding new conditions, input on goals and measures, long-term follow-up, and guidance on data collection.
 - Smaller or under-resourced states need more TA than larger or well-equipped states.
- **Additional training and TA for health care providers**
 - Training and education on NBS and communicating with families about NBS is needed for health care providers.
 - Primary care providers (PCPs) frequently have very little familiarity with the condition, what resources are available, or what next steps the family should take.
 - Increase awareness of the Communication Guide and update ACT sheets.

Goal 2: Potential solutions to address gaps and needs

○ Provide training and TA

- For PCPs on NBS and on specific conditions
- For specialists on confirmatory testing algorithms and guidelines for follow-up
- For hospital staff on dried bloodspot collection and the importance of NBS
- For lab personnel on new screening methods and technology, evaluating cutoffs, and communication with follow-up staff

○ Revise models of training and TA

- Provide support to “early adopter” states to mentor those who are funded after
- Create a repository for materials such as screening validation or verification plans
- Provide continuing education credits for PCPs

Goal 3: Establish, maintain, and operate a system to assess and coordinate follow-up and treatment related to congenital, genetic, and metabolic conditions.

- HRSA NBS programs have played a key role in the success of short-term follow-up, but long-term follow-up is lacking.

“Funding can’t support the weight of Goal 3.”

Goal 3: Current and future needs of NBS system

- **Lack of a national long-term follow-up system:** Perceived needs include the following:
 - Clear definition of long-term follow-up
 - Federal long-term follow-up registry with a clearly defined goal and purpose
 - Guidance regarding who is responsible for collecting data
 - Integration of long-term follow-up into quality measure
 - Additional resources for states to implement
- **State-to-state variability:** There is inconsistency across states for following up with families' and PCPs' knowledge on conditions
 - Access to consultants or specialists who are familiar with the condition and can provide just-in-time education varies across states; not enough genetic counselors in many areas
 - Telehealth can increase access to care but not where internet access is limited
 - Systems are needed for connecting families with psychosocial supports after a positive screen/diagnosis

Goal 3: Potential solutions to address gaps and needs

- **Create a long-term follow-up system and track health outcomes**
 - Collaborate with federal agencies and patient advocacy groups
 - Create a Center of Excellence to support the collection of long-term follow-up data
 - Invest funds in three areas: time for health department staff who have skills to maintain and analyze data, treatment for children who otherwise would have been lost to follow-up, and support more specialists to follow children as they grow
- **Coordinate treatment and support**
 - Provide condition-specific guidance around long-term follow-up treatment and support, including guidance on confirmatory testing methods, cutoffs, and clinical care guidelines
 - Link families to advocacy groups for emotional and informational support
 - Create a clearinghouse of resources for clinicians and families about insurance coverage, treatment options and payment support, and educational needs (e.g., early intervention)

Goal 4: Improve the timeliness of newborn screening from specimen collection through diagnosis

- Timeliness programs were seen as a huge success story in NBS.
- HRSA programs were attributed to making significant improvements in timeliness.

“I believe without NewSTEPs360, I don't think we would be where we are now.”

Goal 4: Current and future needs of NBS system

- **Continued focus on timeliness and QI:** Maintain timeliness standards through ongoing education, QI efforts, and funding.
- **Timeliness data on diagnosis and treatment:** Timeliness should include the timeframe for getting babies into treatment in a timely manner.
- **Condition-specific timeliness goals:** Focus on the most time-sensitive conditions.
- **Lack of education and training for providers on timeliness:** Education and training on timeliness for providers is often left up to states. Some states would benefit from help with provider education and training.

Goal 4: Potential solutions to address gaps and needs

- **Support to improve timeliness of diagnosis:** Work to reduce the amount of time between a positive NBS screen and confirmed diagnosis and treatment by providing timelines metrics for diagnosis and treatment and educating providers on the importance of timely diagnosis and treatment.
- **Funding and training/TA on automated data entry processes:** Automating data entry into the NewSTEPs repository would reduce the amount of burden, specifically staff time, and likely also reduce data entry errors.
- **Education and training for providers:** Provide resources to states and consider using MOC-4 modules, grand rounds, and personal stories to educate providers on the importance of timeliness for certain conditions.
- **Continued state funding to support timeliness:** Some states need continued support with specimen collection, transport of dried bloodspots, and screening indicators.

Goal 5: Develop and provide education to, and engage with, consumers about screening, counseling, follow-up, and treatment to increase awareness, knowledge, and understanding of newborn screening and genetic conditions.

- Current and previously funded HRSA projects have increased the amount, quality, and availability of educational resources for families about NBS
- Educational materials are generally perceived as high quality and accurate, but they lack visibility among all stakeholders.

“We probably lean on them [HRSA] for a lot of our parent education. They’ve been able to give us some documents we can actually put our own state logo on, so we don’t have to be the SMEs, which is great. The ability to use that information and provide it to families has been very helpful.”

Goal 5: Current and future needs of NBS system

- **Timing of dissemination of materials:** Education should happen earlier than it does in the current system. There is a need for more creative targeting of parents in the prenatal period.
- **Format of materials:** Parents typically use the internet to seek information, but the quality of resources online varies.
- **Tailoring of materials:** There is a need to target content appropriately for parents and to provide more education and support about non-medical challenges. Education is particularly lacking for some groups, including those with home births and those who want to opt out of testing.
- **Consistency:** The quality of educational materials varies by state. There is a need for a national, reliable source.

Goal 5: Potential solutions to address gaps and needs

- **Develop a variety of materials targeted to reach diverse audiences:** Focus on making materials relevant and usable at different time points and for specific audiences, develop alternate dissemination strategies to increase the visibility of available resources.
- **Determine effective dissemination strategies:** The communication strategy should involve connections with other stakeholder groups, such as OB-GYNs, PCPs, specialists, and family support organizations.
- **Education on new conditions:** Develop an education and support strategy for families after the diagnosis.
- **Partner with new patient advocacy groups:** Collaborating with condition-specific advocacy organizations to develop materials is particularly helpful because families view them as trusted sources of information.

Goal 6: Improve health equity and health outcomes of individuals with genetic conditions, reduce morbidity and mortality caused by genetic conditions, and improve the quality of coordinated and comprehensive genetic services to children and their families.

- HRSA continues to make improvements in health equity and health outcomes through
 - Support, training, TA provided to the NBS system (e.g., new conditions, timeliness)
 - Education to diverse families

Goal 6: Current and future needs of NBS system

- **Cost of care:** Lack of insurance drives a gap in equity as it serves as barrier to access when the cost of care is prohibitive. The high cost of medical food and specialty childcare is also problematic.
- **Equal access to care:** There are regional gaps in equity, as some states are better equipped to expand their list of conditions and provider services, which can result in loss to follow up.
- **Social support for families:** Children born to families of lower socioeconomic status have higher rates of loss to follow-up with care and families are less likely to be connected to other families for support.
- **Systemic racism and implicit bias:** Systemic racism and implicit bias can impact the care that families receive. Language barriers can also prevent families from accessing care.
- **Additional training and support:** Differences in resources dedicated to training and program improvement can deepen existing disparities between states.

Goal 6: Potential solutions to address gaps and needs

- **Education and training on implicit bias and structural racism:** Provide professional education, training, and TA for NBS staff, state advisory committees, hospital staff, and providers.
- **Long-term follow-up system:** This would track outcomes of babies with a positive screen and between children with the same diagnosis to identify any existing disparities post-diagnosis.
- **System of support and coordination for families:** Develop a system that helps provide more support and coordination for families to access genetic services for their child.
- **Better support for non-English speaking families:** Develop educational materials in different languages and better support services for non-English speaking families.
- **Adequate metrics for measuring program effectiveness:** Develop adequate metrics to measure the effectiveness of programs.

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Recommendations

Policy Recommendations

- **Create strategic plan for NBS**
 - Identify specific goals for addressing current and future gaps
 - Created by a group of NBS experts
 - Address areas of overlap between federal agencies
- **Conduct an evaluation of state NBS programs to assess outcomes and needed areas of improvement**
 - Process evaluation
 - Needs assessment and plan with implementation steps
- **Provide state-level funding to implement new conditions and reduce state-to-state variability**
 - Funding through Title V Maternal and Child Health Services Block Grants
 - Planning grants to states

Infrastructure Recommendations

- Create long-term follow-up registry to track health outcomes
 - Similar infrastructure exists through condition-specific registries and the Newborn Screening Translational Research Network
- Continued focus on interoperability
 - Support states in developing interoperability plans and implementation

Practice Recommendations

- **Continue to support technical assistance and timeliness programs**
 - Knowledge development, leadership, and implementation support
 - Additional methods of providing TA could be considered, such as coaching or expert consultation
 - Tiered support model depending on program size, performance, and need
- **Wraparound support for RUSP conditions**
 - Onboarding new conditions to state panels
 - Education to healthcare providers, both PCPs and specialists,
 - Connecting families to services and supports, both informational and emotional

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