

Appendix F: Newborn Screening and Genetics

Focus Area Name

Newborn Screening and Genetics

Focus Area Contact Information

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Approximate Average Annual Award: \$2,300,000

Funding Opportunity Description

1. Background

State and territorial newborn screening (NBS) programs test infants for diseases that require immediate medical treatment but are not apparent at birth. Nearly 4 million newborns are tested annually in the United States, and thousands of infants and children are saved from severe disability and death through early detection and treatment. Newborn screening laboratories face unique challenges as they implement new tests and conduct ongoing screening to rapidly and accurately identify infants affected by harmful or fatal diseases. Laboratories must rapidly integrate tests for an ever-growing number of diseases, adopt new technologies, manage and interpret increasingly complex newborn screening data, and operate effectively within an NBS community that includes policymakers, advocacy groups, and health care providers. In order to overcome these challenges, NBS laboratories need a national forum to address laboratory practice issues and other emerging issues across the newborn screening system.

Healthy People 2020

This focus area supports the Healthy People 2020 Maternal, Infant, and Child Health Objectives:

Maternal, Infant, and Child Health, Objective 1: Reduce the rate of fetal and infant deaths.

Maternal, Infant, and Child Health, Objective 3: Reduce the rate of child deaths.

Maternal, Infant, and Child Health, Objective 32: Increase appropriate newborn blood-spot screening and follow-up testing.

Other National Public Health Priorities and Strategies

N/A

2. CDC Project Description

a. Approach

I. Purpose

The purpose of the Newborn Screening focus area is to improve national NBS laboratory capability and practice and strengthen the role of NBS laboratories within the newborn screening system. Competent and well-integrated NBS



laboratories will increase the number of newborns screened for harmful or deadly diseases, as well as those identified and referred for life-saving medical treatment.

II. Outcomes

Activities in this focus area should achieve or contribute to the following intermediate outcomes (refer to section ii, “Outcomes” under Part II, A-2-a or the overall logic model in this NOFO for a full list of outcomes of this cooperative agreement):

IO-1. Enhanced communities of practice and other collaborative relationships among and between laboratories and other stakeholders

- Stronger roles for public health laboratories (PHLs) in state NBS programs and the NBS system

IO-2. Established information sharing systems across public health laboratories and with other key stakeholders

- More training for PHLs in existing and emerging NBS testing methods and data interpretation approaches, such as molecular detection, next-generation sequencing (NGS), and bioinformatics
- More guidance for PHLs on emerging NBS techniques and new diseases

IO-3. Improved competence and engagement of public health laboratory workforce

- Larger PHL workforce skilled in newborn screening
- Improved national NBS laboratory capability

IO-4. Enhanced practices, methods, technical capabilities, and infrastructure within the public health laboratory system

- PHLs proficient in NBS testing, including new testing technologies
- PHLs able to rapidly conduct and report newborn screening results
- PHLs able to quickly implement new screening tests and sustain NBS laboratory testing during emergencies

IO-6. Improved public health laboratory detection, surveillance, and response

- Improved coordination of newborn screening laboratory efforts in the newborn screening system.
- Better strategies to address emerging issues across the NBS system

IO-7. Improved quality and safety in public health laboratories

- High-quality and safe NBS laboratory practices in PHLs
- Efficient, interoperable PHL data exchange, transfer, and use for NBS

III. Funding Strategy

CDC funding strategy for this focus area is described in section iv, “Funding Strategy,” under Part II, A-2 (CDC Project Description; a. Approach) in this NOFO. Funds should be used for program resources which could include:



personnel, travel, supplies, equipment, contractual, and consultant support for proposed activities.

The funded recipient is expected to adhere to the requirements of the cooperative agreement. This may include:

- Identifying a designated person with overall responsibility for all activities as well as personnel responsible for each activity
- Participating in implementation, support, and monitoring efforts at least quarterly

Budgets should be submitted with sufficient level of detail so that the technical monitor, project officer, or the grants management officer can determine the necessity, reasonableness, and allocability of costs relative to the proposed grant activities, and their allowability pursuant to the applicable federal cost principles and requirements.

IV. Strategies and Activities

Activities under this focus area should be guided by strategies in the following categories: Policy, Partnership, and Communication (S2), Training and Capacity Building (S3), and Laboratory Quality, Safety, and Informatics for Public Health Testing Services, Surveillance and Response (S4).

S2. Policy, Partnership, and Communication

S2.2. Collaborate and build relationships among laboratory professionals and other stakeholders in public health, healthcare, and beyond

- Develop relationships and engage with partner organizations, including policymakers and advocacy groups.
- Develop partnerships to improve screening test performance, test result interpretation, quality assurance approaches, and training, and technology transfer.

S2.3. Facilitate information exchange and dissemination among practitioners and other stakeholders

- Communicate with PHLs, federal partners, and others about NBS policy, technology, regulation, and practice.
- Disseminate guidance to PHL leadership and staff.
- Disseminate best practices for both routine and emergency laboratory operations.

S2.4. Educate the public and other stakeholders about the role of PHLs

- Identify and address communication and information needs of parents and advocacy groups, including the use of plain writing/plain language in communications.
- Facilitate communication among PHLs, parents, health care providers, partners, and policy makers about NBS science, policy, technology, regulation, and practice.



S3. Training and Capacity Building

S3.1. Identify training and workforce development needs among laboratory professionals in diverse settings

- Conduct training and workforce development needs assessments to identify unique needs for NBS laboratories.
- Use needs assessment results to inform the development and delivery of training and workforce development resources for newborn screening staff and leadership.
- Apply best practices to all training and workforce development products, resources, and events (e.g., CDC Quality Training Standards and Laboratory Competencies).
- Monitor and evaluate effectiveness of training and workforce development products, resources, and events regularly and consistently.

S3.3. Facilitate the development and delivery of training and workforce development resources

- Use needs assessment results and guidance from CDC to develop and deliver training and workforce development opportunities for NBS laboratory staff, including those on laboratory methods and emerging issues in NBS.
- Collaborate with CDC to review training and workforce development products, resources, and events.
- Collaborate with CDC to establish fellow and host site eligibility requirements and performance expectations
- Provide guidance and technical assistance to fellows and host sites.
- Manage operations and funds to support fellows.
- Coordinate with the recipient Training and Workforce Development Program regarding cross-cutting and program-specific data generation to contribute to fellowship program monitoring and evaluation activities.

S4. Laboratory Quality, Safety, Preparedness, and Informatics for Public Health Testing Services, Surveillance, and Response.

S4.2. Identify and address systems to improve the practice of laboratory quality and safety in public health laboratories

- Convene committees, workgroups, or other meetings to develop and communicate guidance about NBS science and policy issues.

S4.3. Identify and address emerging methodological and process improvements in public health laboratories

- Develop guidance about policy, scientific, and management issues in NBS.

S4.4. Provide technical assistance to state and local public health laboratories to support improvements in public health outcomes

- Facilitate on-site assessment and guidance for PHLs integrating new technologies and implementing systems to modernize screening.



- Facilitate improvements in newborn screening laboratory information technology and management infrastructure.

S4.5. Identify and address gaps in laboratory infrastructure and capability to prepare and respond to public health threats.

- Distribute NBS filter paper from emergency stockpile to PHLs experiencing shortages.
- Monitor trends in NBS contingency planning.
- Collaborate with PHLs to develop state NBS contingency plans.

b. Evaluation and Performance Measurement

I. CDC Evaluation and Performance Measurement Strategy

The CDC Evaluation and Performance Measurement Strategy for this focus area uses the guidance from the overall CDC Evaluation and Performance Measurement Strategy described in this NOFO (Part II, A-2-b-i. CDC Evaluation and Performance Measurement Strategy), to address the following specific performance measures (including process measures and outcome measures) for this focus area.

Process measures for the strategies in this focus area may include:

Strategy and Activity	Process Measure
<p>S2.2. Collaborate and build relationships among laboratory professionals and other stakeholders in public health, healthcare, and beyond</p> <ul style="list-style-type: none"> • Develop relationships and engage with partner organizations, including policymakers and advocacy groups. • Develop partnerships to improve screening test performance, test result interpretation, quality assurance approaches, and training, and technology transfer. 	<ul style="list-style-type: none"> • Number and diversity of new and existing partner organizations engaged in and familiar with newborn screening laboratory issues. • Number of partners engaged in screening test performance, test result interpretation, quality assurance approaches, and training, and technology transfer.
<p>S2.3. Facilitate information exchange and dissemination among practitioners and other stakeholders</p> <ul style="list-style-type: none"> • Communicate with PHLs, federal partners, and others about NBS policy, technology, regulation, and practice. • Disseminate guidance to PHL leadership and staff. • Disseminate best practices for both routine and emergency laboratory operations. 	<ul style="list-style-type: none"> • Number of policy, science, and management guidance documents disseminated to newborn screening laboratories. • Number of educational resources and events for partners and stakeholders that increase awareness of newborn screening laboratory capabilities and issues. • Number of best practice documents for routine and emergency newborn screening laboratory operations disseminated to PHLs.



<p>S2.4. Educate the public and other stakeholders about the role of PHLs</p> <ul style="list-style-type: none">• Identify and address communication and information needs of parents and advocacy groups, including the use of plain writing/plain language in communications.• Facilitate communication among PHLs, parents, health care providers, partners, and policy makers about NBS science, policy, technology, regulation, and practice.	<ul style="list-style-type: none">• Number of educational resources, including those in plain language, disseminated to diverse stakeholders, such as parent and advocacy groups, regarding newborn screening laboratory issues.• Number and breadth of connections made among diverse stakeholders regarding newborn screening laboratory and system issues.
<p>S3.1. Identify training and workforce development needs among laboratory professionals in diverse settings</p> <ul style="list-style-type: none">• Conduct training and workforce development needs assessments to identify unique needs for NBS laboratories.• Use needs assessment results to inform the development and delivery of training and workforce development resources for newborn screening staff and leadership.• Apply best practices to all training and workforce development products, resources, and events (e.g., CDC Quality Training Standards and Laboratory Competencies).• Monitor and evaluate effectiveness of training and workforce development products, resources, and events regularly and consistently.	<ul style="list-style-type: none">• Number and type of training needs assessments that identify NBS laboratory needs.• Number of trainings and workforce development products that apply best practices.• Number of evaluations of effectiveness of training products, resources, and events for newborn screening laboratories.
<p>S3.3. Facilitate the development and delivery of training and workforce development resources</p> <ul style="list-style-type: none">• Use needs assessment results and guidance from CDC to develop and deliver training and workforce development opportunities for NBS laboratory staff, including those on laboratory methods and/or emerging issues in NBS.• Collaborate with CDC to review training and workforce development products, resources, and events.	<ul style="list-style-type: none">• Number and diversity of training opportunities for newborn screening laboratory staff, including laboratory methods and/or emerging issues.• Number of trainee and/or fellowships for newborn screening laboratories.



<ul style="list-style-type: none"> • Collaborate with CDC to establish fellow and host site eligibility requirements and performance expectations. • Provide guidance and technical assistance to fellows and host sites. • Manage operations and funds to support fellows. • Coordinate with the recipient Training and Workforce Development Program regarding cross-cutting and program-specific data generation to contribute to fellowship program monitoring and evaluation activities. 	
<p>S4.2. Identify and address systems to improve the practice of laboratory quality and safety in public health laboratories</p> <ul style="list-style-type: none"> • Convene committees, workgroups, or other meetings to develop and communicate guidance about NBS science and policy issues. 	<ul style="list-style-type: none"> • Number of committees, workgroups, and meetings to develop and communicate science and/or policy guidance for newborn screening laboratories.
<p>S4.3. Identify and address emerging methodological and process improvements in public health laboratories</p> <ul style="list-style-type: none"> • Develop guidance about policy, scientific, and management issues in NBS. 	<ul style="list-style-type: none"> • Number of guidance documents about policy, scientific, and management issues relevant to newborn screening laboratories developed.
<p>S4.4. Provide technical assistance to state and local public health laboratories to support improvements in public health outcomes</p> <ul style="list-style-type: none"> • Facilitate on-site assessment and guidance for PHLs integrating new technologies and implementing systems to modernize screening. • Facilitate improvements in newborn screening laboratory information technology and management infrastructure. 	<ul style="list-style-type: none"> • Number of on-site assessments of newborn screening laboratories to improve implementation of new technologies and operations. • Number of resources coalesced, developed, disseminated, and provided to assist newborn screening laboratories in implementing new technologies, approaches, or programs. • Number of resources coalesced, developed, disseminated, and/or provided to assist newborn screening laboratories with improvements in information technology or management infrastructure.
<p>S4.5. Identify and address gaps in laboratory infrastructure and capability to</p>	<ul style="list-style-type: none"> • Number of newborn screening laboratories that received and



<p>prepare and respond to public health threats.</p> <ul style="list-style-type: none"> • Distribute NBS filter paper from emergency stockpile to PHLs experiencing shortages. • Monitor trends in NBS contingency planning. • Collaborate with PHLs to develop state NBS contingency plans. 	<p>successfully used NBS filter paper from emergency stockpile during shortages.</p> <ul style="list-style-type: none"> • Number of resources coalesced, developed, disseminated, and/or provided to assist PHLs with continuous, high-quality newborn screening during an emergency. • Number of resources coalesced, developed, disseminated, and/or provided to assist PHLs with improving or establishing newborn screening contingency plans.
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Outcome measures for this focus area may include:

Outcome	Outcome Measure
<p>IO-1. Established communities of practice and other collaborative relationships among and between laboratories and other stakeholders</p>	<ul style="list-style-type: none"> • Number of PHLs participating in NBS laboratories and/or NBS system networks and communities of practice. • Number of NBS laboratories with active relationships with hospitals, follow-up coordinators, policy makers, communicator and other NBS system stakeholders. • Number and relevance of newborn screening topics that networks, collaborative relationships, and/or communities of practice address.
<p>IO-2. Established information sharing across public health laboratories and with other key stakeholders</p>	<ul style="list-style-type: none"> • Number of PHLs participating in training for existing and emerging NBS testing methods and data interpretation approaches, such as molecular detection, NGS, and bioinformatics. • Number of PHLs using or implementing guidance on new policies and methods related to emerging NBS techniques or new diseases. • Number of newborn screening system stakeholders using or implementing guidance.
<p>IO-3. Improved competence and engagement of public health laboratory workforce</p>	<ul style="list-style-type: none"> • Number of PHL staff able to conduct newborn screening for existing and emerging conditions. • Percent of PHLs with expanded newborn screening laboratory capability.



IO-4. Enhanced practices, methods, technical capabilities, and infrastructure within the public health laboratory system	<ul style="list-style-type: none">• Number of PHLs with expanded newborn screening laboratory capacity.• Percent of PHLs that implement new testing technologies.• Number of PHLs able to more rapidly and accurately detect and report newborn screening results.• Number of PHLs with established and exercised protocols to sustain newborn screening laboratory testing during emergencies.
IO-6. Improved public health laboratory detection, surveillance, and response	<ul style="list-style-type: none">• Number of newborn screening laboratory initiatives that incorporate newborn screening system stakeholders and other partner organizations.• Number of newborn screening laboratories implementing best practices to address emerging NBS issues.
IO-7. Improved quality and safety in public health laboratories	<ul style="list-style-type: none">• Percent increase in the number of PHLs conducting high-quality and safe NBS practices.• Percent increase in the number of newborn screening laboratories effectively utilizing PHL data exchange, transfer, and analytics for NBS.

II. Applicant Evaluation and Performance Measurement Plan

The recipient will be required to submit a detailed Evaluation and Performance Measurement plan within the first 6 months of award and work with CDC staff to ensure that the evaluation plan is feasible and consistent with proposed focus area activities, the intent of this NOFO, and CDC’s evaluation approach.

c. Collaborations

The recipient is expected to use or develop strategic partnerships that best suit their needs in order to maximize the use of resources and improve the sustainability of the NBS project activities.

With CDC funded programs

General guidance for collaborations with CDC funded programs is described in section a, “With other CDC programs and CDC-funded organizations,” under Part II, A-2-iii-1 (Collaborations) in this NOFO. The recipient is expected to collaborate with CDC’s Division of Laboratory Sciences, Newborn Screening and Molecular Biology Branch, and other relevant programs to establish program priorities and activities. The recipient may also consider collaborations with NBS laboratories previously or currently funded by CDC to share expertise, challenges, and successes related to the newborn screening system.



With organizations external to CDC

General guidance for collaborations with organizations external to CDC is described in section b, “With organizations not funded by CDC,” under Part II, A-2-iii-1 (Collaborations) in this NOFO. The recipient may also consider collaboration with NBS laboratories not currently funded by CDC to share expertise, challenges, and successes related to the NBS system. The recipient should establish appropriate collaboration with and support from NBS stakeholders to accomplish diverse activities. Potential collaborators include, but are not limited to: other federal or state agencies involved in newborn screening, national advisory committees, academic institutions, professional organizations, non-profit organizations, advocacy groups, parent support groups, community groups, public health officials, and health care providers.

d. Target populations

The primary target population of this project is PHL professionals supporting state and territorial newborn screening programs. The secondary target population is stakeholders in the newborn screening system, including parents, policy makers, care providers, and others.

e. Organizational Capacity

Refer to section c, “Organizational Capacity of Recipients to Implement the Approach” under Part II, A-2 (CDC Project Description) in this NOFO. The recipient should demonstrate a well-established and trusted role with newborn screening laboratories and among the newborn screening community as a whole. The recipient should also demonstrate broad expertise in the newborn screening field, experience executing the proposed activities, and existing staffing and infrastructure to support the work plan.

f. Work Plan

The recipient is required to provide a work plan for this focus area that provides both a high-level overview of the entire five-year period of performance and a detailed description of the first year of the award. The work plan should follow the general guidance provided in section d, “Work Plan” under Part II, A-2 (CDC Project Description) in this NOFO, and address the specific strategies, activities, outcomes, and performance measures of this focus area. After the award is made, the proposed work plan (including the evaluation and performance measurement plan) may be adjusted in collaboration with the CDC Technical Monitors to ensure integration of the strategies and activities and achievement of the period of performance outcomes.

g. CDC Program Support to Recipient

CDC’s Division of Laboratory Sciences and Newborn Screening and Molecular Biology Branch will provide technical expertise in support of project activities and will meet with the recipient’s newborn screening and genetics staff monthly (and on an ad hoc basis for emerging issues) via conference call to discuss project progress and future actions. In addition, CDC will participate in all relevant stakeholder and other meetings, either in-person or by teleconference.