

INITIAL Survey of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children's Public Health System Assessment

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The purpose of this survey is to inform the Secretary of Health and Human Services ~~Discretionary~~ Advisory Committee on Heritable Disorders in Newborns and Children (Committee) ~~(ACHDNC)~~ about states' ability to add newborn screening (NBS) for [condition x] ~~new conditions~~ using information gathered from most of the state and territorial NBS programs ~~states~~ in the U.S. Your input will provide valuable information and aid the deliberations of the Committee.

Please refer to the [condition x] screening factsheet to help you answer the following questions about the ability of your state or territory to add NBS screening for [condition x] into your NBS program. ~~You have received this survey on behalf of your state newborn screening program. If you are not the correct person to complete and return this form, please ensure that the correct person obtains it. Please consult with others, as needed, from your NBS program.~~ We expect that whoever leads the effort to respond to this survey will need to consult with others within your state, including laboratory and follow-up staff, medical professionals and specialists, prior to completing the survey. When unsure about a response, please provide your best estimate. If you were to answer every question, ~~we~~ as such, we are estimating ~~are estimating that~~ it will take each state an average of 10 ~~person~~ hours to complete this form.

1. ~~A. Does your state NBS screening panel currently include condition x NBS?~~

- ~~Yes (end survey)~~
- ~~No~~

1B. ~~Are you currently involved with any pilot evaluation activities, i.e., research or pre-live reporting results?~~

- ~~Yes: Please describe.~~
- ~~No~~

2.1. ~~Within the last three years, has your state: included... (Please check all that apply):~~

- ~~Included ~~G~~ [condition x] as part of the routine NBS panel? (end survey)~~
- ~~Planned, ~~conducted~~ implemented, or completed Condition x as any type of pilot study or pilot evaluation for [condition x]? (end survey)~~
- ~~Issued a mandate or state-level decision to start screening for [condition x]? (end survey)~~
- ~~None of the above (go to question 2.3)~~

3. ~~Has there been a state-level decision to start screening for condition x as part of NBS?~~

- ~~Yes (end survey)~~
- ~~No~~

4. ~~Which of the following provides NBS laboratory services for your state's NBS program? Please check all that apply.~~

- ~~Your own state's public health or NBS laboratory~~

- ~~o A contracted regional NBS laboratory or other not for profit laboratory~~
- ~~o A contracted commercial laboratory~~
- ~~o Other – please specify:~~
- ~~o None of the above~~

2. Which of the following entities provide NBS laboratory services for your state's NBS program? (check all that apply).

- ~~o Your own state's public health or NBS laboratory~~
- ~~o A state university laboratory for which there is an intra-state agency agreement~~
- ~~o A contracted regional NBS laboratory~~
- ~~o A contracted commercial laboratory~~
- ~~o Other – please specify:~~

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NBS programs consider many factors when deciding to add a condition to their NBS panel. The following question asks you to consider, in general, how much the following factors would be an issue in considering adding [condition x] to your NBS panel.

3. Please ~~categorize~~ indicate if the ~~funding~~ following implementation challenges factors related to NBS program activities for [condition x] in your state would present as a ~~Major~~ challenge, a ~~Minor~~ challenge, or would not be a challenge, given the current status of the NBS Program in your state.

~~Not a Challenge. Please see definitions below. *Not a Challenge using the following ratings:~~

~~Major Challenge = NBS program needs 3 or more years to resolve.~~

~~Minor Challenge = NBS program needs 1-3 years to resolve.~~

~~Not a Challenge = NBS program needs less than 1 year to resolve.~~

Activity Factor	Major Challenge	Minor Challenge	Not a Challenge	Comments
Availability of a validated screening test in your state Providing the screening test				
Ability to conduct S short-term follow-up for of abnormal out of range screening tests results , including tracking and follow-up testing				
Identifying Support to specialists in your state (or region) who can treat newborns and children with for [condition x]				
Treatment A availability of treatment Support to treatment for [condition x] in your state				
Ability to conduct L long-term follow-up for those with late-onset disease or who those identified as carriers (if applicable to [condition x])				
Increasing your NBS fee/ other administrative challenges				
Addressing administrative challenges (please specify in comments section)				

~~Major Challenge = NBS program needs 3 or more years to resolve.~~

~~Minor Challenge = NBS program needs 1-3 years to resolve.~~

~~Not a Challenge = NBS program needs less than 1 year to resolve.~~

5a. 4. Please describe any additional overarching challenges.

~~5. Which of the following best describes the type of screening approach or assay your program would choose for condition x:~~

- ~~Screening approach will detect carriers and we must plan for that incorporate follow-up of those cases into our algorithm~~
- ~~Screening approach will not detect carriers~~
- ~~Screening approach not yet determined~~

For questions 65-78 please assume that [condition x] has been authorized for addition to your state's panel and that funds for laboratory testing and follow-up have been made available.

~~a65a. Other than funding, certain factors related to condition x might make screening easier or more challenging in your state. Please let us know~~The following question considers the various resources needed (e.g. human resources, facilities, etc) by your NBS program in order ~~to be ready~~ to implement screening for [condition x]. ~~by evaluating the following resources~~the degree to which these factors impede or facilitate your ability to screen for condition x in your state. In order to respond to these questions, assume that condition x has been authorized for addition to your state's panel and that funds for both laboratory testing and follow-up are made available. If needed, please speak with your NBS laboratory to help assist with the answers.

~~If funding was made available, based on your state's current NBS infrastructure, to what extent do the factors below impede or facilitate the adoption of screening for condition x in your state?~~

~~5.a – Please complete the following table if you answered “your own state’s public health or NBS laboratory” on question #2. If your answer on question #2 was any of the other options, please skip to 5.b.~~

5.a Resources Needed FactorFactor	Do not have and cannot get within 1 year Do not have but could <u>can</u> get within 1 year	No Impact	Have but needs improvement	Have and need improvement <u>Cannot get within 1 year</u>	Comments
Screening approach for condition x method for [condition x]: [insert name screening method(s) here]					
A second-tier screening approach for [condition x] (if applicable)					
If you selected “your own state’s public health or NBS laboratory” for Question 2:-Quantity and type of [Laboratory equipment needed to screen specimens for [condition x] using flow injection MS/MS* (please describe equipment needed in comments section)]					
Laboratory equipment needed to screen specimens for [condition x] using digital fluorometry*					
Laboratory technical expertise to screen for condition x *					
Laboratory technical expertise to screen for [condition x]					
Sufficient nNumber of technical staff within your laboratory to screen for [condition x].*					

<u>If you selected "A contracted regional NBS laboratory or other not-for-profit laboratory" or "A contracted commercial laboratory" at Question 2: Availability of the screening test in your contracted laboratory--</u>						
<u>Onsite genotyping as part of a second-tier test</u>						
<u>LIMS capacity and instrumentation interface</u>						
<u>Sufficient number of NBS staff to notify and track NBS results</u>						
<u>Access to appropriate diagnostic services after an <u>abnormal or out of range presumptive positive from a screening result is reported</u> (e.g., diagnostic testing, clinical evaluations)</u>						
<u>Genetic counselors, or other staff with the necessary expertise, to cover the expected caseload, including reporting carrier status (if applicable)</u>						
<u>Availability of <u>Specialists to cover expected [condition x] caseloads</u></u>						
<u>Treatment centers for expected [condition x] case-load</u> Availability of treatment for those diagnosed through NBS						
<u>Follow-up protocols for [condition x] cases and carriers</u>						

*Please respond to these factors if you selected "Your own state's public health or NBS laboratory" at question 4.

NOTE SKIP PATTERN (respondents will fill out either 5.a or 5.b., but not both)

~ Please respond to this factor if you selected "A contracted regional NBS laboratory or other not-for-profit laboratory" or "A contracted commercial laboratory" at question 4.

5.b. Please complete the following table if you answered "a state university laboratory for which there is an intra-state agency agreement", "a contracted regional NBS laboratory", "a contracted commercial laboratory", or "other - please specify" on question #2.

<u>5.b Resources</u>	<u>Have Already</u>	<u>Do not have but can get within 1 year</u>	<u>No Impact</u>	<u>Have but needs improvement</u>	<u>Cannot get within 1 year</u>	<u>Comments</u>
<u>Availability of the screening test in the state university laboratory for which there is an intra-state agency agreement, or contracted regional laboratory, or commercial laboratory.</u>						

<u>Availability of a second-tier screening approach for [condition x] (if applicable)</u>						
<u>LIMS capacity and instrumentation interface</u>						
<u>Sufficient number of NBS staff to notify and track NBS results</u>						
<u>Access to appropriate diagnostic services after an abnormal or out of range screening result is reported (e.g., diagnostic testing, clinical evaluations)</u>						
<u>Genetic counselors, or other staff with the necessary expertise, to cover the expected caseload, including reporting carrier status (if applicable)</u>						
<u>Specialists to cover expected [condition x] caseload</u>						
<u>Treatment centers for expected [condition x] caseload</u>						
<u>Follow-up protocols for [condition x] cases and carriers</u>						

~~67bb6. Other than funding, certain factors related to condition x might make screening easier or more challenging in your state. Please let us know indicate the degree* to which these factors impede or facilitate your ability to adopt screeningscreen for [condition x] in your state. In order to respond to these questions, assume that condition x has been authorized for addition to your state's panel and that funds for both laboratory testing and follow up are made available. Please refer to the webinar recording that provides background on condition x. If needed, please consult with laboratory and follow up staff, medical professionals and specialists, prior to completing the survey.~~

~~If funding was made available, to what extent do the factors below impede or facilitate the adoption of screening for condition x in your state?~~

Factor	Will hinder implementation Major Barrier	May hinder implementation Minor Barrier	No Impact	May aid in implementation Minor Facilitator	Will aid in implementation Major Facilitator	Not Applicable	Comments
Predicted run time to screen for [condition x] as it relates to other workload							
Extent to which the screening test for [condition x] can be multiplexed with screening for other conditions							
Other ongoing NBS program activities (e.g., addition of other conditions, other quality improvements)							
Extent to which screening protocol for condition x has been demonstrated in other NBS programs							
Estimated Cost per specimen to conduct screening (personnel, equipment, reagents)							
Estimated Cost of treatment for newborns diagnosed with [condition x]NBS							

Expected clinical outcomes of newborns identified by screening							
Expected cost-benefit of screening in your state							
Advocacy for screening for this [condition x]							
Other non-NBS public health priorities within your state							

**Major barrier- Will prevent testing from being implemented effectively and/or timely.*

**Minor barrier- May compromise testing so it is not performed effectively and/or timely.*

**Minor facilitator- May allow testing to be done effectively and/or timely.*

**Major facilitator- Will allow testing to be done effectively and/or timely.*

~~6b946b47~~ Please describe any additional factors that impede or facilitate adoption of screening for [condition x] in your state.

~~96e8a~~ What ~~is-are~~ the most significant barrier(s) to screening NBS for [condition x] in your state?

~~406d8b~~ What would most facilitate screening for NBS-[condition x] in your state?

~~41-~~

9. If condition x was added to the RUSP tomorrow, about how long would it take in total to initiate screening for condition x in your state (consider the total amount of time from initial interest in screening for condition x to screening every newborn born in your state for condition x. Please estimate the time it would take your NBS program to initiate screening for [condition x] in your state (i.e. get authority and funds to screen for disorder/condition x, go through administrative processes, meet with your state NBS committees and complete all activities need to implementation activities and commence screening for all newborns in your state in order to begin screening (entire process))?

- 12 months or less
- 13 to 24 months
- 25 to 36 months
- 37 to 48 months
- More than 48 months

10. The question above related to the overall timeline. The table below is intended to ask identify about how long would it would take to achieve specific activities on within the overarching timeline. We recognize some of the activities happen in tandem and some cannot begin until a previous activity has been completed. Please estimate the total time needed, in general, for each individual of the activities listed below within your NBS program, the following assuming that condition x was added to your state NBS panel and funds were allocated today, with your current NBS program and laboratory infrastructure?—If needed, please consult with laboratory and follow-up staff, medical professionals and specialists, prior to completing the survey.

Activity	One-year-or less 12 months or less	Years 13 – 24 months	25 – 36 months 2-3 years	37 to 48 months	> 3 years 48 months	Not Applicable	Comment
Obtain authorization to screen for condition x in your state?							
Get Once you received authorization to screen, about how long would it take to have Availability of funds available to implement screening for condition x							
Meet with Advisory committees and other stakeholders Once funds are available, about how long would it take to complete start-up implementation activities (e.g., laboratory validation, reporting systems, and training for follow up) in order to be ready to begin screening for condition x?							
Conduct a pilot/preliminary screening							
Obtain and procure equipment for screening for [condition x]							
Hire necessary laboratory and follow-up staff							
Consult with medical staff and specialists							
Select, develop, and validate the screening test within your							

laboratory <u>IF you are NOT multiplexing</u>							
<u>—Select, develop, and validate the screening test within your laboratory IF you ARE multiplexing</u>							
<u>—Develop a screening algorithm, and follow-up protocols, and train follow up staff</u>							
<u>—Set up reporting and results systems for added condition (e.g., LIMS)</u>							
<u>—Collaborate with specialists and clinicians in the community to determine which diagnostic tests will be recommended upon identification of an out of range NBS result</u>							
<u>—Add the screening test to the existing outside laboratory contract)~</u>							
<u>Conduct an internal validation study for [condition x]</u>							
<u>—Pilot test the screening process within your state, after validation has taken place</u>							
<u>— Implement statewide screening for all newborns, including full reporting and follow-up of abnormal screens after validation and pilot testing</u>							
<u>Entire process from obtaining equipment to implementing statewide screening (assuming that some activities may occur simultaneously)</u>							

*** Please respond to this activity if you selected "A contracted regional NBS laboratory or other not-for-profit laboratory" or "A contracted commercial laboratory" at question 4.*

11. (If applicable to [condition x]) Which of the following best describes the type of screening approach or assay your program would choose for [condition x]:

- Screening approach will detect carriers and we must incorporate follow-up of those cases into our algorithm
- Screening approach will not detect carriers
- Screening approach not yet determined

1312. Are there any special considerations regarding [condition x] that need to be taken into account when assessing the impact on the public health system? (e.g. will it be possible to identify carriers for condition x and how will your state approach carrier status)? (e.g. variants of unknown significance, pseudodeficiencies, age of onset, access to specialists, access to treatment, cost of treatment, etc) Please describe:

1440. 13. Please share any additional information regarding implementation of NBS for [condition x].

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514. Please provide information about the respondent:

Name:
Phone number:
Email address:
Job title:

16 How long have you had this position?

- ~~< 1 year~~
- ~~1-3 years~~
- ~~4-6 years~~
- ~~7-9~~
- ~~More than 10 years~~

157. Who did you consult with to answer these questions? *Please check all that apply.*

- State NBS laboratory experts
- Other NBS program staff
- State NBS advisory board
- State Title V Director
- [Condition x] Specialists
- Primary care providers
- Advocates within your state for [condition x] screening
- Others- please specify: _____
- None of the above

Thank you for completing the survey!