

## INITIAL Survey of the 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 Advisory Committee on Heritable Disorders in Newborns and Children (Committee) about states' ability to add newborn screening (NBS) for [condition x] using information gathered from most of the state and territorial NBS programs 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 screening for [condition x] to your NBS program. Please consult with others, as needed, including laboratory and follow-up staff, medical professionals and specialists, to complete the survey. When unsure about a response, please provide your best estimate. If you were to answer every question, we estimate it will take an average of 10 hours to complete this form.

1. Within the last three years, has your state: *(check all that apply)*
  - Included [condition x] as part of the routine NBS panel? *(end survey)*
  - Planned, implemented, or completed 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)*
  
2. Which of the following entities provide NBS laboratory services for your state's NBS program? *(check all that apply)*
  - Your own state's public health or NBS laboratory
  - A state university laboratory for which there is an intra-state agency agreement
  - A contracted regional NBS laboratory
  - A contracted commercial laboratory
  - Other – please specify: \_\_\_\_\_

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 when considering adding [condition x] to your NBS panel.

3. Please indicate if the following implementation factors for [condition x] would present a major challenge, a minor challenge, or would not be a challenge, given the current status of the NBS Program in your state.

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

4. Please describe any additional overarching challenges. \_\_\_\_\_  
 \_\_\_\_\_

For questions 5-7 please assume that [condition x] has been authorized for addition to your state's panel and funds for laboratory testing and follow-up have been made available.

5. The following question considers the various resources needed (e.g. human resources, facilities, etc) by your NBS program in order to implement screening for [condition x].

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  | Have Already | Do not have but can get within 1 year | Cannot get within 1 year | Comments |
|--|--------------|---------------------------------------|--------------------------|----------|
| Screening method for [condition x]:<br>[insert screening method(s) here]     |              |                                       |                          |          |
| A second-tier screening approach for [condition x] (if applicable)           |              |                                       |                          |          |
| Quantity and type of laboratory equipment needed to screen for [condition x] |              |                                       |                          |          |
| Laboratory technical expertise to screen for [condition x]                   |              |                                       |                          |          |
| Sufficient number of technical staff to screen for [condition x]             |              |                                       |                          |          |
| LIMS capacity and instrumentation interface                                  |              |                                       |                          |          |

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

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

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.

| 5.b. Resources Needed   | Have Already | Do not have but can get within 1 year | Cannot get within 1 year | Comments |
|---|--------------|---------------------------------------|--------------------------|----------|
| 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 |              |                                       |                          |          |
| Availability of a second-tier screening approach for [condition x] (if applicable)  |              |                                       |                          |          |
| LIMS capacity and instrumentation interface   |              |                                       |                          |          |
| Sufficient number of NBS staff to notify and track NBS results  |              |                                       |                          |          |
| Access to appropriate diagnostic services after an abnormal or out of range screening result is reported (e.g., diagnostic testing, clinical evaluations)                             |              |                                       |                          |          |
| Genetic counselors, or other staff with the necessary expertise, to cover the   |              |                                       |                          |          |

|   |  |  |  |  |
|---|--|--|--|--|
| expected caseload, including reporting carrier status (if applicable) |  |  |  |  |
| Specialists to cover expected [condition x] caseload                  |  |  |  |  |
| Treatment centers for expected [condition x] caseload                 |  |  |  |  |
| Follow-up protocols for [condition x] cases and carriers              |  |  |  |  |

6. Please indicate the degree\* to which these factors impede or facilitate your ability to adopt screening for [condition x] in your state.

| Factor  | Major Barrier | Minor Barrier | Minor Facilitator | Major Facilitator | Not Applicable |
|---|---------------|---------------|-------------------|-------------------|----------------|
| 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)       |               |               |                   |                   |                |
| Estimated cost per specimen to conduct screening (personnel, equipment, reagents)                           |               |               |                   |                   |                |
| Estimated cost of treatment for newborns diagnosed with [condition x]                                       |               |               |                   |                   |                |
| 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.

7. Please describe any additional factors that impede or facilitate adoption of screening for [condition x] in your state. \_\_\_\_\_

8a. What are the most significant barrier(s) to screening for [condition x] in your state?

8b. What would most facilitate screening for [condition x] in your state?

9. 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 [condition x], go through administrative processes, meet with your state NBS committees and complete all activities needed to implement and commence screening for all newborns in your state)?

- 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. 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 activity listed below within your NBS program. If needed, please consult with laboratory and follow-up staff, medical professionals and specialists, prior to completing the survey.

| Activity   | 12 months or less | 13 – 24 months | 25 – 36 months | 37 to 48 months | > 48 months | Not Applicable | Comment |
|--|-------------------|----------------|----------------|-----------------|-------------|----------------|---------|
| Obtain authorization to screen for [condition x]               |                   |                |                |                 |             |                |         |
| Availability of funds to implement screening for [condition x] |                   |                |                |                 |             |                |         |
| Meet with Advisory committees and other stakeholders           |                   |                |                |                 |             |                |         |
| Obtain and procure equipment for screening for [condition x]   |                   |                |                |                 |             |                |         |
| Hire necessary laboratory and follow-up staff                  |                   |                |                |                 |             |                |         |

|  |  |  |  |  |  |  |  |
|--|--|--|--|--|--|--|--|
| Select, develop, and validate the screening test within your laboratory IF you are NOT multiplexing  |  |  |  |  |  |  |  |
| Select, develop, and validate the screening test within your laboratory IF you ARE multiplexing  |  |  |  |  |  |  |  |
| Develop a screening algorithm, follow-up protocols, and train follow up staff  |  |  |  |  |  |  |  |
| Set up reporting and results systems for added condition (e.g., LIMS)  |  |  |  |  |  |  |  |
| 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 |  |  |  |  |  |  |  |
| Add the screening test to the existing outside laboratory contract   |  |  |  |  |  |  |  |
| Conduct an internal validation study for [condition x]   |  |  |  |  |  |  |  |
| Pilot test the screening process within your state, after validation has taken place   |  |  |  |  |  |  |  |
| Implement statewide screening for all newborns, including full reporting and follow-up of abnormal screens after validation and pilot testing                          |  |  |  |  |  |  |  |

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

12. 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. variants of unknown significance, pseudodeficiencies, age of onset, access to specialists, access to treatment, cost of treatment, etc) Please describe:

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

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14. Please provide information about the respondent:

- Name:
- Phone number:
- Email address:
- Job title:

15. 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!