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INITIAL Survey of the Advisory Committee on Heritable Disorders in Newborns and Children's Public Health System Assessment - Duchenne Muscular Dystrophy

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 Duchenne Muscular Dystrophy 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 Duchenne Muscular Dystrophy screening factsheet to help you answer the following questions about the ability of your state or territory to add screening for Duchenne Muscular Dystrophy 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. Has your state: (*check all that apply)*
   - o Included Duchenne Muscular Dystrophy as part of the routine NBS panel? (*end survey*)
   - o Involved in a pilot study in any way for Duchenne Muscular Dystrophy (Elaborate please)? (*go to question 2*)
   - o Issued a mandate or state-level decision to start screening for Duchenne Muscular Dystrophy? *(end survey)*
   - o None of the above (*go to question 2*)

2. Which of the following entities provide the majority of the NBS laboratory services for your state's NBS program? (multiple choice*)*
   - 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 (e.g., another state laboratory)
   - o A contracted commercial laboratory
   - o 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 Duchenne Muscular Dystrophy to your NBS panel.

3. Please indicate if the following implementation factors for Duchenne Muscular Dystrophy would present a major challenge, a minor challenge, or would not be a challenge, given the status of the NBS Program in your state.

Factor	Major Challenge	Minor Challenge	Not a Challenge
Availability of a first-tier validated screening test (in-house or contracted)			
Availability of second-tier validated screening test (in-house or contracted)			
Availability of staff to report and track infants with out-of- range results through to diagnosis or resolution			
Identifying specialists in your state (or region) who are confident in the diagnostic evaluation of Duchenne Muscular Dystrophy			
Availability of timely treatment for infants born in your state with Duchenne Muscular Dystrophy (e.g. in-state treatment centers or funds for travel and out of state treatment)			
Increasing your NBS fee			
Addressing administrative challenges (e.g., obtain authorization); please specify in comments section			

4. Please describe any additional overarching challenges. _____
_____

For questions 5-7 please assume that Duchenne Muscular Dystrophy 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 Duchenne Muscular Dystrophy.

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
First-tier screening method for Duchenne Muscular Dystrophy			
Second-tier molecular screening for Duchenne Muscular Dystrophy			
Quantity and type of laboratory equipment needed to screen for Duchenne Muscular Dystrophy			
Laboratory technical expertise to screen for Duchenne Muscular Dystrophy			
Sufficient number of technical staff to screen for Duchenne Muscular Dystrophy			
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			
Specialists to cover expected Duchenne Muscular Dystrophy caseload			
Treatment centers for expected Duchenne Muscular Dystrophy caseload			
Follow-up protocols for Duchenne Muscular Dystrophy			

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
Availability of the first-tier 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 the second-tier molecular screening at a contracted laboratory			
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			
Specialists to cover expected Duchenne Muscular Dystrophy caseload			
Treatment centers for expected Duchenne Muscular Dystrophy caseload			
Follow-up protocols for Duchenne Muscular Dystrophy			

6. Please indicate the degree to which these factors impede or facilitate your ability to adopt screening for Duchenne Muscular Dystrophy in your state.

Factor	Major Barrier	Minor Barrier	Minor Facilitator	Major Facilitator	Not Applicable
Predicted run time to screen for Duchenne Muscular Dystrophy as it relates to other workload					
Extent to which the screening test for Duchenne Muscular Dystrophy can be multiplexed with screening for other disorders					
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 Duchenne Muscular Dystrophy					
Expected clinical outcomes of newborns identified by screening					
Expected cost-benefit of screening in your state					
Advocacy for screening for this Duchenne Muscular Dystrophy					
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 Duchenne Muscular Dystrophy in your state.

_____

8a. What are the most significant barrier(s) to screening for Duchenne Muscular Dystrophy in your state?

_____

8b. What would most facilitate screening for Duchenne Muscular Dystrophy in your state?

_____

9. Please estimate the time it would take your NBS program to initiate screening for Duchenne Muscular Dystrophy in your state (i.e. get authority and funds to screen for Duchenne Muscular Dystrophy, 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)?

- o 12 months or less
- o 13 to 24 months
- o 25 to 36 months
- o 37 to 48 months
- o 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.

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 10.b.

10a.

Activity	12 months or less	13 – 24 months	25 – 36 months	37 to 48 months	> 48 months	Not Applicable
Obtain authorization to screen for Duchenne Muscular Dystrophy						
Availability of funds to implement screening for Duchenne Muscular Dystrophy						
Meet with Advisory Committees and other Stakeholders						
Obtain and procure equipment for screening for Duchenne Muscular Dystrophy						
Hire necessary laboratory and follow-up staff						
Select, develop, and validate the screening test within your laboratory						
Develop follow-up protocols and train follow up staff						
Set up reporting and results systems for Duchenne Muscular Dystrophy (e.g., LIMS)						
Collaborate with specialists and clinicians in the community to determine the approach for the identification of an out-of-range NBS result						
Conduct an internal validation study for Duchenne Muscular Dystrophy						
Pilot test the screening process within your state, after validation has taken place						
Implement statewide screening once validation and pilot testing is complete						

SKIP PATTERN (respondents fill out either 10.a.or 10.b., but not both)

10b.

Activity	12 months or less	13 – 24 months	25 – 36 months	37 to 48 months	> 48 months	Not Applicable
Obtain authorization to screen for Duchenne Muscular Dystrophy						
Availability of funds to implement screening for Duchenne Muscular Dystrophy						
Meet with Advisory Committees and other Stakeholders						
Develop follow-up protocols and train follow up staff						
Set up reporting and results systems for Duchenne Muscular Dystrophy (e.g., LIMS)						
Collaborate with specialists and clinicians in the community to determine the approach for						

identification of an out-of-range NBS result						
Add the screening test to the existing outside laboratory contract						
Implement statewide screening once validation and pilot testing is complete						

11. a. Which of the following best describes the screening approach your program would most likely choose for Duchenne Muscular Dystrophy:

- First-tier CK-MM only (jump to 12)
- First-tier CK-MM, followed by repeat CK-MM for samples above cutoff, followed by third-tier molecular testing (jump to 11b)
- First-tier CK-MM activity followed by second-tier molecular testing for those above cutoff (jump to 11b)
- Other; specify _____ (jump to 12)

11. b. What is your plan for second/third-tier molecular testing? (all go to 12)

- In-house
- Contract to outside laboratory
- Unsure

12. Are there any special considerations regarding Duchenne Muscular Dystrophy that need to be taken into account when assessing the impact on the public health system (e.g. variants of unknown significance, age of onset, access to specialists, access to treatment, cost of treatment, etc.)? Please describe:

_____

13. Please share any additional information regarding implementation of NBS for Duchenne Muscular Dystrophy.

_____

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
- Duchenne Muscular Dystrophy 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!