Commonwealth of Massachusetts

Executive Office of Health and Human Services

Department of Public Health

Bureau of Infectious Disease and Laboratory Sciences

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Diagram

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Newborn Screening Advisory Committee Meeting

Monday, November 4, 2024: 4:00 – 6:00 PM.

Held: Virtually via ZOOM

**Minutes Approved**: 1/8/25

**Members in Attendance:** Karen McAlmon, Nicolas Epie, Catherine (Katie) Brown (DPH), Geoffrey Binney, Anne Comeau, Henry Dorkin, Mary-Alice Abbott, Monica Liao Chang, Amy Sobota, Dallas Reed, Chloe Schwartz, Feria Ladha, Inderneel Sahai, Yvonne Sheldon, Richard Parad.

**Department of Public Health and New England Newborn Screening Program Staff in Attendance:** H. Dawn Fukuda, Mahsa Yazdy, James Ballin, Roger Eaton, Thera Meehan

**Guests and Speakers:** Kylee Noga, Jennifer Yeaple, Elisa Seeger, Miranda McAuliffe, Christie Higuera, Jamie Hale, Catherine Brown (MGH), Susheela Jayaraman, Emilia Wilburn, Kate Segal, Jenn McNary.

**Determination of Quorum:** Confirmed, therefore committee was able to begin meeting.

**Welcome and Introductions:**

Dr. McAlmon welcomed all to the meeting and introductions were completed. There were guests in attendance to participate in the listening session at the end of topic discussions. There were three guest speakers.

**Approval of Minutes**

The minutes from the prior meeting held on November 8, 2023, were approved. Vote was Yes. Meeting Minutes are posted on the NBS Web Page.

**Overview of Pilot Screen Program, Dr. Anne Comeau, Deputy Director, New England Newborn Screening Program**

Dr. Comeau gave a brief overview of the histories of the conditions that have been included on the Massachusetts Newborn Screening Program pilot panels. Her slide presentation is attached.

**2025 Goals**

Dr. McAlmon reviewed the objectives for Massachusetts NBSAC for 2025.

* Meet regularly to determine status of pilot conditions (move to mandate, continue as pilot, or discontinue)
* Meet regularly and advise on timing of supportive actions
* Examine or re-examine conditions not on the Massachusetts panel that are on the RUSP.

**MPSII Brief Review and Update: Dr. Inderneel Sahai**

Dr. Sahai gave a brief review of MPSII and answered any questions. It was moved and voted to add MPS II to the pilot screening program.

MPSII Vote:

Yes. MPSII will be added to the pilot screens.

**NBSAC Website Development: Ms. Jennifer Yeaple**

Website for NBSAC is in development. Ms. Yeaple shared a mock-up of the proposed NBSAC website where all committee information will be posted. All NBSAC member names and affiliations will be included on the website. Information about past and future meetings, meeting materials, and meeting links will be posted on the website. There will be individual sections and/or links, as appropriate. The website will serve as a repository for information and resources for members, interested parties and guests. DPH is in the process of having the website completed and functional as soon as possible.

**New Legislation requiring DMD screening be added to the mandatory panel**

HB4999 An Act promoting access to midwifery care and out-of-hospital birth options

SECTION 18. Section 110A of said chapter 111, as appearing in the 2022 Official Edition, is hereby amended by striking out the first paragraph and inserting in place thereof the following paragraph:

* *The physician attending a newborn child shall cause said child to be subjected to tests for phenylketonuria, cretinism,* ***Duchenne muscular dystrophy*** *and such other specifically treatable genetic or biochemical disorders or treatable infectious diseases which may be determined by testing as specified by the commissioner. The commissioner shall convene an advisory committee on newborn screening to assist the commissioner in determining which tests are necessary; provided, however, that said advisory committee shall convene not less than twice per year.*

NENSP is in the process of operationalizing all procedures needed to institute this screening. Per legislation, the screening must start within 18 months of the passage of the bill.

**Review draft of Newborn Screening Submission Form to Suggest Conditions/Disorders for Addition to the Newborn Screening Panel: Dr. Karen McAlmon**

* The form will be used for DPH to receive recommendations from interest holders regarding conditions and disorders that should be assessed by the advisory committee for inclusion on the Massachusetts newborn screening panel. Guiding principles are included in the form. The full committee will receive submissions after preliminary review by the (yet to be established) NBS Condition Nomination Work Group.
* After presentation of the draft form, the following discussion with suggestions ensued:
  + Add version number and date to ensure appropriate tracking and current form use
  + State that form should only be submitted if all requirements are met
  + Need to establish a work group to initially screen nominations then all submissions should be shared with the full NBSAC
  + Full NBSAC will review conditions/disorders recommended by the work group and will have the discretion to add for review any conditions not specifically recommended
  + Timing for response to a request will vary depending on when it was submitted, when committee meetings occur and the need for expert consultation to the NBSAC. Expectations for turn around time should be discussed further. It is possible it could be a minimum of 6-9 months between submission and disposition.
  + Three to five committee volunteers will be needed to sit on the work group.

**Process for ongoing review of Pilot Screens:**

Dr. McAlmon invited discussion about what our process should be for examining pilots and making recommendations to (1) move those conditions to the mandated screening panel, (2) keep those conditions in pilot for further examination, or (3) discontinue screening for the condition. Dr. Comeau offered the following points of clarification and recommendations:

* Modifications to the conditions included in pilot requires a corresponding change to the educational brochure and redistribution to hospitals. Each time a pilot condition is added or removed, NENSP will update and distribute the brochure.
* An annual review process enables NENSP to correspondingly update program materials annually.
* Removing conditions from pilot or moving conditions to the mandated panel requires a longer amount of time for required approval. This process may be streamlined if the Committee reviews a group of disorders, rather than individual conditions.

Members discussed what this committee might need to move forward with creating a process to review conditions in pilot. It will be important to ensure that recommendations for moving conditions from pilot to mandate include a mechanism to confirm the criteria for mandated screens are met. It was agreed that committee will meet once a year to review pilots and make decisions regarding their future status.

**Possible future plans for Legislative Briefing about NBSAC:**

Recommendation that the NBSAC present a Legislative Briefing about the NBS program. If there are members interested in working on this for future, please inform Dr. McAlmon.

**Possible Lived Experience Testimonials during Disorder Presentations:**

A question was posed to members about the inclusion of lived experience testimonials during presentations on specific disorders/conditions under consideration for newborn screening. The Committee agreed it is important to continue testimonials for each disorder/condition being considered.

**Request for Nomination of Potential New Members:**

There is currently a need for additional representation on the NBSAC. Recommendations include for one or two parent members, an endocrinologist, and a birth hospital representative to join the committee. If you have recommendations, please email those to [Tresa.Glover-Smith@mass.gov](mailto:Tresa.Glover-Smith@mass.gov). Recommendation packets should include:

1) A brief cover letter from the nominator, recommending the individual and describing their interest/expertise in newborn screening and why they would be a strong addition to the committee.

2) A statement of interest from the nominee

3) Resume/CV from the nominee.

**Listening Session:**

**Elisa Seeger, ALD Alliance**

Before I read my prepared comments - I would like to comment on some of the discussion here today:

1. It is not for this committee to relitigate the treatment of any of these conditions which have already been established by the FDA.
2. Why are we comparing updating a brochure to a child’s life? We should not put importance on the cost of updating a brochure as a significant factor in deciding whether to add a condition population-wide.
3. Travel - why is the committee arguing over whether to tell a family about their baby's condition depending on whether they can travel for care? Once again this is not the job or responsibility of this committee; the bigger tragedy is not giving these families the information they need to save their child.

*“Thank you for giving me the time to speak today!  My name is Elisa Seeger, and I am the founder of the ALD Alliance. I started the foundation in late 2012 after losing my son, Aidan to ALD. New York became the first state to start testing every baby for ALD with the passing of Aidan’s Law in 2013. We have advocated throughout the country and am proud to say we are now testing for ALD in 46 states. Our main focus is newborn screening and the reason I wanted to speak today is we are concerned with the pilot that has been ongoing for close to 6 years for MPS1, Pompe, ALD and SMA.*

*While we understand there is a process, after this amount of time we are aware that newborns have been picked up for all 4 conditions and treated as well.*

*Although the Massachusetts newborn screening program has quoted relatively high opt in rates – we are still concerned with the babies that are being missed as this is still currently not population wide.  There are many parents I have spoken to that don’t remember opting in and one mom in particular who is very lucky she did as her son was diagnosed with SMA. Her son could be in a very different situation today if she did not check that box – something she has no recollection of doing.*

*After reaching out to the Massachusetts newborn screening program several times over the last few years, we wrote a letter in August which addresses the 12 requirements for full population screening in Massachusetts, as well as having parent testimonials and clinicians from Massachusetts General and Boston Children’s Hospital as well as institutions across the country in support of population wide testing.  Although all 12 requirements are fulfilled – the pilot is still ongoing.*

*I want to bring awareness to the Massachusetts Newborn Screening Program that we cannot afford to miss one child because a parent did not check a box. The gravity of this will lead to permanent disability or death for that child – something that we could easily stop through population wide screening.*

**Miranda McAuliffe, ALD Alliance**

*Good evening and thank you for the opportunity to address the committee. My name is Miranda McAuliffe. I’m the mother of an 8-year-old son who was diagnosed with ALD in New York.*

*Today I implore the committee to move ALD, SMA, MPS I and Pompe out of an almost six-year pilot program and into full population screening.  While I am educated only about ALD, due to my lived experience with the disease and five years of newborn screening advocacy, it is important to me that all babies be given access to medical monitoring and approved treatments, so that they may grow up to be healthy children.*

*These four conditions were already added to the federal Recommended Uniform Screening Panel years ago - and for longer than you have kept them in pilot.  There is a valid assay to screen at birth and treatment options that are successful if delivered in a timely manner.*

*Please share - why are these four conditions less worthy compared to the others on your panel?  I was told by  Dr. Comeau one year ago that ALD ‘suffers from the fact that it takes usually a minimum of two or three years from the screen to confirm a diagnosis of congenital cerebral ALD that would require transplantation and then time after that in order to determine the incremental benefit of an earlier diagnosis relative to a later diagnosis.’  The benefits have already been determined.  By subject matter experts, the federal RUSP committee, and 46 other states.  Timely treatment for cerebral ALD halts lesions, preserves function, and saves lives.  Why is this state continuing to spend money on a pilot for answers that we already have?*

*I’m also extremely concerned that adrenal insufficiency is not being addressed, as 80% of ALD boys will present with it.  It can be life threatening and is very treatable with steroids that cost pennies but is unfortunately often invisible in the newborn stage.  Why is adrenal insufficiency less important to this committee than cerebral ALD?*

*Why is there not more information being shared about the 28 ALD newborns identified in your state?  Your state is unique in that you have one of the best - if not the best - center of excellence for ALD monitoring and treatment.  This should make follow up reporting a much easier task compared to what other states have to go through - many of which have families that are traveling to this state for monitoring and treatment.*

**Christie Higuera, Massachusetts General Hospital**

*My name is Christie Higuera, and I’m the Program Director at the Center for Rare Neurological Diseases at Massachusetts General Hospital. More importantly, I’m the mother of a nearly 4-year-old boy diagnosed with X-Linked Adrenoleukodystrophy (ALD) in 2020, thanks to Massachusetts’ pilot newborn screening program. At the time, I agreed to the screening without much thought, but that simple decision turned out to be life changing. We caught my son’s condition before it could rob him of his potential.*

*Since his diagnosis, we’ve monitored him closely with frequent blood work and brain MRIs. We’ve been lucky that treatments like steroids, bone marrow transplant, and an FDA approved gene therapy are available here in Massachusetts. Twenty-eight boys, including my son, have been diagnosed with ALD through this program. Two of these boys developed early cerebral ALD but were saved by gene therapy. Without early screening and treatment, cerebral ALD leads to total loss of brain function and eventually a vegetative state. The condition becomes fatal, usually within 2-5 years after the onset of symptoms, due to the widespread damage to the brain.*

*This is why early detection matters so much. It can mean the difference between life and death. Unfortunately, this screening is still in a pilot phase, and 10% of newborns aren’t screened, that’s 10% too many. With 46 other states already testing for ALD, it’s time for Massachusetts to make this screening standard.*

*My son developed adrenal insufficiency, a life-threatening complication of ALD, but thanks to his early diagnosis, we were able to act before it caused irreversible damage. Early detection doesn’t just save lives, it transforms them. Every family deserves the chance to watch their children thrive. When we share knowledge, we give hope, and that hope is power.*

Dr. McAlmon thanked the guests for sharing their testimonials.

**Questions:**

Dr. Nicolas Epie, Director, State Public Health Laboratory, DPH

Is it possible to set test number target for pilot studies? If so, the data can then be analyzed once that target has been achieved?

**Next Steps:**

* Meetings in CY2025 will include a process for a minimum of one time per year to examine conditions in pilot, determine if they meet criteria for inclusion on the mandatory panel, and have members vote to move to the mandatory panel, keep in pilot, or discontinue screening.
* Member should inform Dr. McAlmon if you would like to be a member of the NBS Condition Nomination Work Group.
* Members should inform Dr. McAlmon if you are interested in helping prepare for or participate in a Legislative Briefing in 2025.
* Members should send recommendations for potential committee members to [Tresa.Glover-Smith@mass.gov](mailto:Tresa.Glover-Smith@mass.gov).

**Slides:**



**Meeting Adjourned**