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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Newborn Screening Advisory Committee Meeting

Wednesday, January 8, 2025: 4:00 – 6:00 PM.

Virtually via ZOOM

**Determination of Quorum:** Quorum was met.

**Members in Attendance:** Karen McAlmon, Inderneel Sahai, Olaf Bodamer, Christie Higuera, Catherine (Katie) Brown, Henry Dorkin, Nick Epie, Dallas Reed, Geoff Binney, Richard Parad, Monica Liao, Mary Alice Abbott, Anne Comeau

**Members Absent: Feria Ladha, Christine Mitchell, Chloe Schwartz, Yvonne Sheldon, Birth Hospital Representative (open)**

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

**Guests and Speakers:** Annie Counihan, Jaime Hale, Caroline Villaruz, Davinder Kaur, Binod Kumar

**Welcome and Agenda:**

Dr. McAlmon welcomed all to the meeting and proceeded with agenda items and instructions.

She introduced and welcomed our new members, Dr. Olaf Bodamer and Ms. Christie Higuera.

She reminded all that members should complete the open meeting law form ([download](https://www.mass.gov/doc/certificate-of-receipt-of-open-meeting-law-materials-102120/download)) and forward to Tresa Glover-Smith at tresa.glover-smith@mass.gov. She announcement that the NBSAC website is now on-line at <https://www.mass.gov/orgs/massachusetts-newborn-screening-advisory-committee-nbsac> . Please review the website.

She requested that all review the documents that were sent out prior to the meeting, specifically: .

* The member list— Please forward any corrections or updates to Tresa Glover-Smith at [tresa.glover-smith@mass.gov](mailto:tresa.glover-smith@mass.gov).
* The meeting dates for the remainder of the year

Quarterly meetings and back up meetings have been scheduled for the 2025 calendar. We would like at least one meeting to be in person this year, likely summer or fall of 2025. We should consider having a symposium (an half or day-long meeting in which multiple agenda items that require longer discussion or member education for a variety of votes might be covered).

The Committee is still looking to fill a vacancy with a parent/consumer, and a hospital administration representative. If you know of anyone that may be interested, please invite them to submit a statement of interest and resume to Tresa.Glover-Smith@mass.gov.

**Member Introductions were completed.**

**Minutes were Approved**:

11/8/24 Meeting Minutes Review and Vote for Approval

Member Role call to approve minutes

12 Yes

0 No

1 Abstain

Minutes were approved.

**Brief Overview**

Dawn Fukuda presented on the committee’s Roles and Responsibilities in service to children and families in the Commonwealth. The role of newborn screening is to test infants for treatable disorders. Please refer to the presentation for the full information on the roles and responsibilities (NBSAC Roles and Responsibilities, 1-8-2025).

**Update on Pilot Screens and Processes**

To begin the annual review of the screens in the Commonwealth’s current Pilot Program, Dr. Anne Comeau started the discussion with a presentation beginning with the oldest pilots. After the presentation of each disorder, the committee needed to make recommendations about whether to continue it on the pilot panel, to move it to the mandatory panel, or to discontinue it as a targeted disorder based on the NBSAC Guiding Principles. These included:

1. A set of four disorders that have been included in the pilot program since 2015: dienoyl-CoA reductase deficiency, malonic acidemia, medium/short chain L-3-OH acyl-CoA dehydrogenase deficiency, Hyperornithinemia, Hyperammoninemia, Homocitrullinemia Syndrome
   1. No births have been identified with these disorders.
   2. There was a lengthy discussion about the legal ramifications of missing a disorder, what other states are doing and the difference between by products and specific screening. Note: Most of these (except malonic acidemia) could be listed only as “by-products” of screening done for other disorders. No final decision was made as the Committee felt we needed additional information about the issues raised. Discussion will be finalized at the next meeting.
2. A set of four disorders that were included in the pilot program since 2018
   1. Dr. Comeau reviewed the Massachusetts experience with screening for Spinal Muscular Atrophy (SMA), including screening data and clinical outcomes. She presented a matrix aligning each of the Committee’s Guiding Principles to frame the Committee discussion. **After discussion, the Committee voted unanimously to recommend addition of SMA to the mandatory panel. Vote: 13:0**
   2. Dr. Sahai reviewed the New England experience with screening for X-linked Adrenoleukodystrophy (X-ALD), including screening data and clinical outcomes. After discussion, the Committee determined that the treatment available for the congenital cerebral form of X-ALD was known to be beneficial and therefore met criteria in the NBSAC Guiding Principles. **The Committee voted to recommend addition of X-ALD to the mandatory panel. Vote: 11 yes, 0 no 1 abstain**
   3. There was no time to discuss **MPSI** or **Pompe** and **discussion of both were moved to the next meeting.**
3. A set of two disorders (**MPSII** and **GAMT**) that were only recently added to the pilot panel and for which screening has not yet been implemented were noted. **No recommendations are needed for these.**

Unfinished business will be discussed at the February 12th, 2025, meeting

**Listening Session:**

There were no requests for guests to speak at this meeting

**Meeting Adjourned**