Amendments to 105 CMR 270.000: Blood Screening of Newborns for Treatable Diseases and Disorders

Massachusetts Department of Public Health and
University of Massachusetts Medical School
New England Newborn Screening Program

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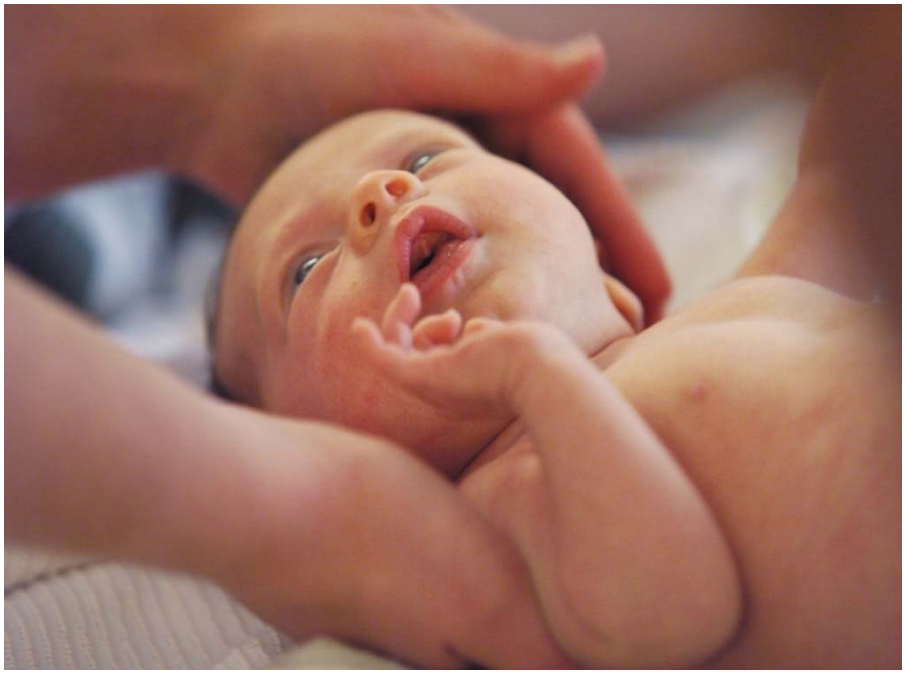

I. Background

- DPH has established a Newborn Screening Program to ensure that newborns are tested for treatable genetic, biochemical or infectious disorders or diseases.
- The DPH Newborn Screening Program is operated by the New England Newborn Screening Program at the University of Massachusetts Medical School and has been in continuous operation since 1962, screening approximately 80,000 newborns per year.
- The disorders or diseases tested for are determined by the Commissioner with the advice of the Advisory Committee on Newborn Screening ("Advisory Committee").

I. Background *continued*

- Pursuant to M.G.L. c. 111, §110A, the Commissioner has the authority to mandate that newborns be screened for specified disorders or diseases unless the parents/guardians have a religious objection.
- For some disorders or diseases that do not meet the criteria to be added to the mandated screening list, but for which there is some presumption of benefit to the newborn, the Commissioner may recommend inclusion of the disorder as a pilot study.
- There are currently 30 disorders or diseases included on the mandated screening list and others are included as pilots.

II. Proposed Amendments

Significant proposed amendments include:

- Definitions: New definitions were added and others were revised for clarity.
- Mandated Screening: Two new screening tests were added to the list of mandated screening: Severe Combined Immunodeficiency (SCID) and Carnitine-acylcarnitine translocase deficiency (CACT). Both have been previously tested as part of the pilot program.
- Pilots: The list of optional pilot program tests have been removed from the regulations. They will be listed in the Newborn Screening brochure and on the NENSP website.

II. Proposed Amendments (cont'd)

- Collection and Submission of Newborn Blood Specimens.
  - The time period in which specimens must be submitted has been changed from 24-72 hours to 24-48 hours to better align with the optimal window of time for testing and turnaround time for optimal outcomes for certain disorders.
  - The procedures for opting out of newborn screening for religious reasons are clarified and a refusal form is now available from the NENSP to assist providers in documenting parent refusal.
  - The health care provider attending to a newborn will be required to provide information to parents about optional newborn screening tests offered as pilot studies and document verbal consent for optional pilot screening on the collection device.

II. Proposed Amendments (cont'd)

- Confidentiality. Proposed revisions clarify confidentiality requirements and specify for what purpose and to whom newborn screening results may be released, including for research.
- Storage and Use of Residual Specimens and Data.
  - This new section would add requirements for the storage and use of specimens remaining after newborn screening is completed and require NENSP to comply with DPH policy on retention of residual blood spots;
  - Storage of residual dried blood spots would be reduced to a period of 15-16 years (prior policy was 22 years);
  - Parents will be able to request destruction of their child's residual blood spot after it is no longer needed for the testing and follow up of newborn screening services for their child.

III. Request for Approval

- After initial presentation to the PHC in February 2016, DPH held a public comment period for this regulation.
- During this time, DPH received no comments regarding proposed changes to this regulation.
- As no comments were received, DPH requests approval to move forward with amendments to the regulation as originally proposed.

Questions?