

STATE OF CONNECTICUT

Connecticut Department of Public Health
Dr. Katherine A. Kelley State Public Health Laboratory
395 West Street; Rocky Hill, CT 06067

March 8, 2017

Re: Change in Notification Practices for Positive Newborn Screening Hemoglobin Results

Dear Primary Care Provider and Office Manager,

The Connecticut Newborn Screening (CT NBS) Program has reviewed its processes for reporting abnormal NBS results suggestive of Hemoglobin (Hb) disease or trait. **Effective immediately**, we will no longer be notifying **parents** directly of screen positive Hb results. Best practice dictates that parent notification of an abnormal Newborn Screening (NBS) result, suggestive of Hb disease or trait, take place through the Primary Care Provider's (PCP) office.

You can expect to receive notice of all screen positive Hb results for your patients by telephone, followed by a written notification faxed to your office. In the case of suspected Hb disease, we will continue to notify you of referral to a specialist at one of Connecticut's regionally based diagnostic and treatment centers:

- Dr. Farzana Pashankar at the Yale School of Medicine Pediatric Hematology and Oncology Program, 203.785.4640
- Dr. Donna Boruchov at the Connecticut Children's Medical Center (CCMC) Center for Cancer & Blood Disorders, 860.545.9360

It is important to remember that NBS results are not diagnostic, and further diagnostic work-up is necessary for children who screen positive for Hb disorders and traits.

You will find a letter included in this communication, notifying you of a screen positive result for one of your patients. We are in the process of updating all of our printed materials including our PCP notification letters and recommendations. In the interim we recommend the following:

- Review the appropriate American College of Genetics ACT Sheet/Confirmatory Algorithm at: http://www.acmg.net/Admin/ACT_Sheets_and_Confirmatory_Algorithms/NBS_ACT_Sheets_and_Algorithm_Table/ACMG/Resources/ACT_Sheets_and_Confirmatory_Algorithms/NBS_ACT_Sheets_and_Algorithms_Table.aspx?hkey=aa84b9b4
- Consultation with one of the above named diagnostic/treatment centers
- Referral for genetic counseling when appropriate
- Referral to one of the parent advocacy and support agencies when appropriate:
 - Citizens for Quality Sickle Cell Care (Greater Hartford area): <http://www.cqsc.org/index.php/home>
 - The Sickle Cell Disease Association of America Southern Connecticut: <http://www.scdaaofsouthernct.org/>

Connecticut Newborn Screening Short Term Follow-Up Unit
Telephone: (860) 920-6628 Fax: (860) 920-6633

Also included in this communication is a compilation of links to resources and fact-sheets that may be beneficial to parents.

It is important to notify the CT NBS Program immediately if the infant named in the attached letter is **not** followed by your practice. Please keep in mind that the infant's name may have changed since the time of the initial NBS. You may need use the infant's date of birth, mother's name or other identifying information to locate this infant in your system.

Lastly, we request that you notify the CT NBS program at the fax number below of the final result of confirmatory testing completed by your office related to this NBS result.

Please feel free to contact the CT NBS program at the number below with any questions.

Sincerely,

Marie Burlette, RN, BSN, MPH
Supervising Nurse Consultant
Connecticut Newborn Screening
Short Term Follow-up Unit

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Resources for Parents

Diagnostic Testing

Connecticut Children's Medical Center:

- Blood Test: Hemoglobin Electrophoresis
<http://www.connecticutchildrens.org/index.php/healthinfo/parents/doctors-and-hospitals/medical-tests-and-exams/blood-test-hemoglobin-electrophoresis/>
- Genetic Testing: <http://www.connecticutchildrens.org/index.php/healthinfo/parents/doctors-and-hospitals/medical-tests-and-exams/genetic-testing/>

KidsHealth.org:

- Hemoglobin Electrophoresis: [Hemoglobin Electrophoresis: http://kidshealth.org/en/parents/test-electrophoresis.html?ref=search](http://kidshealth.org/en/parents/test-electrophoresis.html?ref=search)

Hemoglobinopathies

- Baby's First Test: Hemoglobinopathies (general information on hemoglobinopathies detected by NBS, follow-up testing and parent resources): <http://www.babysfirsttest.org/newborn-screening/conditions/hemoglobinopathies>

Disease/Trait Specific Information

Connecticut Children's Medical Center:

- Sickle Cell Disease: <http://www.connecticutchildrens.org/index.php/healthinfo/parents/diseases-and-conditions/heart-and-blood-vessels/sickle-cell-disease/>

Yale Health Network:

- Hemoglobin S: http://healthlibrary.ynhh.org/Search/167,hemoglobin_s
- Sickle Cell Disease in Children: <http://healthlibrary.ynhh.org/Search/90,P02327>
- Hemoglobin E Trait: <http://healthlibrary.ynhh.org/Search/160,12>
- Hemoglobin C in Children: <http://healthlibrary.ynhh.org/Search/160,9>

Genetic Alliance:

- Alpha Thalassemia Trait: <http://www.diseaseinfosearch.org/Alpha+Thalassemia+Trait/328>
- Sickle Cell Disease: <http://www.diseaseinfosearch.org/Sickle+cell+anemia/6589>
- Sickle Cell Trait: <http://www.diseaseinfosearch.org/Sickle+Cell+Trait/6591>
- Sickle Cell Hemoglobin C Disease: <http://www.diseaseinfosearch.org/Sickle+cell+hemoglobin+C+disease/9842>

SaveBabies.org:

- A Family Story, Newborn Screening and Sickle Cell Disease: <http://www.savebabies.org/blog/2013/03/a-family-story-newborn-screening-sickle-cell-disease/>

Centers for Disease Control and Prevention:

- What you should know about sickle cell trait
 - English: https://www.cdc.gov/ncbddd/sicklecell/documents/SCD%20factsheet_Sickle%20Cell%20Trait.pdf
 - Spanish: https://www.cdc.gov/ncbddd/spanish/sicklecell/documents/2013_sicklecelltraitfactsheet_spanish.pdf
 - French https://www.cdc.gov/ncbddd/sicklecell/documents/factsheet_sickle_cell_trait-french.pdf
- Facts about Thalassemia:
 - <https://www.cdc.gov/ncbddd/thalassemia/facts.html>

Resources for Parents Continued

Disease/Trait Specific Information

KidsHealth.org:

- Thalassemias: <http://kidshealth.org/en/parents/thalassemias.html>
- Beta Thalassemias: <http://kidshealth.org/en/parents/beta-thalassemia.html?ref=search>

Fact Sheets for Parents

The Screening, Technology and Research in Genetics (STAR-G) Project:

- Alpha Thalassemia: http://www.newbornscreening.info/Parents/otherdisorders/Alpha_thal.html
- Sickle Cell Disease: <http://www.newbornscreening.info/Parents/otherdisorders/SCD.html>

Advocacy/Support

- Citizens for Quality Sickle Cell Care (Greater Hartford): <http://www.cqsc.org/index.php/home>
- Sickle Cell Disease Association of America Southern Connecticut: <http://www.scdaaofsouthernct.org/>