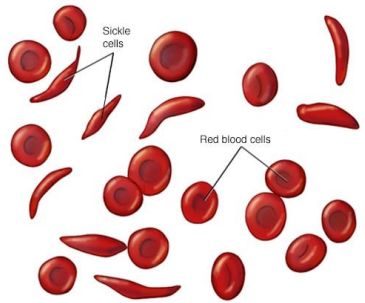
September 24, 2024

Overview of Connecticut's Sickle Cell Disease Collaboration

Sickle Cell Disease (SCD)

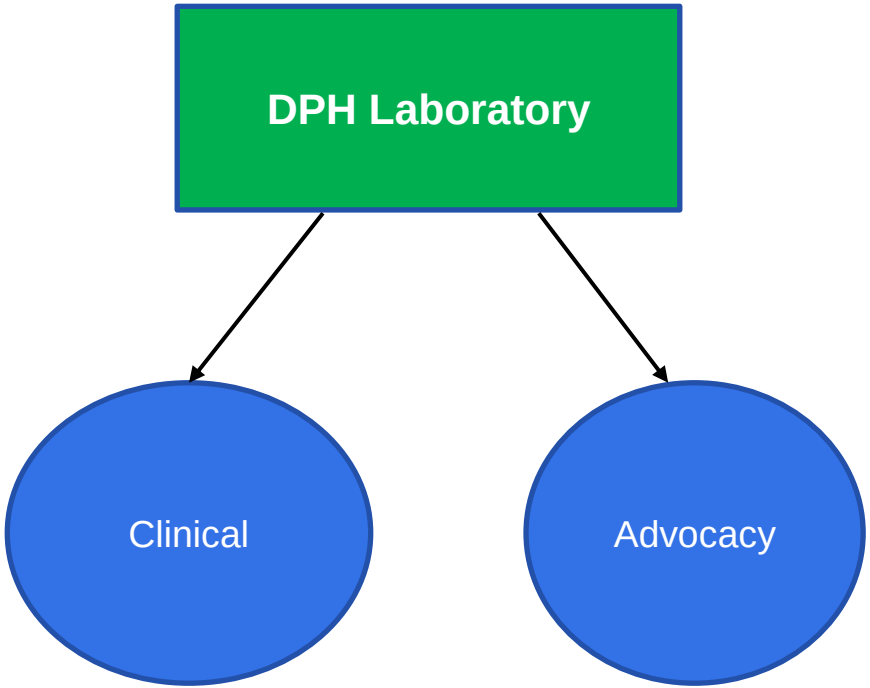
- Sickle Cell Disease (SCD) is in a group of inherited red blood cell disorders that are present at birth.
- Red blood cells contain hemoglobin, which is a protein that carries oxygen.
- Healthy red blood cells are round and move through blood vessels to carry oxygen throughout the body.
- When someone has SCD, the hemoglobin is abnormal, and the red blood cells become hard and sticky and look like a C-shaped farm tool called a “sickle.”
- The sickle cells die early, which causes a constant shortage of red blood cells and sickle cells can get stuck and clog the blood flow leading to pain and serious health problems such as infections, acute chest syndrome and also stroke.



Sickle Cell Disease (Continued)

- There are several types of SCD. The specific type of SCD a person has depends on the genes they inherited from their parents.
- A person can also have Sickle Cell Trait (SCT), where they inherit a hemoglobin “S” gene from one parent and a normal gene (one that codes for hemoglobin “A”) from the other parent.
- People with SCT usually do not have any of the signs of the disease but can pass the abnormal gene to their children. If both parents have the trait, there is a 25% chance that any child conceived may have SCD.
- It is estimated that Sickle Cell Disease (SCD) affects 90,000 to 100,000 people in the United States, with most people being Black or African American. In the United States, SCD occurs in about 1 out of every 365 Black or African American births and 1 out of every 16,300 Hispanic American births.
- Approximately 1 in 13 Black or African American babies is born with Sickle Cell Trait (SCT).

Overview of Connecticut's Program



Overview Connecticut Newborn Screening Program

General

- Section/Branch: State Public Health Laboratory/NBS Program
- Funding: General Funds
- 36,436 (99.7%) newborns screened 2023
- Can detect 72 disorders through bloodspot screening
- Most recently added:
  - ✓ Mucopolysaccharidosis Type-II (MPS-II)
  - ✓ Guanidinoacetate methyltransferase(GAMT) deficiency
  - ✓ Validating method for Krabbe

Goals

- All newborns receive a timely, valid NBS
- Cost-effective, reliable & sensitive NBS testing
- All infants with a PP NBS result receive an appropriate diagnostic work-up
- Prevent serious illness, permanent disability and death in affected infants
- Collect and analyze NBS QI data & case data to promote early and accurate detection of NBS disorders

CT NBS Program: a Centralized System for Testing, Treatment & Follow-up

CT NBS Program

- ✓ Validates, implements & refines screening tests
- ✓ Makes initial contact with PCP following abnormal result & reports abnormal result to CT NBS Network (funded by DPH)
- ✓ Reports all hgb trait results with recommendations for follow-up to child's PCP by phone & in writing
- ✓ Submits deidentified confirmed case & timeliness data for MCHBG and to national NBS data repository (MOU in place)
- ✓ Collects and analyzes NBS QI data & case data

The CT NBS Network:

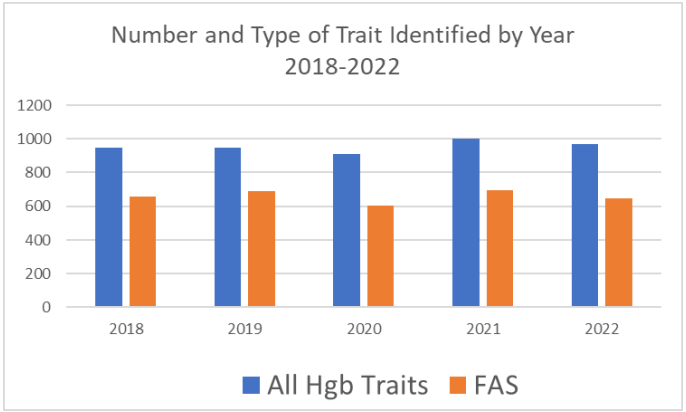
- ✓ Coordinates with PCP/Parent to start diagnostic process
- ✓ Sends short-term f/u data to CT NBS (diagnosis confirmed vs. ruled out)
- ✓ Maintains CT confirmed case data registry
- ✓ Provides care coordination for children who confirm with a disorder
- ✓ Collects data on global & disease specific outcome measures
- ✓ Sends long-term f/u data to CT NBS

Connecticut NBS Hemoglobinopathy Screening

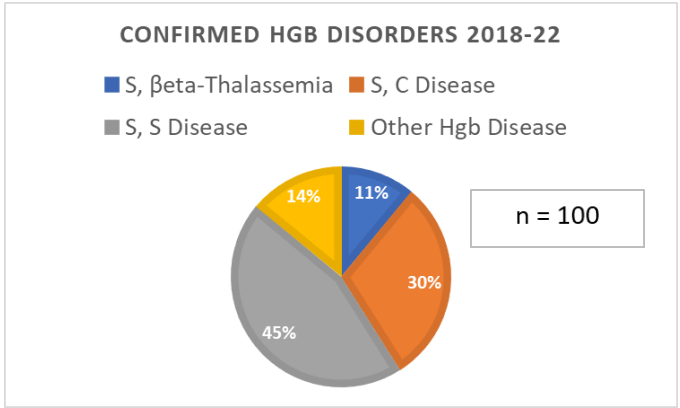
The Advisory Committee on Heritable Disorders in Newborns & Children Recommends Screening for:

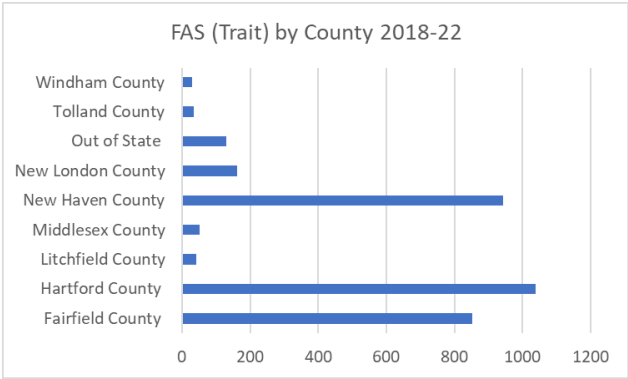
- ❑ **Core RUSP**
  - ✓ **S, βeta-Thalassemia**
  - ✓ **S, C Disease**
  - ✓ **S, S Disease (Sickle Cell Anemia)**
- ❑ **Secondary RUSP**
  - ✓ **Various other Hemoglobinopathies**

CT NBS Stats



- ❖ Hgb FAS is suggestive of Sickle Cell Trait.
- ❖ Hgb FS is suggestive of Sickle Beta Thal or Sickle Cell Disease
- ❖ All abnormal Hgb results require diagnostic confirmation





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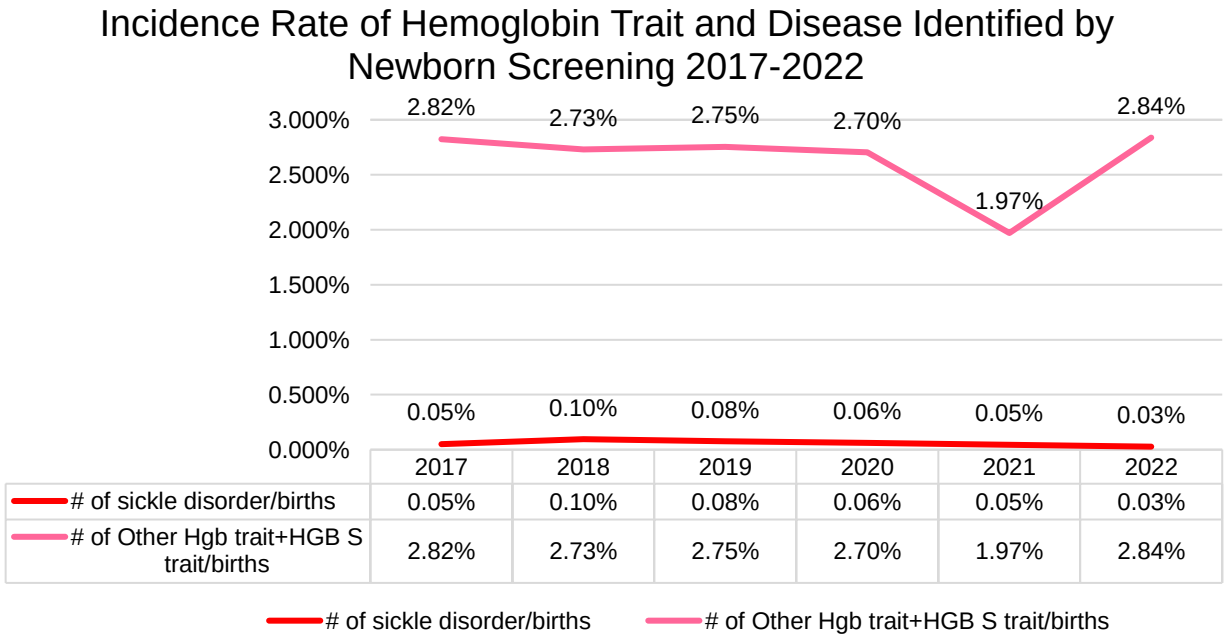
Public Health

The CT NBS Program received funding through the State Newborn Screening System Priorities Program (NBS Propel) HRSA-23-065 to improve short-term and long-term follow-up, and help families understand and navigate the process from confirmation of a diagnosis to treatment, and follow-up across the lifespan of the affected individual. These activities include expanding access to diagnosis and treatment resources for providers and families of infants with NBS disorders with a focus on reaching underserved/marginalized populations, and empowering families to actively engage at all levels of the newborn screening system

- Identified opportunities for improvement:
  - Notification and education of families and healthcare providers of trait results
  - Tracking of diagnostic outcome following trait reports
  - Connecting families with a hematologists and genetic counselors.
  - Identifying and responding to care gaps for children with confirmed SCD
  - Identifying long-term follow-up outcome measures to track for children with confirmed SCD

DPH Laboratory

- The Connecticut Newborn Screening Program at the DPH Laboratory receives a blood spot card for 99.99% of babies born in the state which are screened for many medical conditions including SCT and SCD.
- When a newborn screens positive for SCD, the CT Newborn Screening Program reports the result to the baby's pediatrician and a specialist in blood disorders (a pediatric hematologist). The specialist works with the family and pediatrician to order diagnostic tests and determine a plan for follow-up and treatment.



Sickle Cell Trait

In 2022 approximately 645 babies in CT are identified as having sickle cell trait.

More than 80% of adults with sickle cell trait do not know their status.

Why is it Important?

- Potential health risks associated with sickle cell trait
  - -- pain crises
  - -- heat stroke
  - -- muscle cramps or weakness

- Allows time to discuss screening and testing options in future pregnancies, and review other options like preimplantation testing.

- Individuals or their parents with SCD traits can become advocates for the condition, raising awareness, and supporting research and initiatives for access and coverage for better management and treatment.

Sickle Cell Trait: Current State

Newborn screen suggestive of sickle cell TRAIT

DPH sends fax to PCP recommending:

1)They notify the family

2) They obtain follow-up testing at 6 months

3) Resource provided for the family to schedule a no-cost telehealth appointment with a genetic counselor

Sickle Cell Trait: Need for Change

Of 645 sickle cell traits reported in 2022:

- 1 scheduled an appointment with the genetic counselor (and then was a no-show for the visit)

Goals of this Project

► More families will access follow-up resources (recommended labs, genetic counseling)

► More individuals will be aware of their trait status

SCD Pediatric Clinical Treatment in Connecticut

Pediatric hematology services are provided by **Connecticut Children's (CC)** and **Yale New Haven Health (YNHH).**

- Provide comprehensive, family-centered care
- Offer patient and family education, family support programs, and genetic counseling
- Conduct research to advance SCD treatment
- Enhance the transition process from pediatric to adult care.
    - Baseline assessments of adolescent and family in preparation for transition to adult care.
    - Identifying areas of improvement in transition processes statewide.
    - Implement identified needed changes to improve transition processes.
    - Repeating patient/family evaluation to ensure consistent outcomes.
    - Measuring impact of changes to transition process.

SCD Adult Clinical Treatment in Connecticut

Adult hematology services are provided by the **University of Connecticut Health Center (UConn Health)** in partnership with the **New England Sickle Cell Institute (NESCI)** and **Yale New Haven Hospital (YNHH).**

Conduct a baseline assessment of SCD adult and address the areas of need

- Identify adults with SCD not receiving care in a CT
- Address areas of need and develop a multi-disciplinary and multi-agency (Department, OEC, DOE) action plan

UConn Health/NESCI will also:

- Support and create linkages to medical homes for primary care, and coordinate access to a medical home, including linkages to Access and Safety Net Programs
- Improve state-wide SCD Emergency Department protocols
- Conduct community outreach, and education to CT residents about SCD and increase family support activities to those diagnosed with SCD/Trait

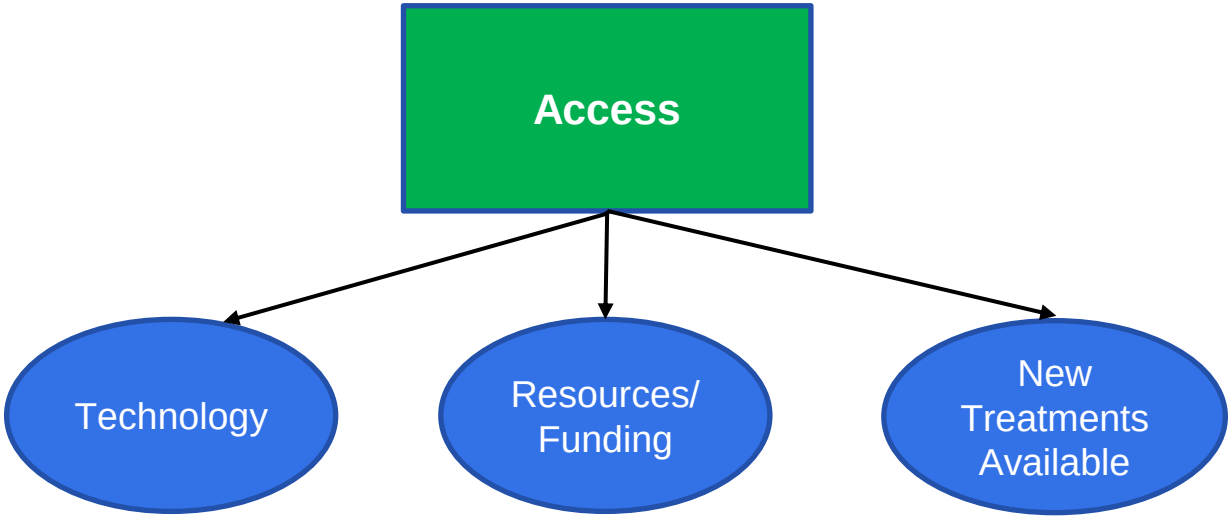
SCD Advocacy

- Sickle Cell Disease Consortium: https://sicklecellconsortium.org/connecticut/
- Community Support Groups
    - Michelle's House: https://www.michelleshousect.org/
- Parent Engagement Groups
    - Connecticut Children's SCD Family Advisory Council
    - UConn Connecticut and Rhode Island Sickle Cell Disease Community Advisory Board
- Parents Available to Help (PATH CT) https://pathct.org

Improvements

- Improvements:
    - Cost of care- conversations with payors, state Medicare and Medicaid
    - Funding- look for more funding opportunities
    - Engage and educate individuals living with SCD and their families
    - Examining data and sharing with stakeholders
    - Advance Equity and Engagement

Limited Access for SCD



Statewide Resources

- The Connecticut DPH: www.ct.gov/dph
- Yale-New Haven Health: https://www.ynhh.org/services/hematology/Sickle-Cell-Program
    - Adult Medicine: https://www.ynhh.org/services/hematology/Adult-Sickle-Cell-Clinic
    - Pediatric Medicine: https://www.ynhh.org/childrens-hospital/services/cancer-and-hematology/sickle-cell-disease
- University of Connecticut (UConn Health):
    - New England Sickle Cell Disease Institute: https://health.uconn.edu/cancer/patient-services/centers-and-interdisciplinary-clinics/new-england-sickle-cell-institute/
- Connecticut Children's: https://www.connecticutchildrens.org/specialties-conditions/pulmonary-medicine/sickle-cell-pulmonary-clinic
- SCDAA Southern CT Inc: https://www.michelleshousect.org/
- Connecticut Newborn Screening Network: https://www.connecticutchildrens.org/specialties-conditions/connecticut-newborn-screening-network

Funding

- **Name of Project:** HRSA-23-065 State Newborn Screening System Priorities Program (NBS Propel)
- **Name of Funding Agency:** HRSA (Health Resources and Services Administration)
- **Date of Grant Period:** July 1, 2023 through June 30, 2028 (5 years)
- **Project:** Work with Sickle Cell Disease Stakeholders in CT to explore and document current state processes, data, and staffing, and to develop a plan for education and implementation of a statewide carrier status follow-up initiative.
    - Develop state-specific SCD/T materials for parents and providers
    - Work with SCD stakeholders to explore areas of improvement in reporting/follow-up
    - Incorporate parent and provider feedback into initiatives.

CT Sickle Cell Partners

- The Connecticut DPH: Community, Family Health, and Prevention Branch
- The Connecticut Public Health Laboratory- Newborn Screening Program
- Connecticut Children’s
- Yale New Haven Health
- University of Connecticut Health (UConn)
- Connecticut Newborn Screening Network
- New England Sickle Cell Institute (NESCI).

Questions?