Michigan Department of Community Health

Newborn Screening for Hemoglobinopathies

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Newborn Screening Follow-up and Medical Management
Disorders Screened

- **43 Metabolic disorders**
  - 25-30% of total cases detected

- **10 Non-metabolic disorders**
  - 70-75% of total cases detected
    - Hemoglobinopathies - 5
    - Endocrine disorders - 2
    - Immunodeficiencies - 2
    - Cystic fibrosis
Hemoglobinopathies

- Target - Sickling conditions
  - Sickle cell anemia (SS)
  - Sickle β thalassemia (Sβ° Sβ+)
  - SC disease

- Screen for Hb A, S, C, D, E and Bart’s by HPLC
Hemoglobinopathies (cont.)

- Detect some cases of $\beta^0$ thalassemia (Low A) and most cases of hemoglobin H disease (↑ Bart’s) by HPLC

- Detection rate $\sim 1:2,000$

- Number detected (2013) = 55
**Hemoglobinopathies Screening Goals**

- Penicillin prophylaxis by 4\textsuperscript{th} month of life; vaccine schedule
  - 35% deaths pre NBS

- Prevent early deaths

- Early follow-up for case management and patient care.
Prophylactic Penicillin Study (PROPS) 1983 - 1985

- Randomized double-blind placebo-controlled study
- 85% decrease in the rate of pneumococcal infection for children receiving penicillin
- Begin penicillin prophylaxis by 4 months of age

NBS Program Overview

83 Hospitals
90 Midwives

~111,000
~750

negative reports

MDCH NBS Laboratory

Positives, early, and unsats

Education/Training
Quality Assurance Reports
Monitoring

MDCH NBS Follow-up Program

Positives, early, and unsats

PCP Medical Home

Cystic Fibrosis
U of M

Endocrine
U of M

Metabolic
CHMMC

Hemoglobinopathies
SCDAA-MI
CHM

Severe Combined
Immunodeficiency
CHM/Wayne
State

Hemoglobinopathy Quality Improvement Committee
(HemQIC)
MDCH HemQIC
(Hemoglobinopathy Quality Improvement Committee)

Reviews MDCH NBS system for provision of screening diagnostic treatment services for the hemoglobinopathies.
NBS Information

- newbornscreening@michigan.gov
- www.michigan.gov/newbornscreening
- www.michigan.gov/sicklecell