

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 ($S\beta^{\circ} S\beta^{+}$)
 - SC disease
- Screen for Hb A, S, C, D, E and Bart's by HPLC

Hemoglobinopathies (cont.)

- Detect some cases of β^0 thalassemia (Low A) and most cases of hemoglobin H disease (\uparrow Bart's) by HPLC
- Detection rate \sim 1:2,000
- Number detected (2013) = 55

Hemoglobinopathies

Screening Goals

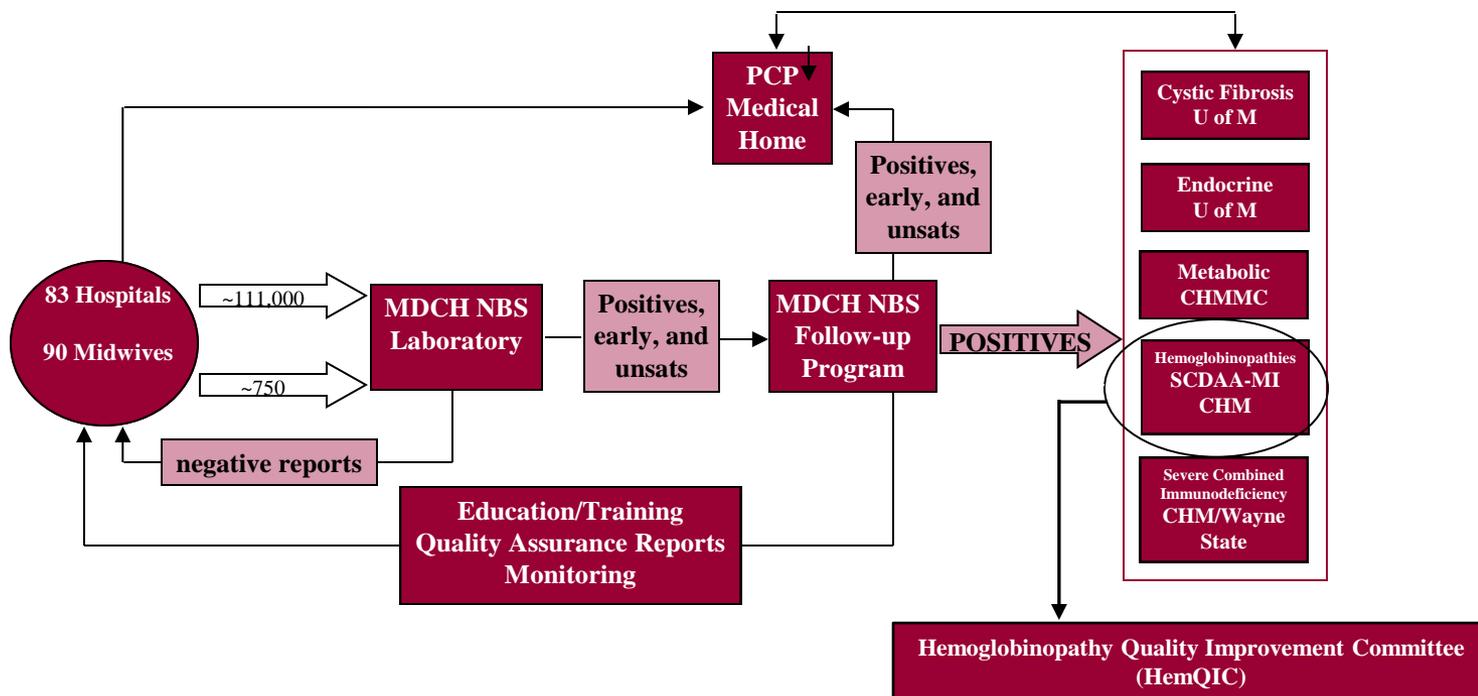
- Penicillin prophylaxis by 4th month of life; vaccine schedule
 - 35% deaths pre NBS
- Prevent early deaths
- Early follow-up for case management and patient care.

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

Gaston MH, Verter JI, Woods G, et al. Prophylaxis with oral penicillin in children with sickle cell anemia: a randomized trial. *N Engl J Med.* 1986 Jun;314(25):1593–1599.

NBS Program Overview



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