Michigan Department of Community Health

MI Newborn Screening Program:

An Overview of Follow-up & Case Management for Sickle Cell Conditions

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Session Goals

- Understand purpose, process and impact:
  - Newborn Screening Program
  - Sickle Cell Disease Association of America (SCDAA-MI)
- Describe health outcomes and disparities affecting individuals living with a sickling condition.
  - Medical Implications
  - Access/Barriers to Care
- Identify opportunities for continued collaboration between SCDAA-MI and MIHP to support quality and program success.

Michigan's Newborn Screening Program

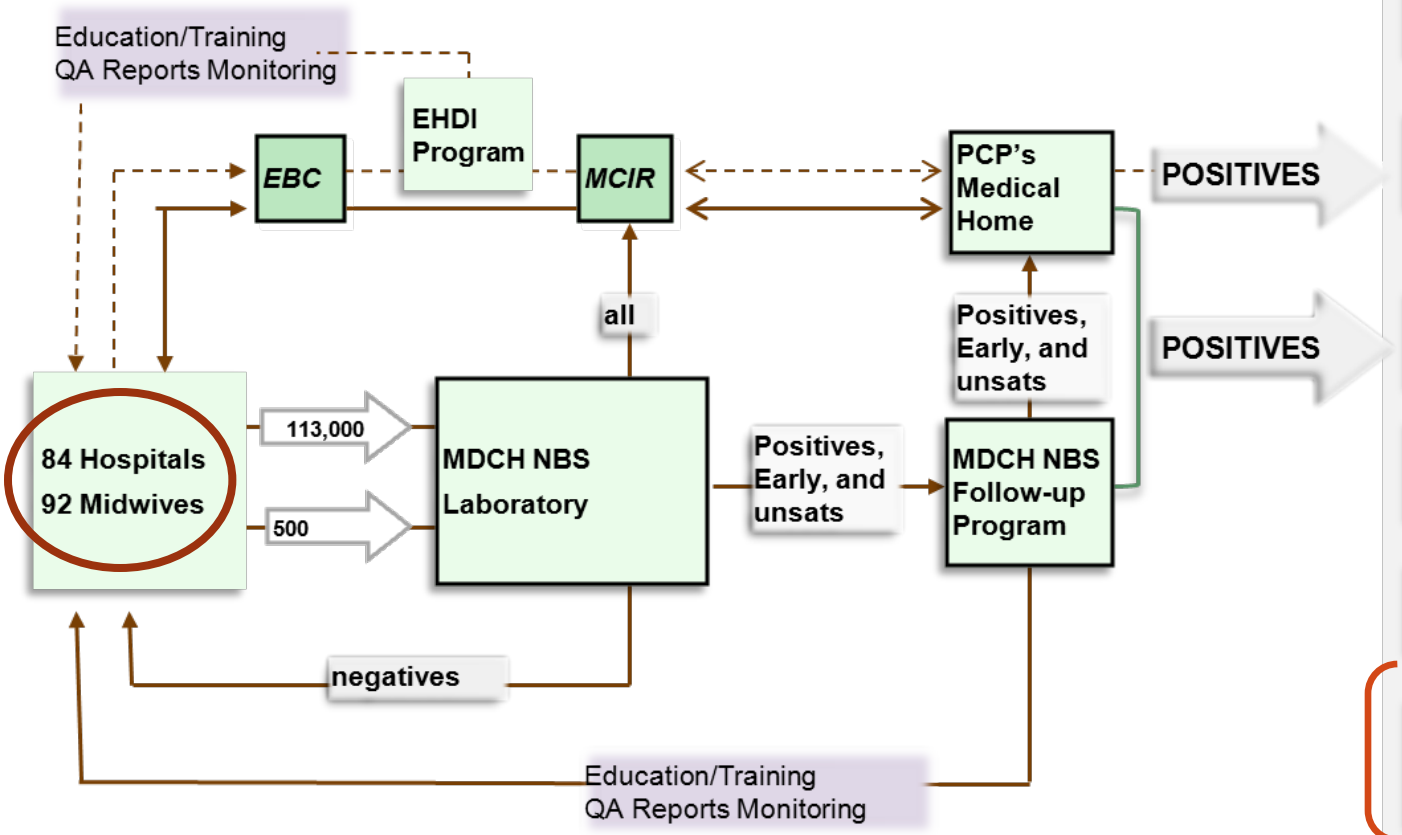
- A statewide public health program to find babies with rare but serious disorders
- Find newborns quickly before symptoms occur to assure early diagnosis and treatment
  - Mostly genetic
  - Often no family history
  - May not otherwise be detected at an early age
- Prevent early deaths
- Reduce human & financial costs of disease

Michigan's Newborn Screening Panel

- 14 Amino Acid Disorders
  - Ex: PKU, Maple Syrup Urine Disease
- 13 Fatty Acid Oxidation Disorders
  - Ex: Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
- 14 Organic Acid Disorders
  - Ex: Propionic Acidemia
- 2 Endocrine Disorders
  - Ex: Congenital Hypothyroidism, Congenital Adrenal Hyperplasia (CAH)
- 5 Groups of Hemoglobinopathies
  - Ex: Sickle cell anemia
- 7 Others: Biotinidase Deficiency, Cystic Fibrosis, Galactosemia, Severe Combined Immunodeficiency and T-cell related lymphocyte deficiencies, Hearing Loss, Critical Congenital Heart Disease

About 270 Michigan babies, or 1 out of every 400 screened each year, are found to have a condition diagnosed by NBS. From 1965 through 2012, over 6.6 million children were screened and more than 5,100 diagnosed.

Michigan Newborn Screening



MI NBS Follow-up Coordinating Centers

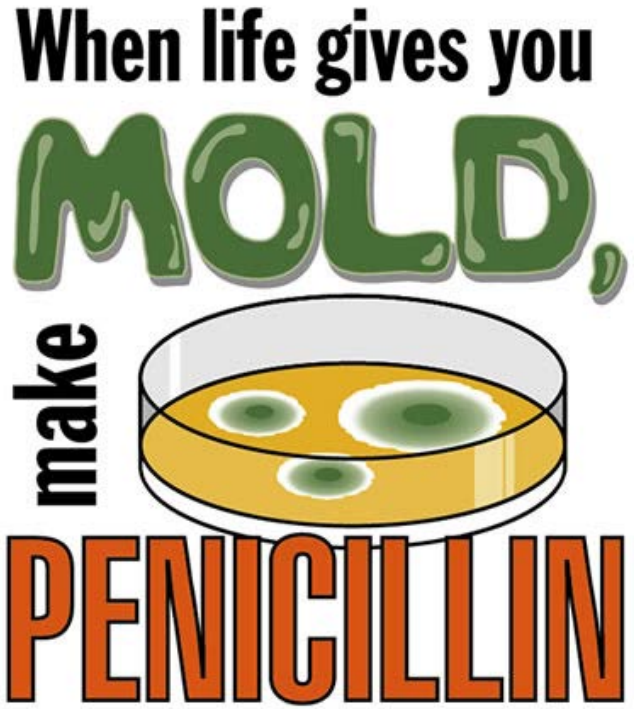
- Hearing Loss: Pediatric Audiology
- Cystic Fibrosis: U of M
- Endocrine: U of M
- Metabolic: CHM/Wayne State
- Hemoglobinopathies: Sickle Cell Association of MI/CHM
- Severe Combined Immunodeficiency: CHM/Wayne State

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**
- **Screening Goals**
  - Prevent early deaths
  - Penicillin prophylaxis by 4th month of life; vaccine schedule 35% deaths pre NBS
  - 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.

Hemoglobinopathies (cont.)

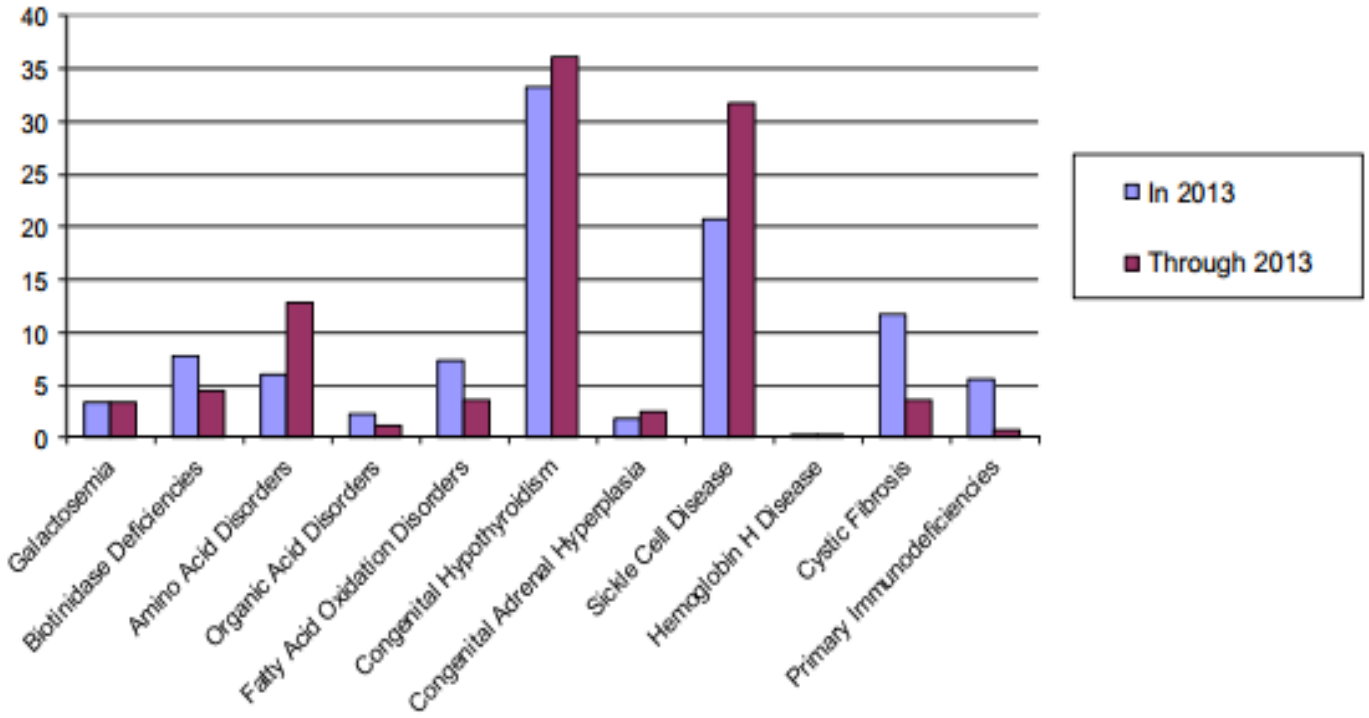
2013 Data:

- Detection rate ~ 1:2,043
- Number detected = 56
- Cases from 1987 - 2013 = 1,733
- African Americans = 87.5% of cases
  (incidence in African Americans is 1 in 417 screening in Michigan)
- Carriers identified = 2,772

Statewide approximately 2,800 residents suffer from SCD and 140,000 carry the sickle cell trait.

Hemoglobinopathies (cont.)

Percent Distribution of Disorders Identified in Newborns via Newborn Screening, Michigan Residents, in 2013 and through 2013

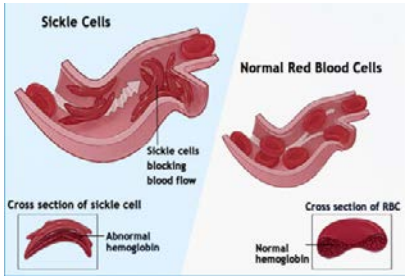


What is Sickle Cell Disease?

- Sickle cell disease (SCD) is a genetic disease of the red blood cell characterized by vaso-occlusion and hemolysis.

Vaso -Occlusion

- All cells have the genetic defect but are not always sickled



- Sickled cells block flow within blood vessels

- Area of decreased flow is deprived of oxygen ⟶ tissue damage and pain

Hemolysis

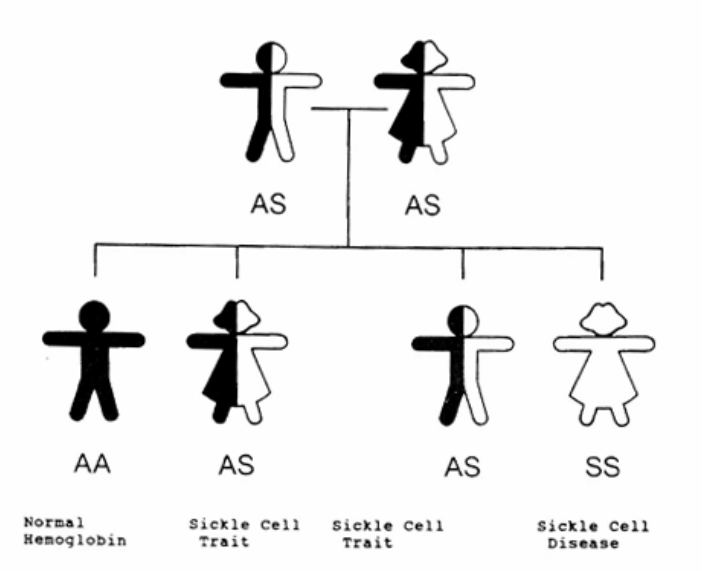
- Normal red cell lives 120 days

- Sickle cells may only last a few days: the cells break apart increasing bilirubin levels

- Bone marrow must work harder to try and compensate

Genetic Disease: Inheritance

If both parents have Sickle Cell Trait (AS):



For each pregnancy there is a 25% chance of inheriting the usual hemoglobin (AA), a 50% chance of inheriting Sickle Cell Trait (AS), and a 25% risk of inheriting Sickle Cell Disease (SS).

THESE CHANCES ARE THE SAME FOR EVERY PREGNANCY

- Both parents must have an abnormal hemoglobin trait to have a child with SCD.
- If both parents have trait there is a 1 in 4 (25%) chance that each baby will have SCD.

Effects of Sickle Cell Disease

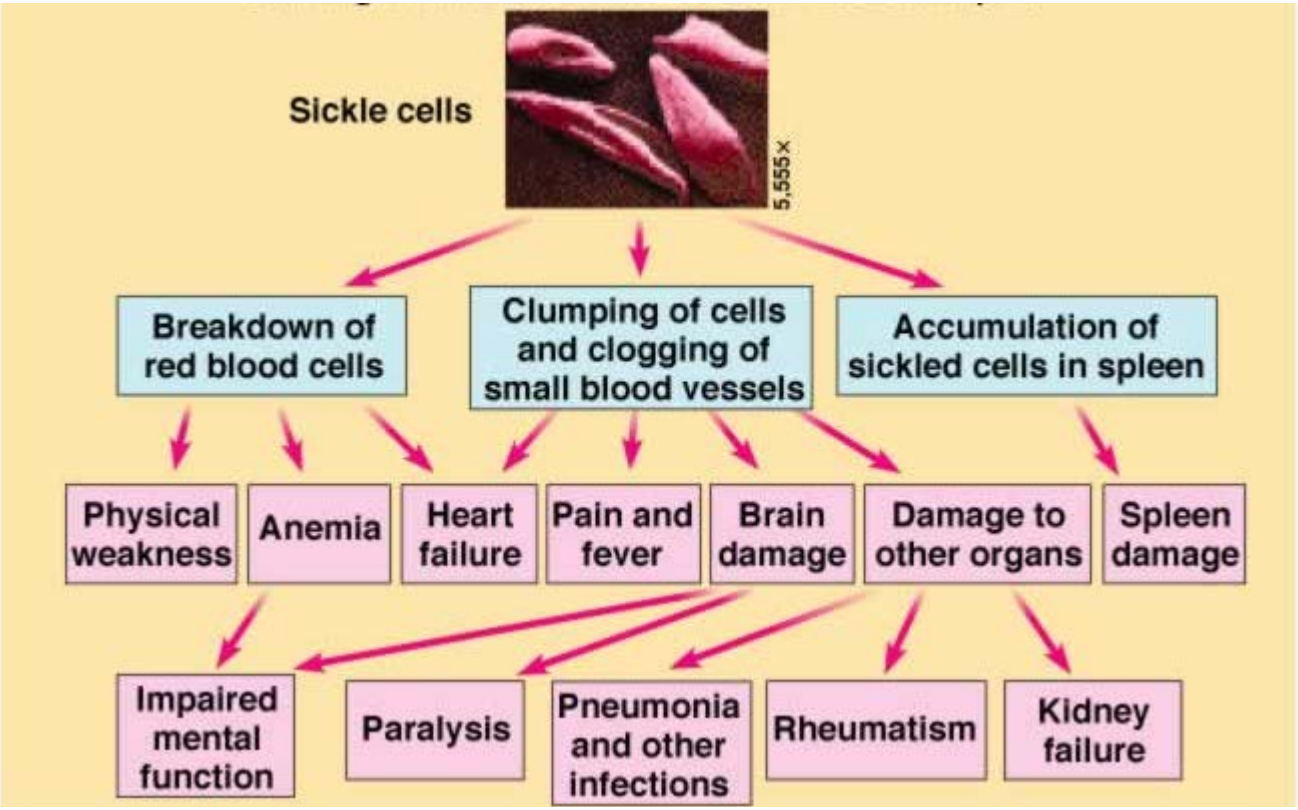


Image Source: Clinical Cases: Biochemistry for Medics

http://usmle.biochemistryformedics.com/answer-case-study-sickle-cell-disease-a-15-year-old-african-american-female-presents-to-the-emergency-room/

Treatments for Sickle Cell Disease

- Penicillin prophylaxis until age 5 years

- Immunizations: Prevnar, Pneumovax and Influen

- Blood Transfusions
  - Prevent stroke or protect lungs

- Hydroxyurea
  - Increase levels of fetal hemoglobin
  - Red blood cells live longer and sickle less
  - Can cause reversible neutropenia
  - Potential carcinogen or teratogen
  - Biggest problem is compliance

Cure for Sickle Cell Disease

- Bone marrow or stem cell transplant
- Can cure sickle cell, but risk of serious and fatal complications
- Reserved for the sickest children with sibling matches

Broad Impacts on Children & Adults

- Acute and chronic pain
- School/work absences; Managing work/careers
- Difficulty concentrating and other cognitive issues
- Receiving adequate medical treatment and support
- Stigmatization

Broad Impacts (cont.)

- Worry about their future
- Impact on families
- Maintaining relationships
- Emotional impacts
- Transition

Considerations for MIHP

- Discuss with parents the importance of penicillin prophylaxis adherence, vaccinations, and hydroxyurea.
- Become knowledgeable about sickle cell disease/trait inheritance and provide information to families.
- Refer families to the SCDAA-MI.
- MDCH to develop a public health strategic plan to address sickle cell disease across the lifespan.

Sickle Cell Disease Association of America- MI Chapter

Our Mission

TO MAXIMIZE THE LIFE POTENTIAL OF INDIVIDUALS LIVING WITH SICKLE CELL DISEASE

TO ENABLE INDIVIDUALS WITH SICKLE CELL TRAIT TO MAKE INFORMED DECISIONS THAT THEY BELIEVE ARE IN THEIR BEST INTEREST WITH RESPECT TO FAMILY PLANNING

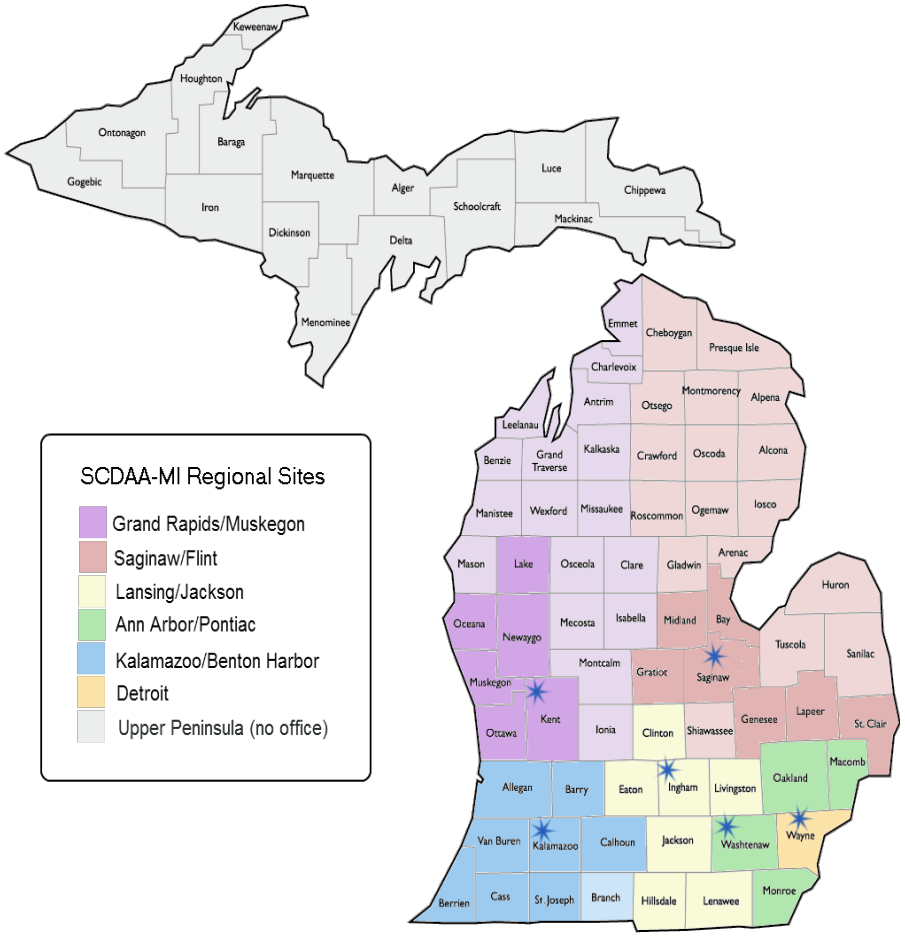
TO PROVIDE EDUCATION, TESTING AND INFORMATION FOR THE GENERAL PUBLIC

SERVICES PROVIDED

- Follow-up, laboratory testing, genetic counseling
- Education, medical referrals
- Program assistance, career development
- Community event presentations and displays
- Parent club support group, summer camp program

Satellite Offices

- Benton Harbor/Kalamazoo
- Grand Rapids/Muskegon
- Saginaw/Flint
- Lansing/Jackson



Covering 95% of at risk population

Newborn Screening

We serve as the central coordinating center for hemoglobinopathy screening for the MDCH

Cases referred from Lansing

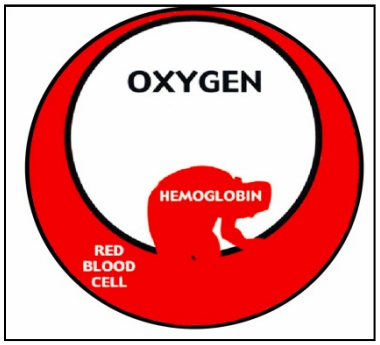
Disease

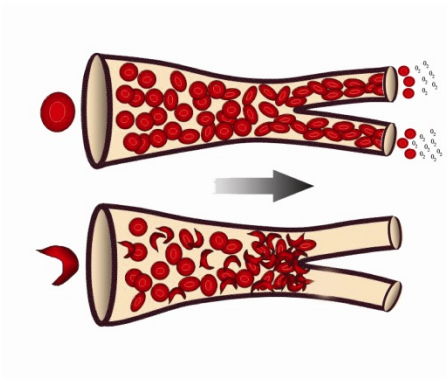
- Initial contact with family
- Disease education sessions
- Repeat test
- Penicillin Initiation
- Monitor Adherence

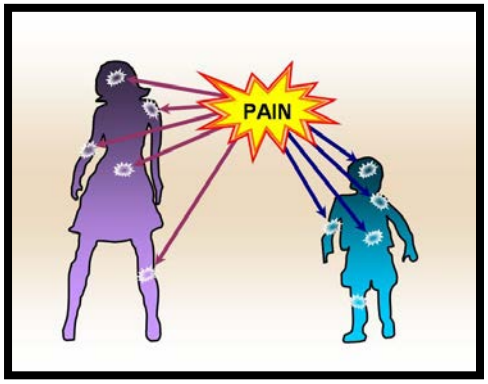
Sickle Cell Trait

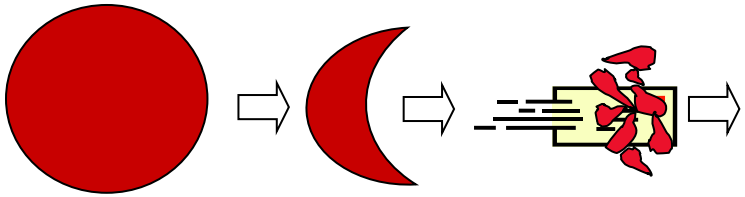
- Letter
- Phone follow-up

Education



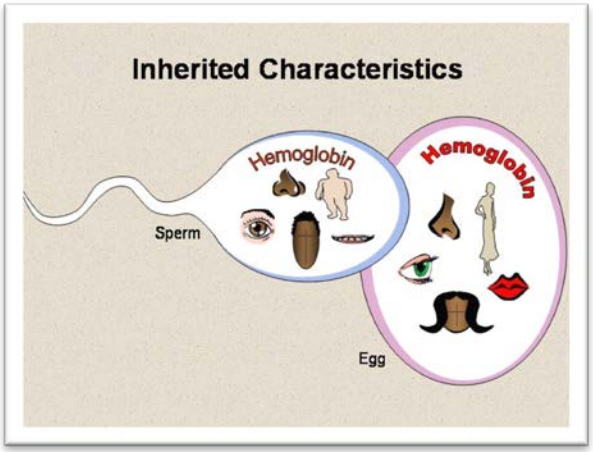


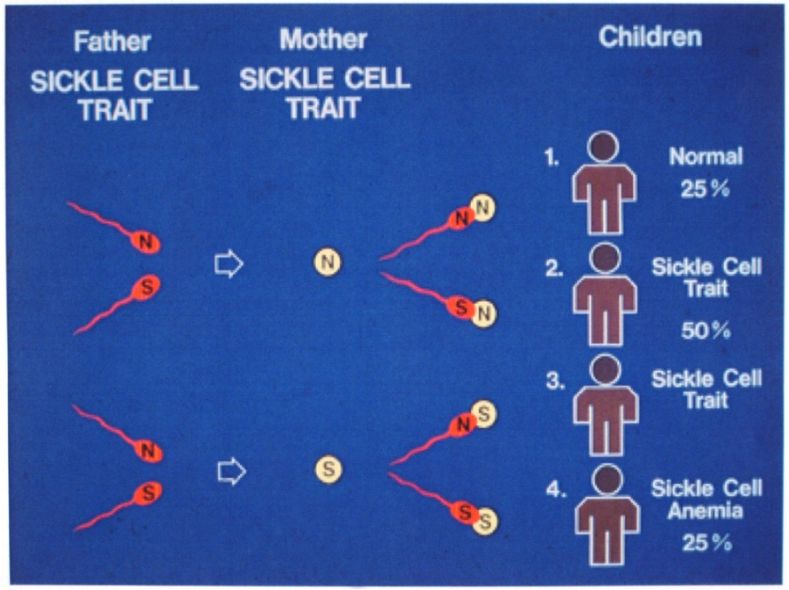


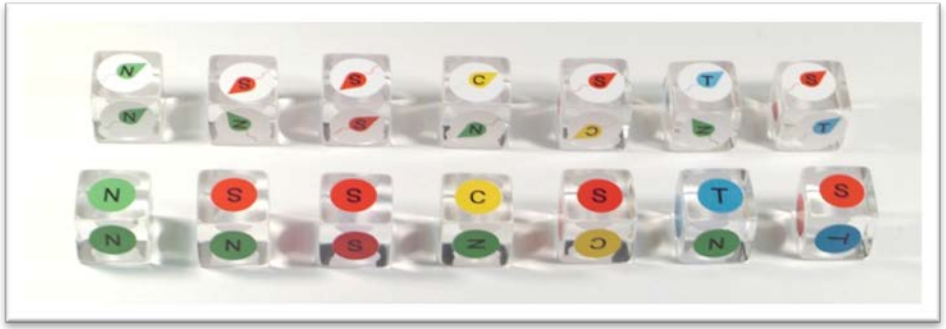


ANEMIA
Fatigue

Education (cont)







Informed decisions about future childbearing

Education (cont)

Medical Assistance

- Help clients find doctors
- Help obtain medical insurance (i.e. Medicaid & CSHCS)

Basic Needs

- Assist with emergency needs:
  - Shut off notices
  - Evictions
  - Food
  - Clothing

ADVOCACY

- Attend Individualized Education Program (IEP) planning or 504 Plans in schools
- Communicate with teachers
- Hospital visits
- Referrals (i.e. Support groups, SSI/SSD, WIC, Early On)

Community Awareness/Education

- Health fairs
- Presentations
- Trait counseling and education for families

Assessments

- Completing psychosocial assessments
- Health Status Assessments (help identify needs or concerns that the patient may have as well as identifying the successes)

Annual Summer Camp

- Sponsor children with SCD
- Gives children an opportunity to be around other children experiencing the same challenges
- Provides a unique opportunity for children to gain self confidence and independence
- Helps parents overcome issues of over protectiveness which may occur when raising a child with a chronic illness

Awareness Walk

Sickle Cell Disease Association of America, Michigan Chapter

Presents a

SICKLE CELL

AWARENESS & MYTH DISPELLING

WALK

Saturday, September 27, 2014

Registration at 8:00 am Walk begins at 9:00 am *SHARP!*

8450 W. 10 Mile Rd., Royal Oak, MI 48067

$25.00 Pre– Registration at sicklecellwalkzoo.org

Includes: T shirt & Admission to Zoo

For more info contact SCDAA at: (800) 842-0973

Awareness Walk cont.

Myth Dispelling

Instilling Hope

Every child is different

Discourage overprotection

Hertz Nazaire
Haitian Artist

In Summary

- **Newborn screening** is an important Michigan public health program that benefits babies with rare but serious disorders
- **Sickle cell disease** is a challenging illness that consists of daily pain, time consuming trips to Emergency Departments, stigma and a compromised quality of life.
- **Public health programs** play a critical role in positive health outcomes for families impacted by sickle cell disease.

Michigan Department of Community Health

Thank you for your time and we welcome your questions☺

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