west central ohio comprehensive sickle cell center/ohio newborn screening program for hemoglobinopathies

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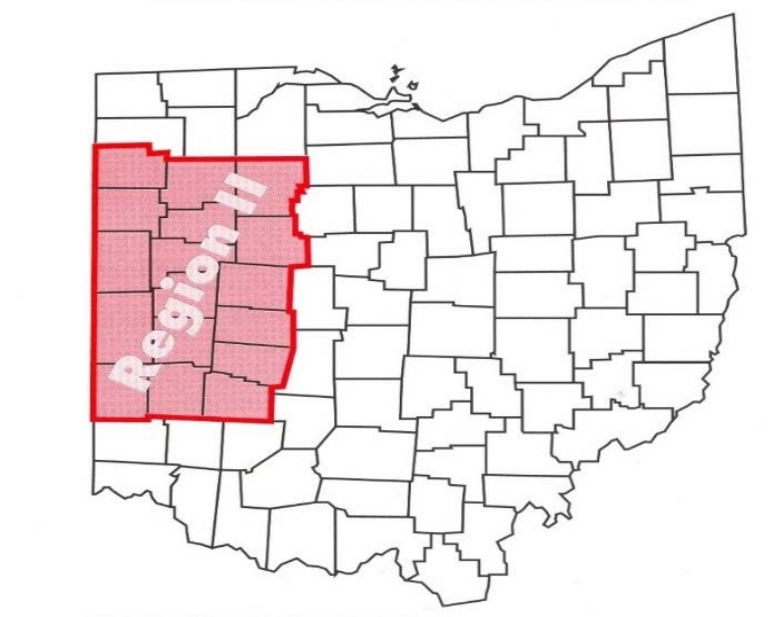
ohio regional sickle cell projects (ORSCP)

- Six Regional Sickle Cell Projects throughout the state of Ohio (covers all 88 counties in Ohio)
- Each RSCP is funded by a grant from the Ohio Department of Health (ODH) Sickle Cell Services Program/Maternal, Child and Family Health

region II sickle cell project



Region II is located in the Hematology (blood) & Oncology (cancer) Department at Dayton Children's Hospital

Seventeen counties – **Allen**, Auglaize, Champaign, **Clark**, Darke, **Greene**, Hancock, Hardin, Logan, Mercer, Miami, **Montgomery**, Paulding, Preble, Putnam, Shelby, Van Wert

goals of ORSCP

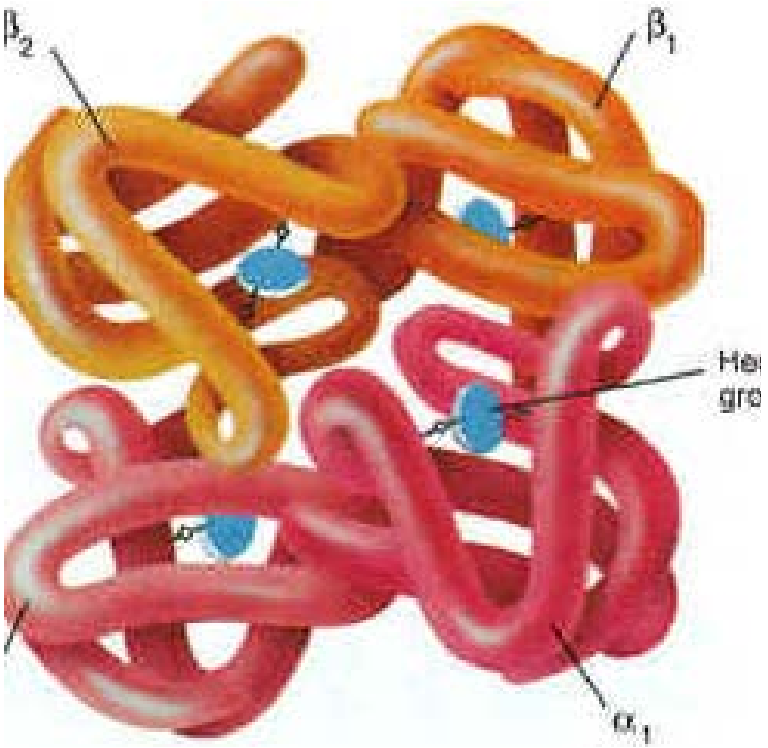
- Early identification of children with sickle cell disease and related hemoglobinopathies and integration into systems of services and care
- Increase awareness, knowledge, and skill level of Ohio's health care professionals' and providers' about the special health care needs related to sickle cell disease, sickle cell trait, and related hemoglobinopathies
- Expand public and community awareness of sickle cell disorders, traits, and available programs/services

what are hemoglobinopathies?

- Include **sickle cell disease** (SCD), **sickle cell trait** (SCT) and thalassemias
- Some can cause life-threatening symptoms, while others do not cause medical problems or even signs of the condition
- Some may require no medical treatment
- Severe cases that are left untreated can cause a shortage of red blood cells (anemia), organ damage, or other serious complications

what is hemoglobin?

- A protein substance found in the red blood cells
  - Helps carry oxygen throughout the body
  - Molecule made up of alpha and beta globin
- Defined by letters of the alphabet, people and places
  - Over 800 different types
  - Everyone has a hemoglobin type (different from blood type)
  - Inherit one gene from each parent

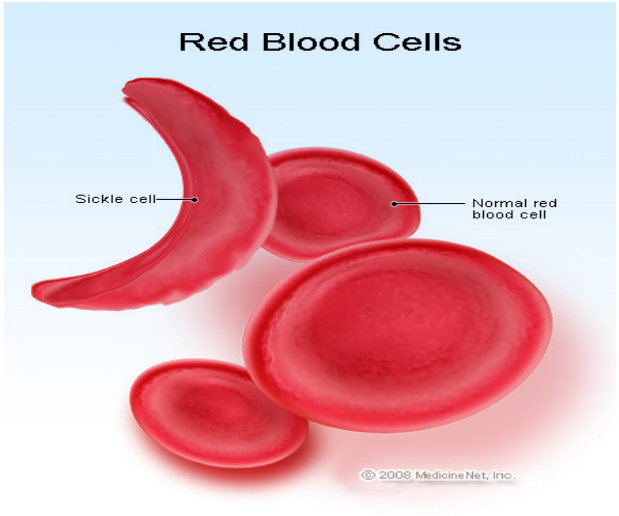


a closer look at some common hemoglobin (Hb) types

- Normal Hemoglobin (Adult):
  Hb A
- Fetal Hemoglobin:
  Hb F
- Most common types of SCD include:
  Hb SS (Sickle cell disease)
  Hb SC (Hb SC disease)
  Hb Sickle Beta +/0 Thalassemia
- Rare types of SCD include:
  Hb SD
  Hb SE
  Hb SO

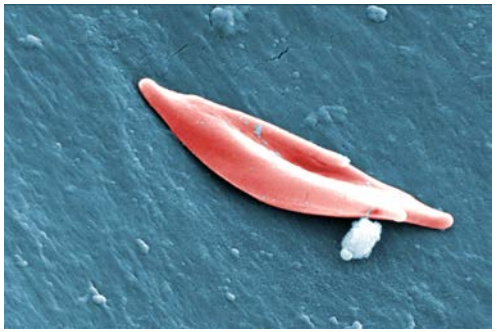
what is sickle cell trait?

- A “carrier” state when a person inherits one **abnormal** gene (“S”) from one parent and one **normal** gene (“A”) from the other parent



what is sickle cell disease?

- Most common inherited blood disorder in the U.S.
- A term used to describe a group of disorders that affect the hemoglobin in the red blood cells
- Red blood cells produced of an abnormal type of hemoglobin called hemoglobin S that changes the structure of the red blood cells (RBC)
- A life-long condition, characterized by pain and does not have a universal cure



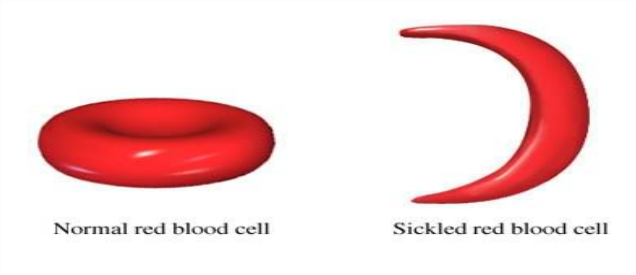
let’s compare

Hemoglobin A (normal hemoglobin)

- Lifespan of 120 days
- Round, soft, pliable
- Flows easily through blood vessels

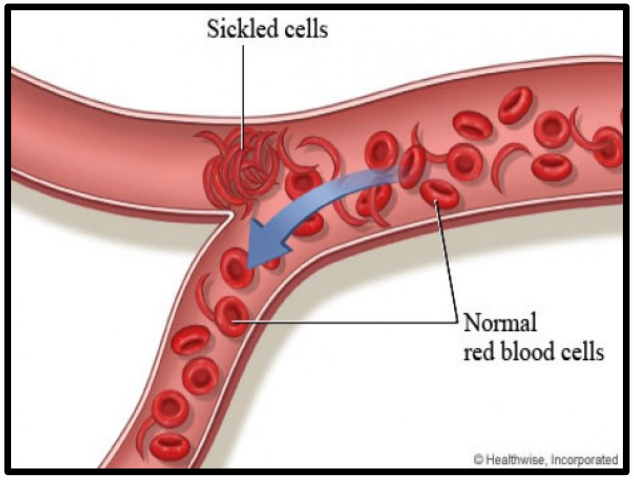
Hemoglobin S (abnormal sickle hemoglobin)

- Lifespan of 14 days
- Jagged, stiff, sticky
- Tend to clump together, occlude blood flow



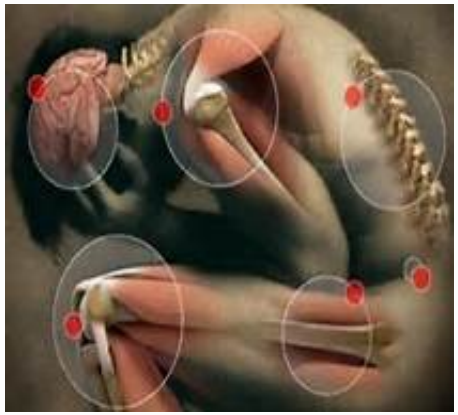
what EXACTLY happens?

- Mutation forms abnormal hemoglobin molecules
- After releasing oxygen, sickled red blood cells clump together, block blood vessels, and decrease blood flow
- Decreased blood flow damages tissue, blood vessels, organs, and bone



common complications

- Pain episode “crises”
- Infection/fever
- Acute chest syndrome (ACS)
- Anemia
- Gallbladder issues
- Splenic sequestration
- Stroke
- Priapism (males only)
- Avascular necrosis (AVN)
- Leg/ankle ulcers (mostly in adults)
- Delayed growth and development
- Psychosocial issues



the sickle cell iceberg

Sickle Cell Disease

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what can trigger a sickling crisis?

- Dehydration
- Temperature extremes
- Low oxygen
- Infection
- Strenuous activity
- Emotional stress
- ???

management of scd

- Preventive management
  - Prophylactic antibiotics
  - Vaccines
  - Hydration
  - Daily medication
    - ✓ Folic acid
    - ✓ Hydroxyurea
    - ✓ L-glutamine/Endari

- Management of complications
  - Pain management
  - Blood transfusion
  - Surgery
  - Mental health resources

who is at-risk for scd?

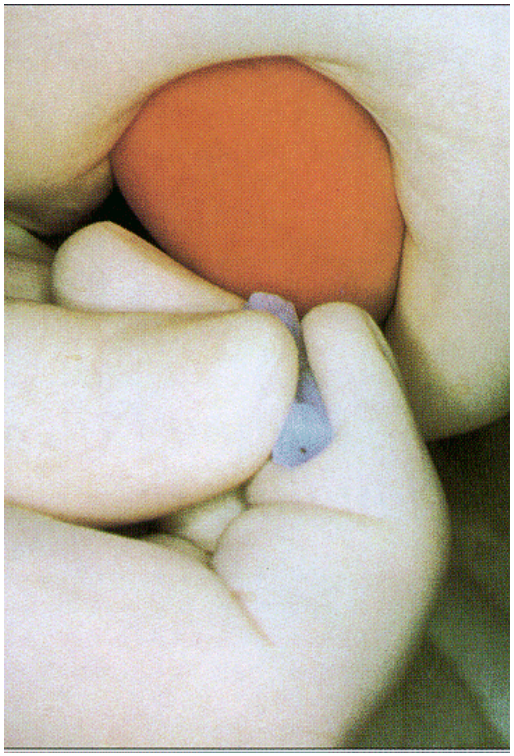
- Anyone
  - According to the Centers for Disease Control and Prevention (CDC), approximately 100,000 Americans are affected by SCD in the U.S.
  - Affects millions of people worldwide
  - More common in people with heritage

    from Africa, Asia, Europe, Mediterranean areas (Turkey, Greece, Italy), Middle East, Central and South America
- Family history of sickle cell trait/disease and/or thalassemia

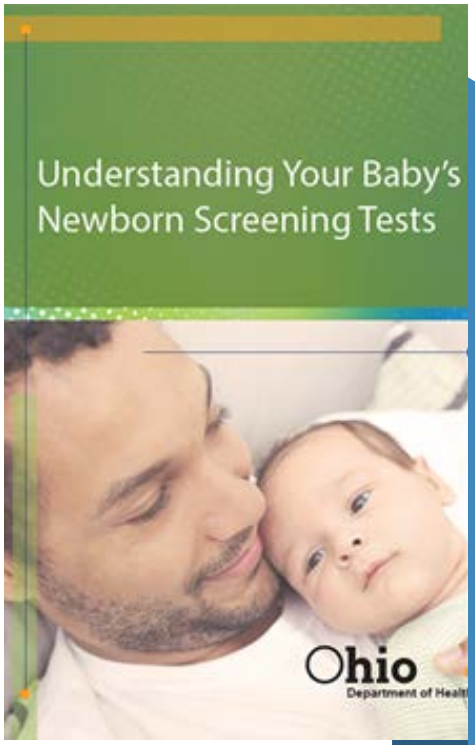
importance of the Ohio NBS program for hemoglobinopathies

- Identify newborn babies who may be at risk for rare but serious hemoglobinopathies (hemoglobin disorders)
- If problems are found early, treatment may help prevent serious problems or, in some cases, death
- Does not diagnose illnesses but shows which babies require more testing to confirm or rule out disease or illness
- Critical for the baby to receive treatment (treatment may be lifesaving); however, not all disorders can be treated

ohio newborn screening program for hemoglobin disorders



- Ohio currently mandates screening of all newborns for over 40 genetic disorders
    - Hemoglobinopathies were added to NBS panel in March of 1990
    - Sample collected at about 24-48 hours of life but no later than 5 days of age
    - Screenings performed on dried heel stick blood specimen collected at birth hospital
    - All newborns are tested regardless of race/ethnicity



understanding your baby's newborn screening tests

importance of hemoglobin counseling

- To eliminate or reduce parents' anxiety
- To provide parents with education and understanding about their child's hemoglobin results
- To provide accurate information and clear up any misconceptions or preconceived notions
- To offer parents an option of having a free hemoglobin screening test done if unaware of hemoglobin status

Ohio Department of Health Newborn Screening Program
Diagnosis and Case Disposition Form

The Newborn Screen Results on this patient were positive for a possible hemoglobin disorder

NBS Results ☐

Baby's Birth Name:	**Mother's Name:**	**Doctor's Name:**		**Birth Hospital Name:**
DOB:				
Baby's AKA				
MedRec No.				
Sex:				
Weight:				
Transfusion:				
Race:	**County**	**Kit #**	**Time collected**	**Region**
				RSCP fax: (937) 641- 5885

Disposition Information: (Check all that apply)

- ☐ Patient seen by Regional Sickle Cell Project (RSCP)- Project Name: _____
  Services provided by RSCP: _____Diagnostic Evaluation _____Hemoglobin Counseling _____Treatment* _____Other:
- ☐ Patient seen by Non-RSCP provider- Provider Name: _____ Provider Phone: ( )_____
  Services provided by Non-RSCP: _____Diagnostic Evaluation _____Hemoglobin Counseling _____Treatment* _____Other
- ☐ Family Declines: _____Referral to Sickle Cell Project _____Diagnostic Evaluation _____Hemoglobin Counseling _____Treatment
- ☐ Patient has been lost to follow-up (check all that apply): ____Moved out of State _____No current address/phone _____ No Provider
  No Response to RSCP contact _____Other: _____
- ☐ Case referred to local health department (LHD) or Child Protected Services (CPS)- Name of agency _____
- ☐ Patient Expired. Date of death _____

Diagnostic and Treatment Information

☐ Initial Submission	☐ Updated Submission
☐ FS	☐ FSA Beta thal	☐ FAS
☐ FSC	☐ FA+Barts (Alpha Thal)	☐ FAC
☐ FC	☐ FA+ other Hb	☐ FAE
☐ FE	☐ FA (normal)	☐ FAD
	☐ AF	☐ FAG

Other Diagnosis not listed above: _____ Date of Confirmatory Testing: _____

Lab Method: ☐ Hemoglobin Electrophoresis ☐ HPLC ☐ Isoelectric Focusing (IEF)

* Prophylactic antibiotics initiated: ☐ Yes (Date initiated) _____ ☐ No ☐ Not Applicable

Person Completing Form: _____ Date: _____ Phone: ( ) _____

Fax this completed form and confirmatory lab report to RSCP fax (937) 641-5885

ohio newborn screening program rules

Complete information on the Ohio Administrative Code (OAC) Rules governing the NBS Program may be obtained at www.odh.ohio.gov/rules/final/3701-55.aspx

or

Ohio Department of Health Bureau of Public Health Labs Newborn Screening Program

8995 East Main Street, Building 22

Reynoldsburg, OH 43068-3342

Telephone (888)-ODH-LABS

Fax (614) 644-4648

physician guidelines for hemoglobin follow-up and testing

- **Newborn Screening results should be COMMUNICATED** to the parents as soon as possible
  - Parents should ask about these results during the baby's first health check-up
  - The initial Newborn Screening result should be **CONFIRMED** within 2 months of age by performing a Hemoglobin Electrophoresis

An abnormal newborn screening result does not always mean the baby has a disorder

physician guidelines for hemoglobin follow-up and testing (cont.)

- **Parents should be NOTIFIED and INFORMED** by the Provider of Record (POR) of the confirmatory test results
- The reporting of the confirmatory result…
    - Will lessen parents' anxiety
    - Will help lessen the duplication of testing
    - Is necessary for patient awareness and reproductive decision-making
    - **Remember, babies born in Ohio before to March 1990 were not tested for hemoglobinopathies**

physician guidelines for hemoglobin follow-up and testing (cont.)

The physician must PROVIDE and **DOCUMENT** hemoglobin counseling services

- Simply informing the patient/family of the diagnosis is **NOT** sufficient
- Detailed counseling is necessary to educate the family about the specific hemoglobin variant, genetic implications for future pregnancies and other related concerns
- For liability reasons, all hemoglobin counseling efforts should be documented in the patient's medical chart

physician guidelines for confirmatory testing and follow-up (cont.)

- **The POR must SEND** documentation of the FINAL DIAGNOSIS (confirmatory testing), laboratory reports, consults and treatment information (if applicable) to the Cynthia Moon, Project Director/NBS Coordinator at 937-641-5885 (fax) for case closure
- **The Ohio Department of Health requires that all NBS cases be closed within 4 months from the infant's date of birth**

family planning

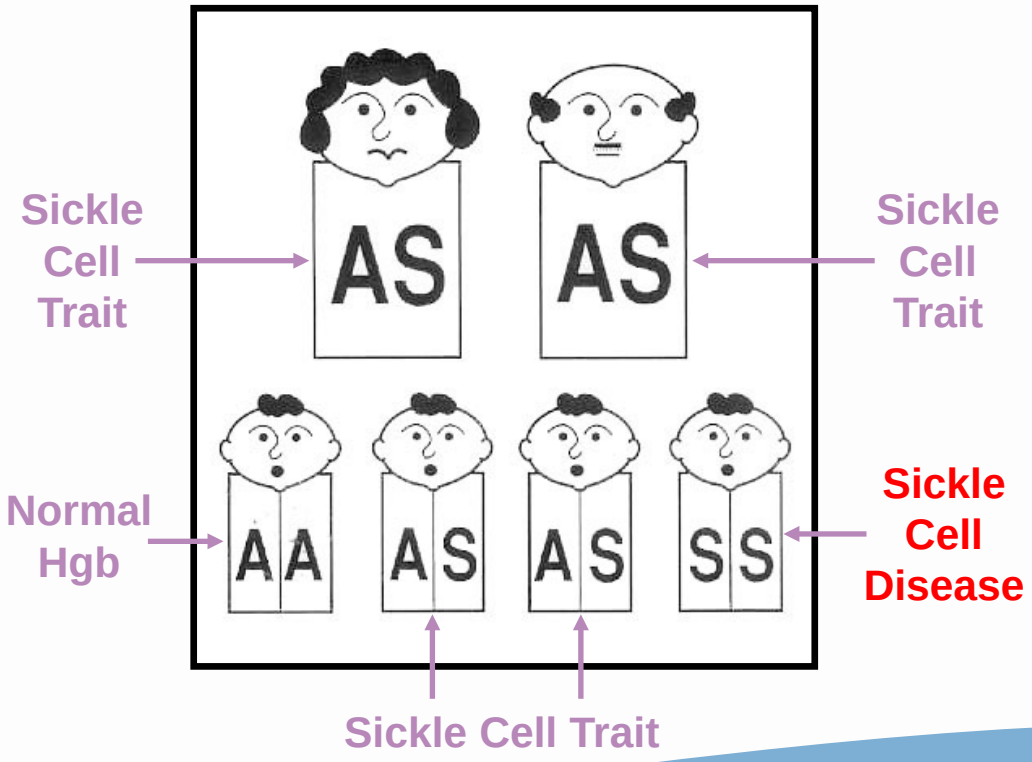
- It is very important for couples to know their hemoglobin type before having children
- Remember, screening for hemoglobinopathies began in March 1990, so there is a population born before 1990 who may not know if they carry the gene for sickle cell
- If both parents have sickle cell trait, there is a 25% chance with each pregnancy of having a baby born with sickle cell disease

sickle cell disease and pregnancy

- Before pregnancy:
  - Partner should be tested for SCT
  - Meet with genetic counselor to discuss the risks to their children and provide further information

- During pregnancy:
  - Prenatal testing can be done
  - Chorionic villus sampling (CVS) and amniocentesis
  - Usually conducted after the second month of pregnancy

inheritance pattern



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cures for sickle cell disease

- Bone Marrow Transplant (BMT)
  - Donor match (usually a sibling)
  - Chemotherapy is needed to destroy patient's bone marrow
  - Very risky (graft vs. host disease)
  - Very expensive

- Experimental cure underway
- Gene therapy
  - No donor needed
  - Chemotherapy is needed to destroy patient's bone marrow
  - Normal hemoglobin gene introduced into patient's own bone marrow cells
  - New marrow replaces old marrow (sickled cells disappear)

references

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