

# Sickle Cell Trait

Terianne Lindsey, MSN, RN, CPNP  
Missouri Newborn Screening Coordinator  
Sickle Cell Disease Nurse Practitioner  
Washington University School Medicine

# What is Sickle Cell Trait?

- An inherited condition in which the individual is carrying both A and S genes in the red blood cells called hemoglobin
- Hemoglobin is the pigment responsible for the red color of the blood
- Normal hemoglobin is referred to as hemoglobin A
- Sickled hemoglobin is referred to as hemoglobin S

# Sickle Cell Trait

- The type of hemoglobin you have present in your blood cells was inherited from your parents
- All of a persons features are inherited and are passed to the individual from parents by genes
- Genes are bits of information that code what you look like
- One gene comes from each parent

# Sickle Cell Trait

- Trait is a common word for a condition when a person inherits an abnormal gene from one parent and a normal gene from the other parent
- If a person gets an abnormal gene from both parents, it is called disease
- The individual with trait will have more of the normal gene present than the abnormal gene

# Sickle Cell Trait

- A person with SCT can pass either normal hemoglobin (A) or sickle hemoglobin (S) to a child
- If two people with SCT pass on sickle hemoglobin to a child, the child will have SCA
- There is a 1 in 4 chance with each pregnancy of two people with SCT will have a child with SCA

# Sickle Cell Trait

- In planning a family, it is wise to ask your partner to be tested
- Only if both parents are tested can they know exactly what kind of hemoglobin their children could have
- They should look at inheritance patterns for the possibilities of having a child with SCD
- A counselor can tell them if any of their future children could have a form of SCD

# Hemoglobinopathy Carriers Identified by Neonatal Screening

## Screening Results

- FAS
- FAC
- FAE
- FA Other

## Possible Condition

Sickle cell trait

Hb C carrier

Hb E carrier

Other Hb variant carrier

# **GENETIC COUNSELING**

# What is Genetic Counseling?

- A communication process between health professionals and family members that begins as soon as a genetic problem is identified
    - Diagnosis
    - Information
    - Support
    - Follow-up counseling
- (Williams, Pediatric Nursing, 1986)

# Purpose of genetic counselors

- Help the individual or family
  - Comprehend the medical facts, including the diagnosis, probable course of the disorder, and available management options
  - Understand how heredity contributes to the disorder and the recurrence risks

(Williams, Pediatric Nursing, 1986)

# Purpose of genetic counselors

- Understand the options for dealing with the recurrence risks
- Choose the best course of action for the family
- Learn to cope with an affected family member's disorder and the recurrence risks

(Williams, Pediatric Nursing, 1986)

# Genetic Counseling

- Education
  - Enable individuals to make informed decisions, in their own interest, about future family planning
- Decision-making
  - Focus on education and informed decisions, about current pregnancy
- Advice, personal opinions, and societal positions must not be given or implied

# Parent's Options

- Avoid pregnancy completely
  - Contraceptives
    - Condoms
    - Foams
  - Sterilization
    - Vasectomy
    - Tubal ligation

# Parent's Options

- Request pre-implantation genetic diagnosis
  - DNA analysis of one of the cells after division to test for SCD
  - If genetically normal, implant into mother to complete pregnancy

# Detection

- Amniocentesis
  - Sixteen to eighteen weeks
  - Results 2-3 weeks
- Chorionic villus sampling
  - Twelve to fourteen weeks
  - Results within 48 hours

# Genetic Counseling

- The family planning options open to persons with SCT
- SCT is not an illness, so no restrictions need to be placed on his or her activities
- The variability in severity of SCD
- Both parents must have the trait for the child to have SCD
- The 25 percent chance that each pregnancy will result in a child with SCD if both parents have the trait

# Genetic Counseling

- SCT trait is not considered to be a health problem
- Individuals who test positive should be informed about implications for their health and family planning
- Despite mandatory NBS programs implemented in most states, children with SCT may not recall or understand the implications by the time they reach childbearing age

# **Inadequate Community Knowledge about Sickle Cell Disease among African-American Women**

Boyd JH, Watkins AR, Price CL, Fleming  
F, DeBaun MR

Journal of the National Medical  
Association January 2005

# Background

- Sickle cell disease (SCD) is the most common genetically inherited disease affecting African Americans (AA)
- Approximately one in 12 AA will have sickle cell trait (SCT), and approximately one in 400 AA newborns will be diagnosed with SCD annually

# Background

- Prior to the 1970's, few programs were dedicated to providing the public with information on SCD
- Early community-based surveys conducted in large urban areas demonstrated limited awareness of SCD among AA in these communities

# Background

- In 1972, the National SCD Control Act set the foundation for universal screening of newborns for SCD
- The national legislation promoted community awareness of SCD by providing education and genetic counseling programs to patients and families with SCT and SCD
- The aim of this study was to assess existing knowledge about SCD among AA women in St. Louis, MO

# Methods

- A cross-sectional telephone survey of AA women
- Age 18-30 years
- Recruited through random-digit dialing in six zip codes
- Greater than 75% AA residents

# Methods

- Questions asked exploring four content domains about SCD
  - General knowledge
  - Genetics
  - Management
  - Educational resources

# Results

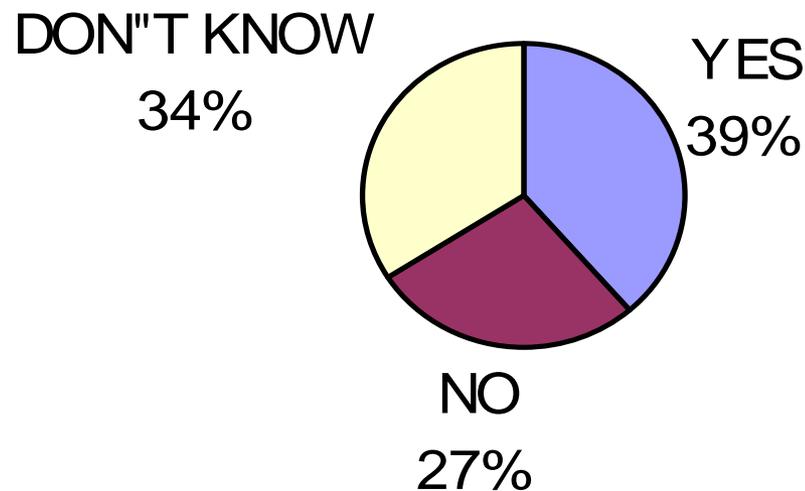
- Total 264 women contacted
- 30% unable to participate because unaware of SCD
- 162 included in study

# Results

- 91% believed SCD was hereditary
- 9.3% understood inheritance pattern
- 11% unaware of their SCT status
- Most recognized complications of SCD
  - pain (94%)
  - infection (80%)
  - strokes (40%)

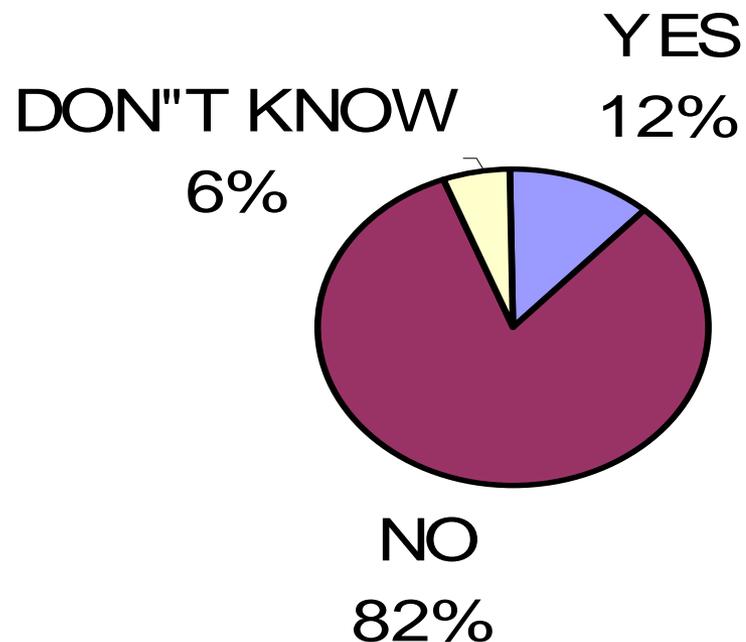
# Knowledge of Phenotypes Associated with Sickle Cell Disease in 162 African American Women between 18 and 30 Years of Age living in St. Louis

**Do you know if there are different types of traits that can lead to sickle cell disease?**



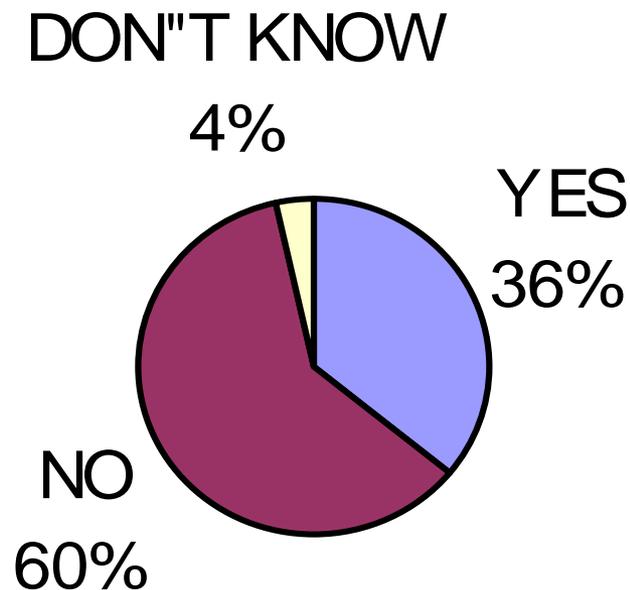
# Knowledge of Phenotypes Associated with Sickle Cell Disease in 162 African American Women between 18 and 30 Years of Age living in St. Louis

Have you ever heard of  $\beta$ -thalassemia trait?



# Knowledge of Phenotypes Associated with Sickle Cell Disease in 162 African American Women between 18 and 30 Years of Age living in St. Louis

**Have you ever heard of C trait?**



# Conclusions

- New strategies are needed to enhance awareness of SCD among AA of childbearing age

# Prenatal Genetic Counseling for Hemoglobinopathy Carriers: A Comparison of Primary Providers of Prenatal Care and Professional Genetic Counselors

Rowley PT, Loader S, Sutera CJ,  
Kozyra A

American Journal of Human Genetics,  
1995

# Introduction

- Health personnel trained in medical genetics are insufficient to meet the demand for genetic services
- Methods must be found to enable primary care providers to offer commonly needed genetic services themselves

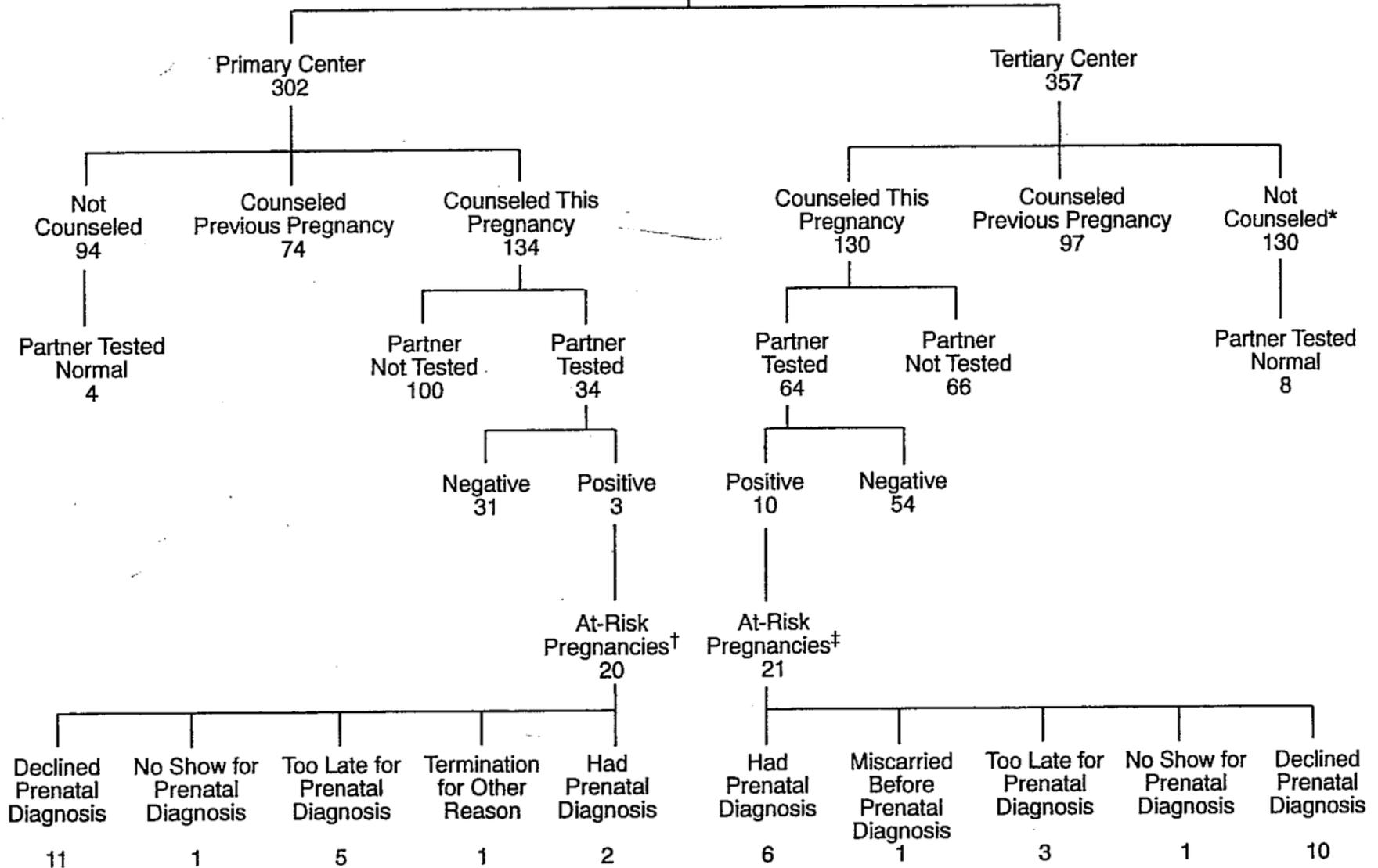
# Genetic Counseling

- Would prenatal screening for hemoglobinopathies be more efficient if counseling were given by primary prenatal care provider rather than a tertiary provider?
- Could primary providers be trained to offer genetic counseling as effectively as experienced genetic counselor?

(Rowley et. al., Am. J. Hum. Genet., 1995)

Pregnancies Screened  
32,988

Sickle Trait Pregnancies  
659

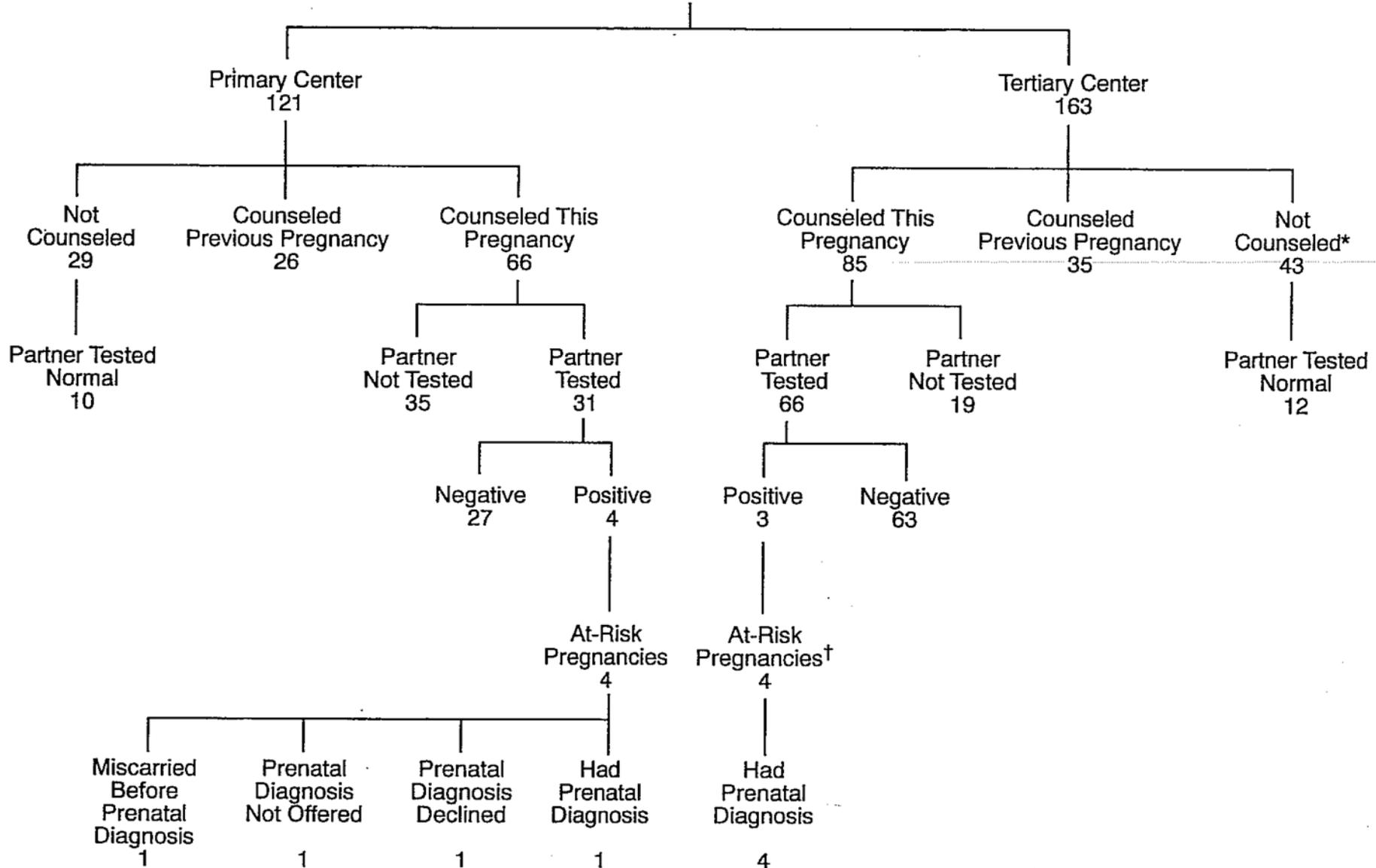


Pregnancies Screened

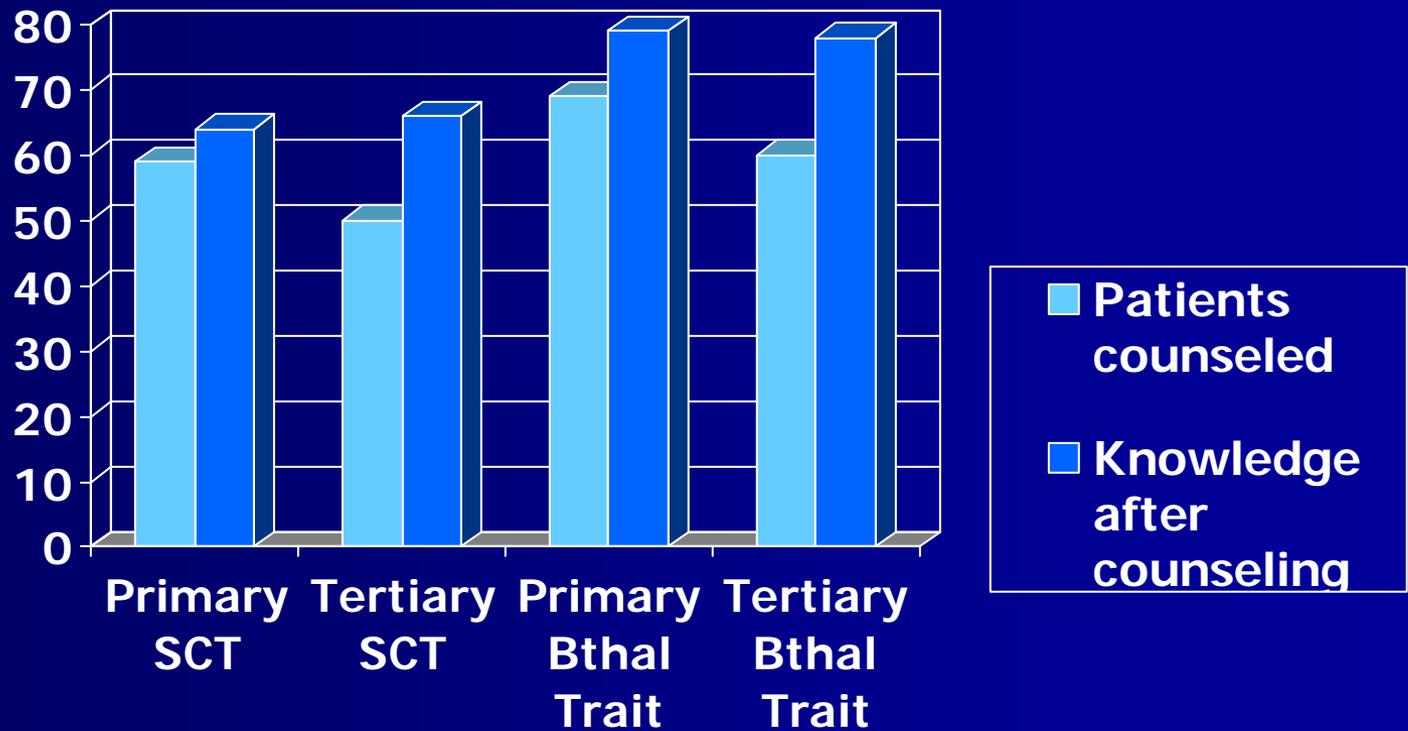
32,988

$\beta$ -Thalassemia Trait Pregnancies

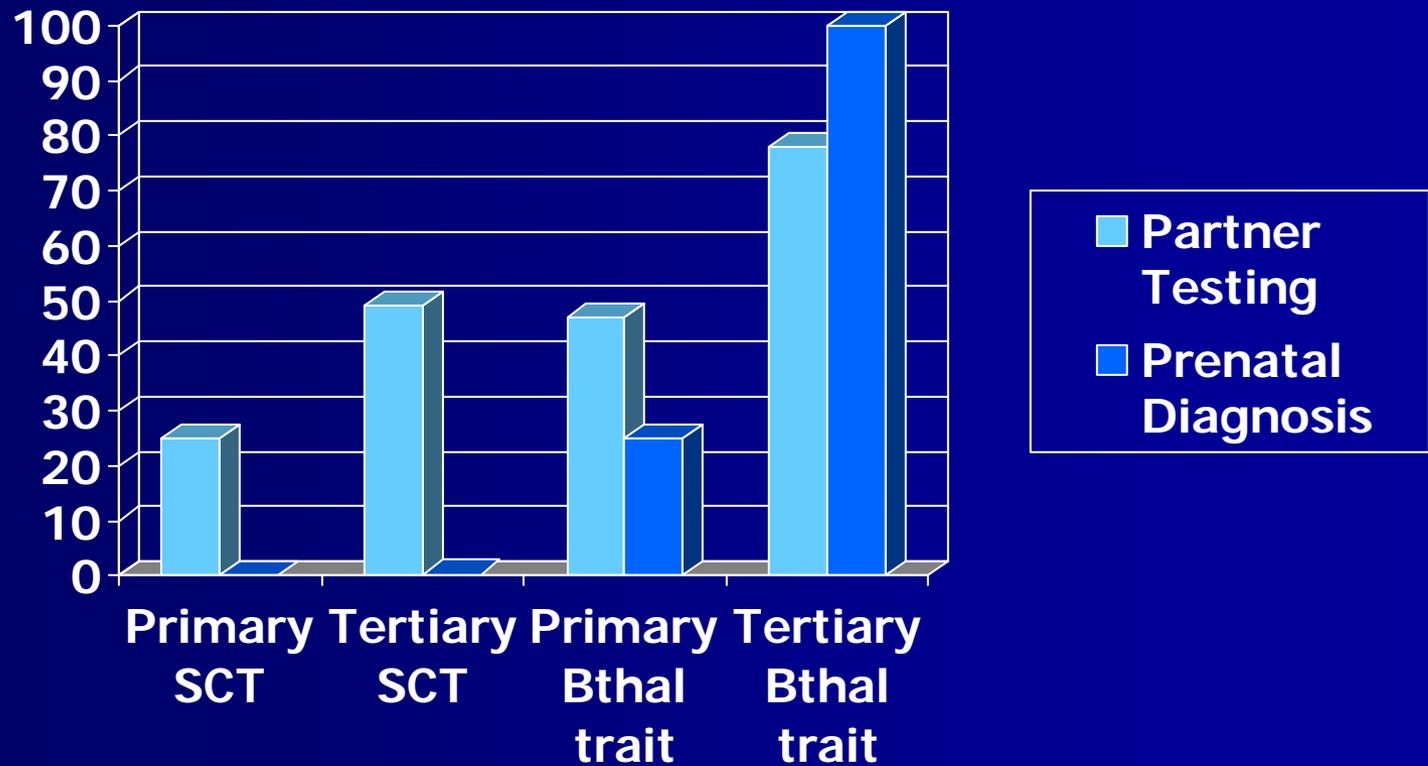
284



# Results



# Results



# A Community Based Sickle Cell Trait Educational Program

Allison King, MD, MPH

Washington University School of Medicine

Community Partners:

Myrtle Hilliard Davis Comprehensive Health  
Center

Prince Hall Masons of Missouri

# The Setting – St. Louis, MO

- Over 19,000 African Americans in the metro area carry sickle cell trait
  - S trait
  - C trait
  - Thal trait
- Genetic counseling is required for these traits
- No genetic counseling is available in our area

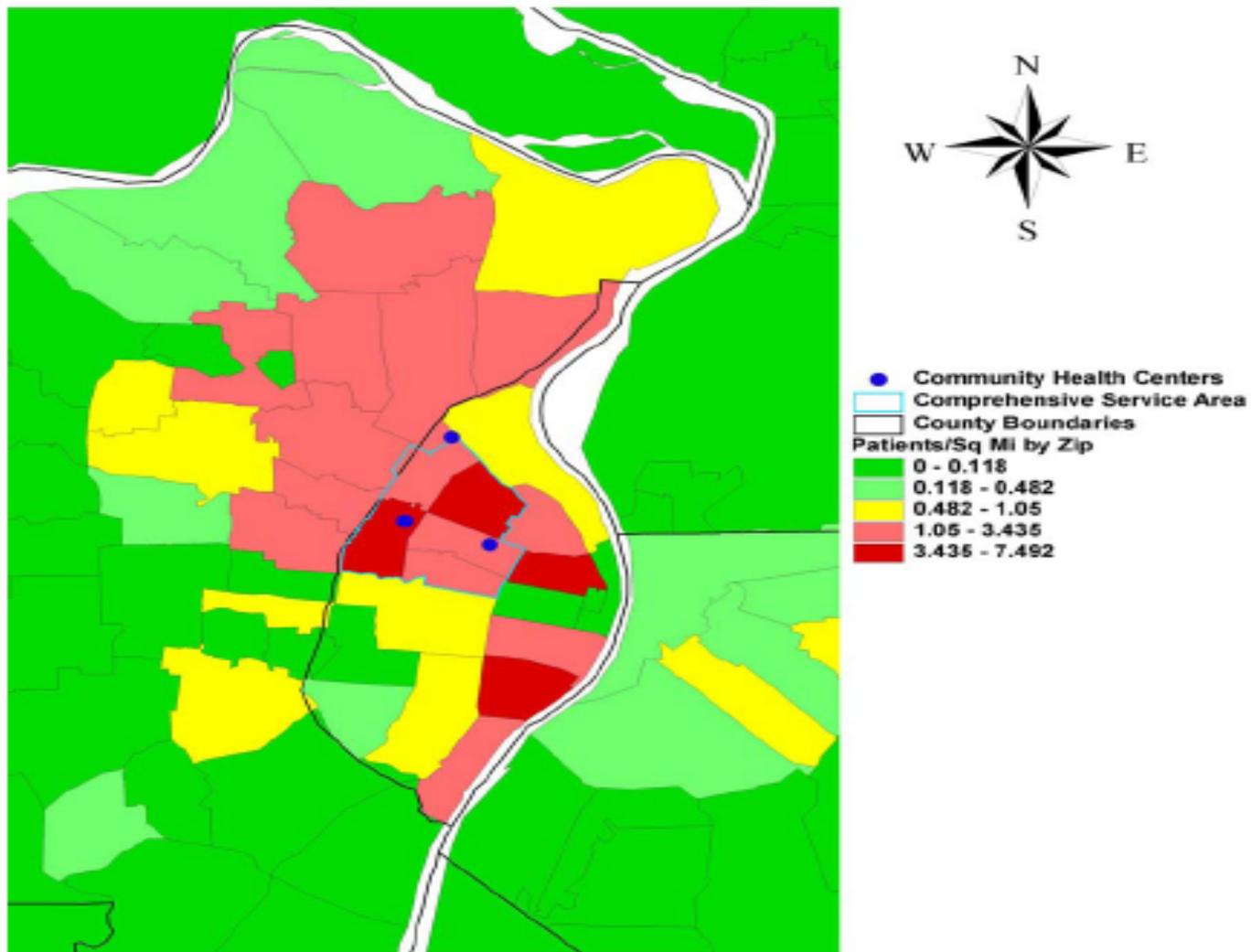
# Goals of Intervention

- To increase the number of young adults in the target population who:
  - have a general understanding of SCD
  - are aware of their sickle cell trait status
- To empower young adults with sickle cell trait about the risk of having a child with sickle cell disease

# Partners

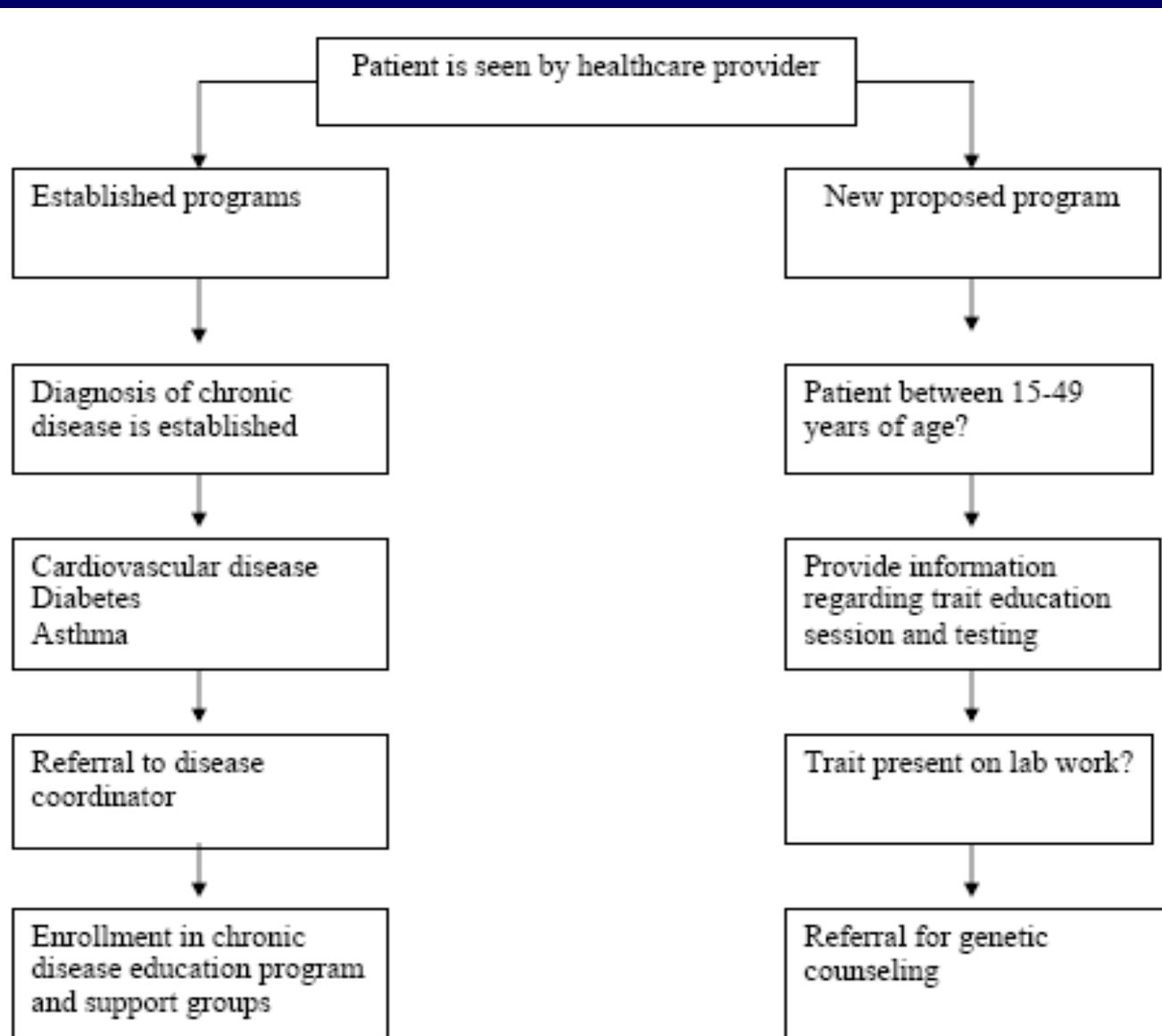
- Myrtle Hilliard Comprehensive Health Center
  - Federally funded public clinic system
  - Serves over 77,000 patients
    - Internal Medicine, Pediatrics, OB, Dental, Podiatry
- Prince Hall Masons of Missouri
  - Local Masons group
  - Donate funds and volunteer for St. Louis Children's Sickle Cell Disease Treatment Center
  - Michael Johnson
    - Team Chaplain
    - Adult with SC disease

# Sickle Cell Patient Density by Zip Code



10 0 10 Miles

# Concept



29,391 active patients in Comprehensive's clinic system



11,750 patients between the ages of 15-49 years



470 (~4%) patients will attend educational sessions  
(based on rate of participation in our previous experience with  
community based parent education programs)



376 (80%) patients will agree to have blood drawn for trait testing  
Patients provide primary and secondary contact information



Estimate 1:10 or 38 patients will test  
positive for sickle trait



RN-Genetic Counselor sends letter  
to home and calls to schedule genetic  
counseling session. Three attempts  
will be made to home and secondary  
contact.

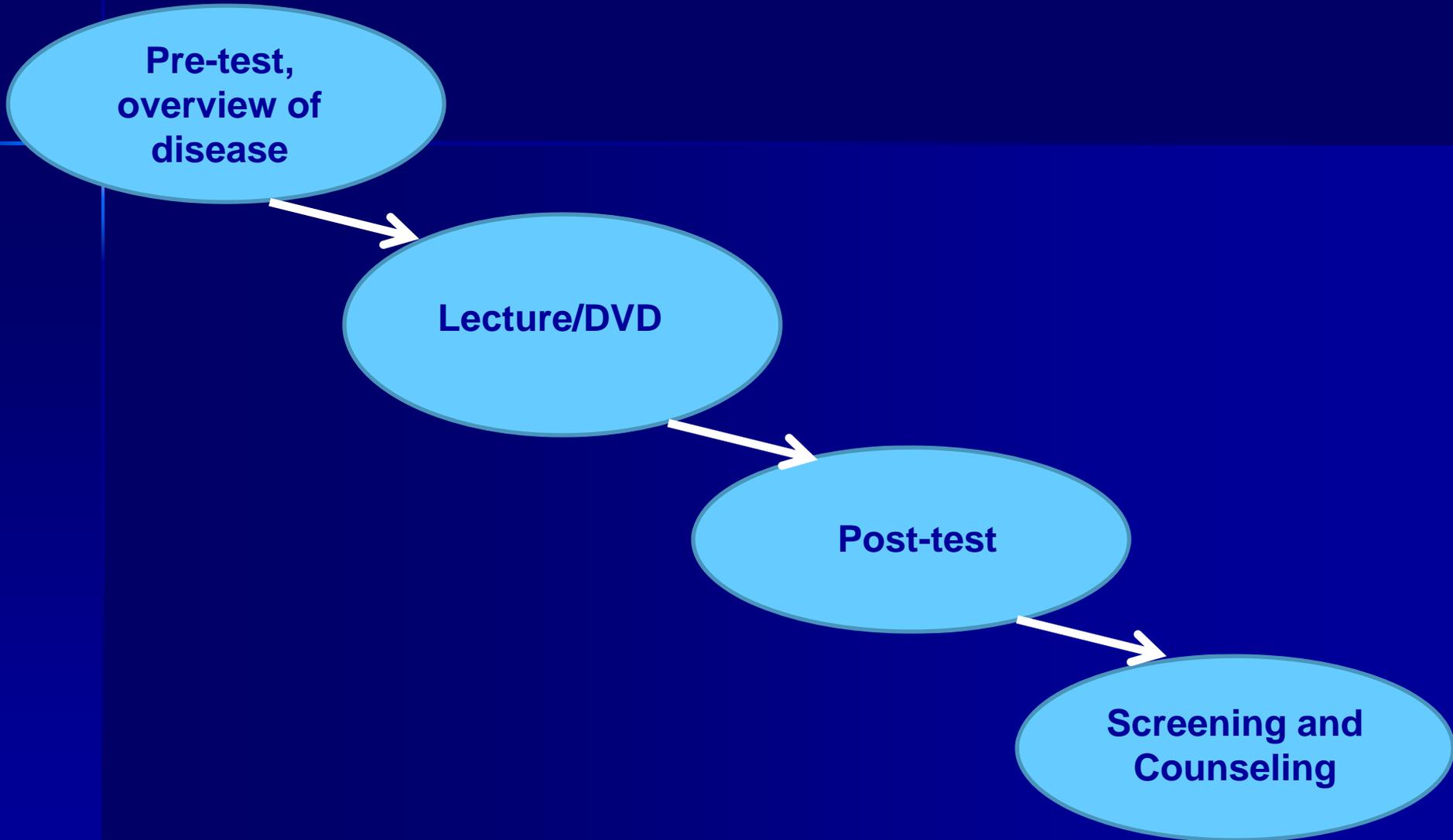


338 will test negative for trait



RN-Genetic Counselor sends  
letter to home and calls to inform

# Intervention



# Evaluation

| <b>Primary Intervention</b>  | <b>Primary Evaluation</b>   |
|--|---|
| Identify 15-49 yr olds<br>Invite for educational sessions<br>Offer trait testing | <ul style="list-style-type: none"><li>■ Proportion who attend sessions and have blood drawn for trait testing</li><li>■ Success = 50% or more</li></ul>               |
| Complete genetic counseling for individuals who have sickle cell trait           | <ul style="list-style-type: none"><li>■ Proportion of patients with sickle cell trait who attend genetic counseling session</li><li>■ Success = 50% or more</li></ul> |
| <b>Secondary Outcomes</b>  | <b>Secondary Assessments</b>  |
| Effectiveness of educational session   | <ul style="list-style-type: none"><li>■ Pre vs. post-tests</li><li>■ Satisfaction survey</li></ul>  |
| Effectiveness of genetic counseling session                                      | <ul style="list-style-type: none"><li>■ Pre vs. post-tests</li><li>■ Satisfaction survey (lecture vs. DVD)</li></ul>  |

