

# **SICKLE CARRIER DETECTION:**

***LESSONS LEARNED OVER 75 YEARS***

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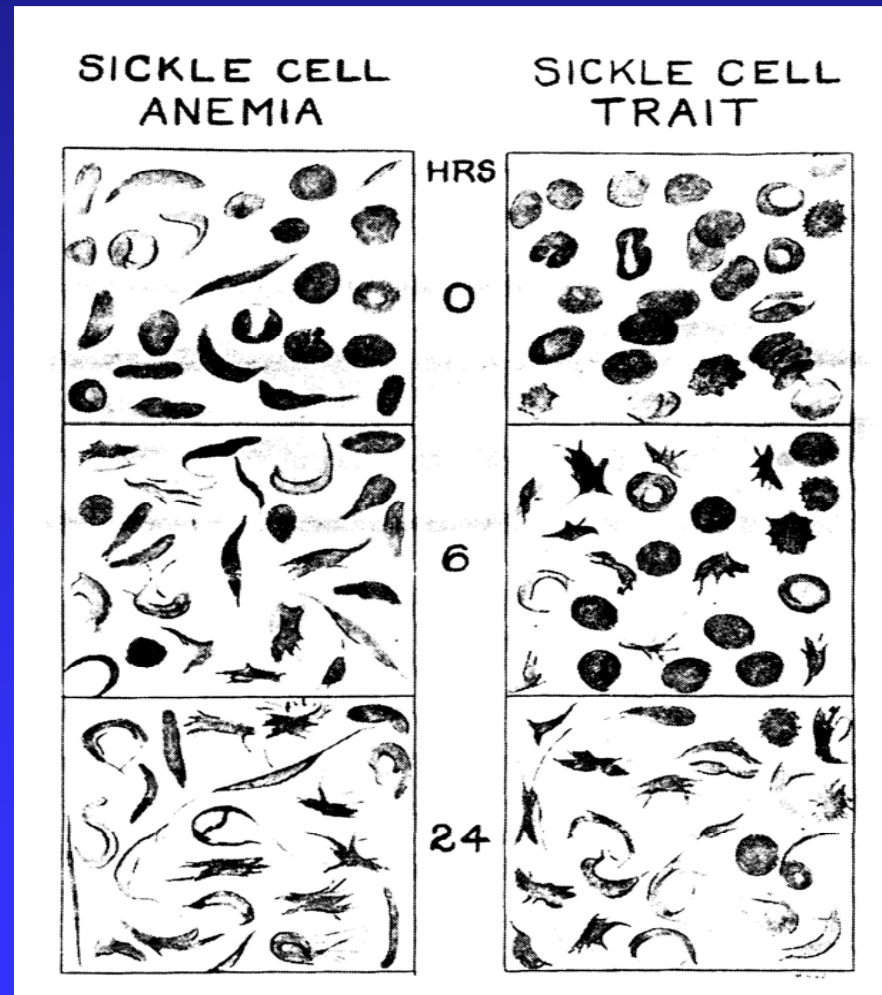
**Winship Cancer Institute**

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Grady Health System**

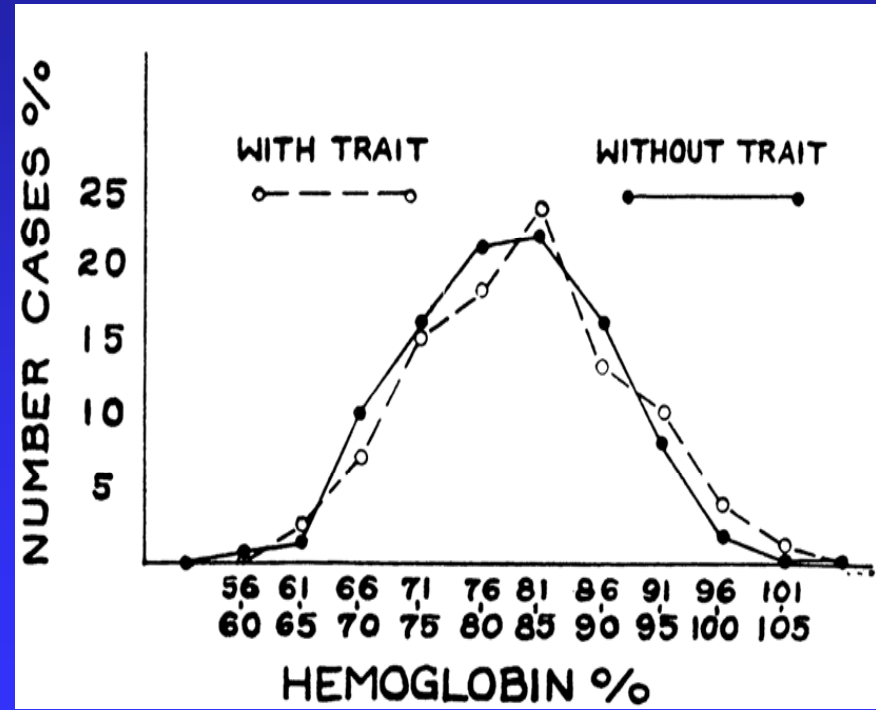
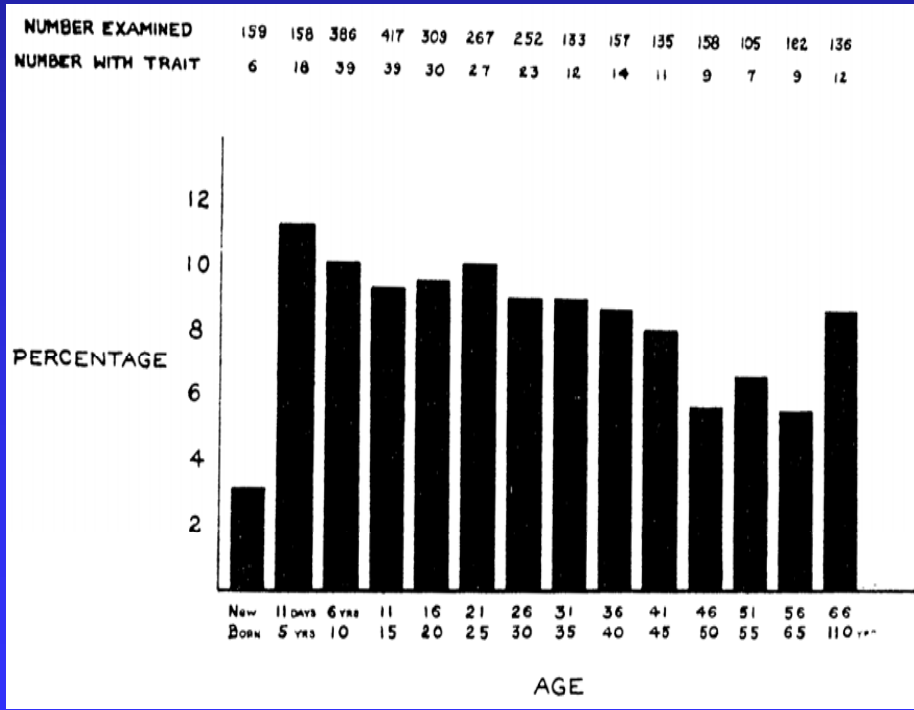
# SICKLE TRAIT

*Diggs Ann Intern Med 1933;7:769-778*



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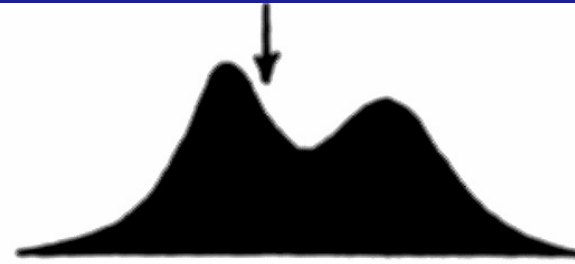


# SICKLE CELL AS A MOLECULAR DISEASE

Pauling L, Itano HA, Singer SJ. Science 110:543 - 548, 1949



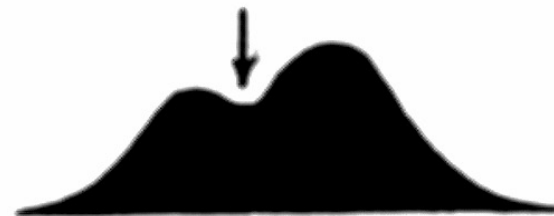
a) Normal



c) Sickle Cell Trait



b) Sickle Cell Anemia



d) 50-50 Mixture of a) and b)

# National Sickle Cell Anemia Control Act of 1972. Public Law 92-294

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- **Goals were to:**
  - Reduce morbidity and mortality from sickle cell disease
  - Increase awareness and educate the public
  - Develop new modes of therapy
- **Outcome**
  - NIH - Comprehensive Centers focused on research and treatment (15-10)
  - HRSA (HSA) - Sickle cell screening and education clinics focused on screening, education, information, counseling, and referral of disease
  - CDC – Training, proficiency testing, and reference laboratory

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Manley AF. Legislation and funding for sickle cell services, 1972-1982. AmerJ Pediatr Hemae/Onc 1984;6:67-71.

# **National Sickle Cell Anemia Control Act of 1972. Public Law 92-294**

**“Misguided but well-intentioned individuals began programs for mass screening with inadequate training and poor methodology, often without providing counseling of the persons who were being tested. Misguided legislators rushed to push for obligatory legislation mandating premarital and preschool testing for sickle cell anemia. A fair share of fraudulent benefit promoters and bogus organizations entered the field of rip-off profiteer, which led to the disease being called by some the “ghetto hustle”.”**

**Scott RB Historical review of legislative and national initiatives  
for sickle cell disease. Amer J Pediatr Heme/Onc 1983;5:346-351.**

# EARLY SICKLE CELL TRAIT SCREENING AND EDUCATION

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- **Neal-Cooper F, Scott RB. Genetic Counseling in Sickle Cell Anemia: Experiences with Couples at Risk. Pub Health Reports 1988;103:174-178.**
    - Little or no impact on reproductive behavior
    - “..since screening, education, and counseling have become available, a more positive attitude towards consideration of inherited illness, especially sickling can now be noted among blacks.”
  - **Bowman JE. Is a national program to prevent sickle cell disease possible? Amer J Pediatr Heme/Onc 1983;5:367-372.**
    - The only effective way to prevent sickle cell is prenatal diagnosis and selective abortion.
    - That other social and economic problems in the community should take precedent.
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# NEWBORN SCREENING SPREDS

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## NEWBORN SCREENING FOR SICKLE CELL DISEASE AND OTHER HEMOGLOBIN- OPATHIES

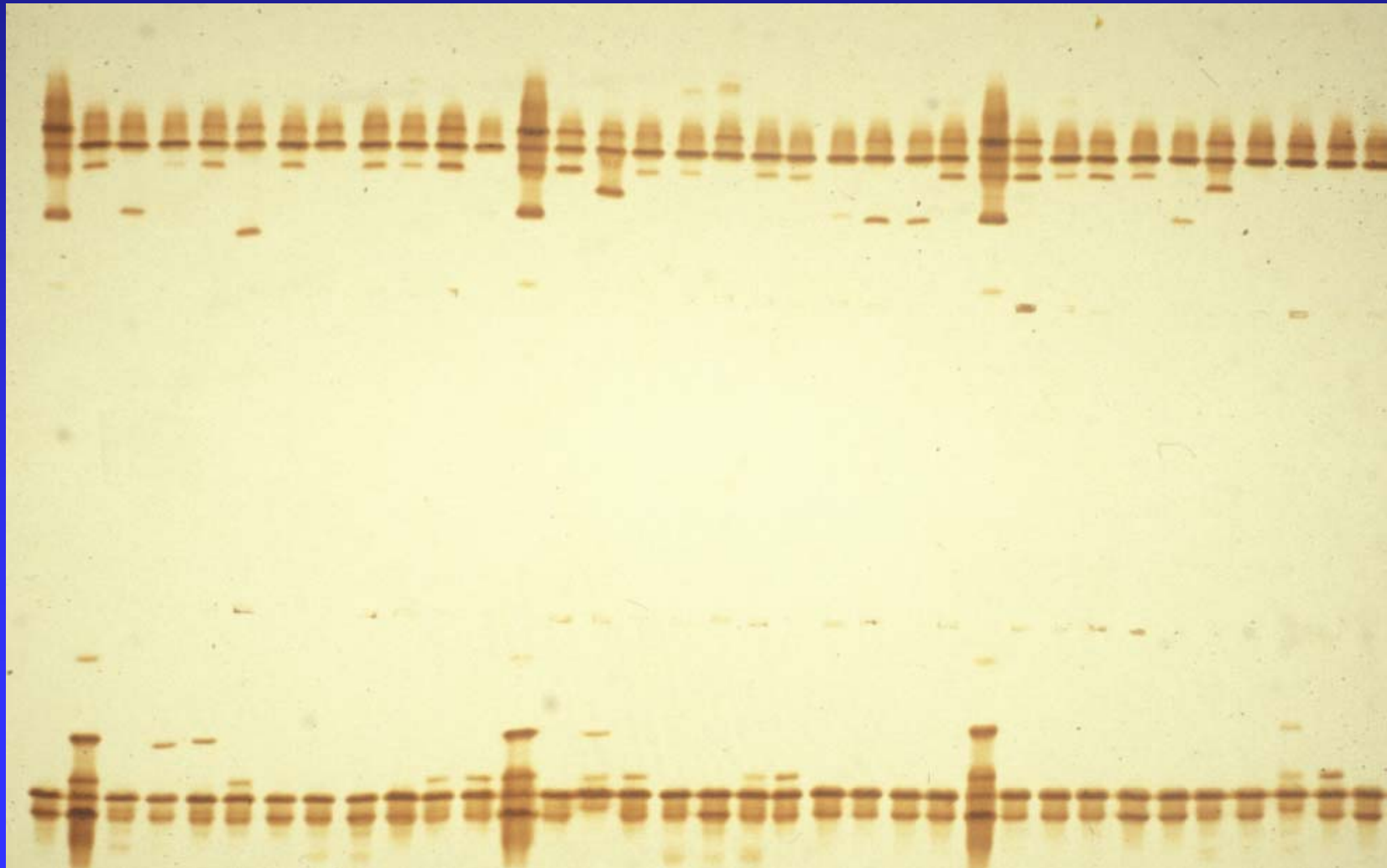
National Institutes of Health  
Consensus Development  
Conference Statement

Volume 6 Number 9  
April 6-8, 1987





# DIAGNOSIS AT BIRTH



# NEWBORN SCREENING 2005

*NNSIS at NNSGRC [www.genes-r-us.uthscsa.edu](http://www.genes-r-us.uthscsa.edu)*

<b>Total Screened</b>	<b>4,190,705</b>
<b>Disease</b>	<b>3,419</b>
Hb FS	1,680
Hb FSC	1,099
Hb FSA	640
<b>Carriers</b>	<b>72,111</b>
Hb FAS	53,532
Hb FAC	14,814
Hb FAE	3,695

# ISSUES CREATED

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- **Follow-up of carriers**
    - Notify carriers' parents
    - Providing education and counseling
    - Follow-up when infant reproductive
  - **Testing couples at risk and extended family**
    - Methods: hemoglobin ELP and CBC
    - Resources
      - Laboratory
      - Health educators
      - Genetic counselors
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# ISSUES CREATED

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- **Negative impact of detecting carriers**

*Rowley: NY State J Med 1986;141:593; Am J Dis Child 1979;133:1248; Am j Dis Child 1983;137:341; Science 1984; 225:138.*

- Stigmatization
- Reduced maternal bonding
- Discrimination

- **Misdiagnosis of carrier state**

- Carriers followed as individuals with disease
  - Over treatment for pain
  - Miss serious medical conditions
- Individuals with disease followed as carriers
  - Unnecessary diagnosis and treatment
  - Vocational problems

- **Does sickle cell trait have impact on wellness**

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# HEALTH IMPACT OF SICKLE TRAIT

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- **Decreased urinary concentrating ability**
    - *Sears Am J Med 1978;64:1021-1036.*
  - **Recurrent hematuria**
    - *Heller et al. N Engl J Med 1979;300:1001-1005.*
  - **Increased incidence of sudden death at extremes of human endurance**
    - *Kark et al. N Engl J Med 1987;317:781-787.*
  - **Pain, splenic infarction, sequestration at altitude**
    - *Kark JA, Ward FT Semin Hematol 1994;31:181-225.*
  - **Increased incidence of pulmonary emboli – mild hyper coagulable state**
    - *Auston et al. Blood 2007;110:908-912.*
  - **Rare tumor – Renal medullary carcinoma**
    - *Davis et al. Am J Surg Path 1995;19:1-11.*
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# HEALTH IMPACT OF SICKLE TRAIT

## Sickle trait and its association with birthweight and urinary tract infections in pregnancy

*Division of Maternal-Fetal Medicine, Department of Gynecology and Obstetrics, Johns Hopkins University School of Medicine, 600 North Wolfe Street, Baltimore, MD (USA)*

*Int. J. Gynecol. Obstet.*, 1990, 33: 19—21

International Federation of Gynecology and Obstetrics

## Bacteriuria in pregnant women with sickle cell trait

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*Departments of Obstetrics and Gynecology,<sup>a</sup> Pathology and Laboratory Medicine,<sup>b</sup> and Biometry and Epidemiology,<sup>c</sup> Medical University of South Carolina, Charleston, SC*

*American Journal of Obstetrics and Gynecology* (2006) **194**, 1366–70

# HEALTH IMPACT OF SICKLE TRAIT

## CONTROVERSIES IN NEUROLOGY

SECTION EDITOR: E. S. ROACH, MD

### Sickle Cell Trait Is a Risk Factor for Early Stroke

*Meredith R. Golomb, MD, MSc*

### Sickle Cell Trait Is Not a Risk Factor for Stroke

*Michael M. Dowling, MD, PhD*

### Sickle Cell Trait

*Innocent Until Proven Guilty*

*E. S. Roach, MD*

ARCH NEUROL/VOL 62, NOV 2005

WWW.ARCHNEUROL.COM

**1781**

**Those who cannot remember the  
past are condemned to repeat it.**

**George Santayana *The Life of  
Reason, Vol 1, 1905***