SICKLE CARRIER DETECTION: LESSONS LEARNED OVER 75 YEARS

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SICKLE TRAIT

Diggs Ann Intern Med 1933;7:769-778
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The image includes two bar charts and a line graph. The bar chart on the left shows the number of individuals examined and the number with the trait by age group. The bar chart on the right shows the number of cases by age and hemoglobin level. The line graph compares the number of cases with and without the trait.
National Sickle Cell Anemia Control Act of 1972. Public Law 92-294

- 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

“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”.”

EARLY SICKLE CELL TRAIT SCREENING AND EDUCATION

  – 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.”

  – 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.
NEWBORN SCREENING SPREDS

NEWBORN SCREENING FOR SICKLE CELL DISEASE AND OTHER HEMOGLOBINOPATHIES

National Institutes of Health Consensus Development Conference Statement

Volume 6 Number 9 April 6-8, 1987
DIAGNOSIS AT BIRTH
<table>
<thead>
<tr>
<th>Total Screened</th>
<th>4,190,705</th>
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</thead>
<tbody>
<tr>
<td><strong>Disease</strong></td>
<td>3,419</td>
</tr>
<tr>
<td>Hb FS</td>
<td>1,680</td>
</tr>
<tr>
<td>Hb FSC</td>
<td>1,099</td>
</tr>
<tr>
<td>Hb FSA</td>
<td>640</td>
</tr>
<tr>
<td><strong>Carriers</strong></td>
<td>72,111</td>
</tr>
<tr>
<td>Hb FAS</td>
<td>53,532</td>
</tr>
<tr>
<td>Hb FAC</td>
<td>14,814</td>
</tr>
<tr>
<td>Hb FAE</td>
<td>3,695</td>
</tr>
</tbody>
</table>
ISSUES CREATED

• 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
ISSUES CREATED

• Negative impact of detecting carriers
  – 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
HEALTH IMPACT OF SICKLE TRAIT

- Decreased urinary concentrating ability
- Recurrent hematuria
- Increased incidence of sudden death at extremes of human endurance
- Pain, splenic infarction, sequestration at altitude
- Increased incidence of pulmonary emboli – mild hyper coagulable state
- Rare tumor – Renal medullary carcinoma
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)

International Federation of Gynecology and Obstetrics

Bacteriuria in pregnant women with sickle cell trait

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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

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

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