

**Sickle Cell Disease: Health Objectives Working Group  
Blood Diseases Branch  
National Heart, Lung, and Blood Institute (NHLBI)  
Baltimore, MD**

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## **INTRODUCTION**

In 1979, the U.S. Surgeon General released a report to develop health objectives for the U.S. population. This set the Nation's health agenda for the coming decade and was soon followed by the 1990 Health Objectives for the Nation. At that time, most objectives emphasized primary prevention, although several addressed secondary prevention for people with clinical diagnoses such as hypertension and sexually transmitted diseases. Updated objectives were released in 1990, titled Healthy People 2000. At that time, communities were encouraged to use the Federal objectives as templates and to develop their own objectives. These initial efforts at improving and tracking population health were instrumental in generating new data systems for measuring progress toward the objectives. Healthy People 2000 and Health People 2010 emphasized health improvements and elimination of disparities among population groups. These objectives targeted reducing morbidity and mortality. Over time, these and other efforts have led to improved data systems that have yielded data for health and quality care indicators.

Currently (2006), there is one sickle cell disease (SCD) health objective in Healthy People 2010. The section on Maternal, Infant, and Child Health includes the objective to "reduce hospitalization for life-threatening sepsis among children aged 4 years and under with sickling hemoglobinopathies" (Objective 16-21). Although the National Hospital Discharge Survey is listed as a potential data source, it is unclear whether it can be used to track progress for this one objective.

On September 14-15 2006, the NHLBI convened a Working Group to develop health objectives for people with SCD. The working Group was charged with identifying health priorities for SCD that should be used to drive and prioritize subsequent efforts, and that should include appropriate measures by which to evaluate efforts.

## **OPENING PRESENTATIONS**

On Thursday, September 14, Dr. Ellen Werner welcomed the participants and introduced two speakers: Dr. Charles Peterson, Director of the Division of Blood Diseases and Resources at the NHLBI; and Dr. Marie Mann, of the Health resources and Services Administration (HRSA). They discussed their previous work with SCD and the importance of developing health objectives for patients, families, and health care providers. Dr. Kathryn Hassell distributed examples from the current Healthy People 2010 and discussed their application to SCD. Dr. Werner then reviewed the history of health objectives and health promotion and disease prevention activities in the Federal Government.

## **METHODS**

Participants were divided into five breakout groups and were charged with developing recommendations for the following topics:

1. **Health Status:** objectives related to physical and medical status, life expectancy, quality of life, and functioning.
2. **Health Promotion:** objectives related to activities to prevent complications, self-management, health behaviors, health education, and reduction of risk behaviors. [It also includes social support systems, patient knowledge, and coping.]
3. **Health Services:** objectives related primary and specialized health services, medical tests, access to health care, and emergency services.
4. **Health Workforce:** objectives related to the availability of a wide range of health care providers and their skills and knowledge of SCD.
5. **Talking Points:** important questions to answer and compelling data-supported facts about SCD that could be used to raise awareness about SCD.

The Working Groups were instructed that the objectives were to be **Specific, Measurable, Ambitious, Reachable, Time-bound, and Trackable** (i.e., SMARTT). In addition, participants were told to develop objectives that are:

- Important and understandable to the community and relate clearly to the vision
- Prevention-oriented, with emphasis on health improvements that can be achieved through population-based and health service-based interventions
- Action-oriented, including suggestions for a set of interim steps needed to achieve the target objective
- Useful and relevant to states, localities, and the private sector; include activities targeted to community organizations, such as schools, worksites, health settings, and churches.
- Measurable including health outcomes, behavioral and health service change, and community capacity
- Stated in an affirmative tone
- Designed to permit continuity and comparability of measurement with Healthy People 2000 objectives
- Supported by, or have the potential to be supported by, sound scientific evidence.

After each breakout developed a list of objectives, the groups moved to the next topic to allow each group to review the other groups' objectives, which were listed on flip charts. Each group added its input to the previous group's objectives. Finally, the entire Working Group convened to select which objectives they all agreed on, those to be deleted, and those that needed more work. They also reviewed and discussed the talking points.

On Friday, September 15, the participants reviewed and finalized the objectives identified on the previous day. The group voted unanimously to include the June 2003 Consumer Working Group objectives as generic models for stating the SCD objectives.

## HEALTH STATUS

### Background Information

**Children.** Children with SCD have significant physical and mental health morbidity. The onset of cerebral vasculopathy early in childhood leads to variable degrees of cognitive impairment, an increased risk of stroke (~11 percent cumulative incidence), and a substantial cumulative incidence (30 percent of children by age 18 with hemoglobin SS) of microvascular cerebral ischemia and infarct. Acute Chest Syndrome (ACS), and pain are other common complications. Pediatric patients have an average 1 hospitalization every 2 years for pain. However, recent single center longitudinal data show that the vast majority of infants with SCD live to adulthood in contrast to earlier natural history studies

SCD can be diagnosed with newborn screening. The incidence of clinical stroke has declined with the use of Trans-Cranial Doppler (TCD) ultrasound screening, and subsequent treatment with chronic transfusions to reduce the recurrence of stroke and silent stroke [need results of trial]. Other therapies are being developed to reduce the incidence of ACS and ameliorate its complications. Hydroxyurea, which is FDA-approved for symptomatic adults with SCD, is being used more widely in children as a result of previous and on-going NIH-funded studies of its safety and efficacy in children.

A comprehensive system is needed for transitioning children with SCD to successful adult life in a developmentally appropriate context—focusing on families of SCD infants and, subsequently, on the individuals with SCD.

**Adolescents.** Treatment of adolescents is challenging because their normal developmental changes and growing independence are complicated by SCD manifestations and the stress of living with a chronic disease. Adolescents' growth, nutrition, and sexual development are impacted by SCD. Pain and anemia leads to decreased participation in normal adolescent activities and impacts self-image. Depression and anxiety may be present. The status of mental health functioning in SCD is not well understood.

Disease management is more complex as adolescents age and have more complications, such as leg ulcers, increased pain, priapism, and avascular necrosis (AVN). Chronic transfusion can lead to iron overload and the need for specialized services such as chelation.

The transition from adolescent to young adult involves gaining autonomy from parents. This is difficult because children have grown to trust their family members and depended on their care and love. The often long-standing relationship with the pediatric healthcare team must be broken and replaced with a new adult-focused healthcare system, which is unfortunately often underfunded, understaffed and fragmented.

**Adults.** The majority of adults with SCD live long, productive lives even though they face increasing challenges. Chronic complications greatly impact adult health and function in the areas of pulmonary dysfunction, renal disease, chronic skin ulcers, hypertension, and heart disease. Emergency Department physicians see the more impacted patients; for example, 5 percent of SCD patients represent 85 percent of emergency room (ER) admissions [AHCR Database; Is there data from the CSSCD Natural History Study. However, the overall health

status of the approximately 50,000 adult SCD patients has not been well characterized . A goal is to achieve good health maintenance for SCD as well as for any other co-morbid health conditions.

### **Objectives**

- By 2010, determine the current health status of SCD patients, including:
  - Life expectancy
  - Prevalence of disease-related complications
    - Pulmonary hypertension
    - Cerebral vascular disease
    - Renal disease
    - Retinopathy and blindness
    - Avascular necrosis and orthopedic complications
    - Depression and mental health status
    - Common diseases and co-morbidities (e.g., diabetes mellitus, hypertension, coronary artery disease, obesity).
  - Prevalence of medication use, particularly opioid analgesics
  - Prevalence of renal transplants, hematopoietic stem cell transplants, and dialysis
  - Prevalence of disability and functional impairments
  -
- By 2020:
  - Increase life expectancy by 10 percent.
  - Increase transplants (renal, hematopoietic stem cells) by 10 percent
  - Increase utilization of hydroxyurea and chelation to 80 percent of eligible patients.
  - Decrease the percentage of adult patients with serious iron overload to less than 1 percent.
- By 2015, establish the use of Transcranial Doppler (TCD) ultrasound to screen all at-risk children with SCD by age 6.
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- By 2015, increase to 100 percent the proportion of patients who, by age 18 have an eye exam by an optometrist or ophthalmologist, including a retinal exam by dilated fundoscopy.

- By 2010, implement developmentally appropriate transition programs throughout the lifespan at 10 to 20 demonstration sites.
- By 2010, implement a Health Insurance Portability and Accountability Act (HIPAA)-compliant, state-specific SCD registry linking mortality and health status, to be used for service delivery, tracking, and followup. Develop a program to desensitize persons with SCD to voluntary enroll in such a registry.
- By 2010, implement a system for transitioning individuals with SCD to successful adult life, focusing on families of infants detected in newborn screening and subsequently on the individuals with disease in a developmentally appropriate context. [ same as above]

## HEALTH PROMOTION

### Background Information

**Health Maintenance Behaviors.** As with the general population, SCD patients need good health maintenance behaviors. This can be achieved by increasing education about the need to maintain health checkups. Both adult and pediatric patients need disease-specific education, yearly health checkups, specialty care, resource education, and support networks.

**Risk Behaviors.** SCD patients and the general population have similar incidence of risks related to smoking, alcoholism, and substance abuse. However, the effects of these risk behaviors on the health status of the SCD population are likely more severe. SCD patients are more likely to have ER admissions. [Eckman; Bellevue]

Despite the prevalence mis-conception, opioid addiction is not a common problem among SCD patients; it affects 4–5 percent of adults with SCD. Healthcare providers are themselves three times more likely to be addicted. Pseudo-addiction is often due to suboptimal analgesic management and leads to stigmatization.

**Coping.** Data indicate that the vast majority of SCD patients make positive adjustments to their disease and develop many coping strategies. But we can do better. Many patients with decreased social support networks have difficulty coping with the disease, and data indicate this is a predictor of hospitalization. Maternal coping predicts more success than children’s coping for SCD and other diseases .

**Patient Empowerment and Self-Care.** Patients’ health status depends in part on their sense of empowerment and their ability to provide self-care and interact with providers. They need to acquire the confidence to acknowledge their health care needs and be able to access support that may be required. Both patients and their health care providers need to be empowered to successfully manage their health and wellbeing. There is a need to present a more positive image of SCD patients, highlighting those that lead productive and successful lives.

**Patient Knowledge.** Parents of children with SCD need education about general health as well as SCD-specific information. Patients require resources to help them learn about SCD and manage self-care.

## Objectives

- By 2010, increase by 50 percent the SCD patients and their families who feel they are partners with their health care providers.
- By 2015, increase patient self-awareness to recognize the negative impacts of SCD.
- By 2015, increase patient awareness about the steps necessary to prevent complications of SCD.
- By 2015, increase to 80 percent the patients who recognize when they need access to social support or support services.
- By 2010, develop demonstration projects to create programs to educate patients and their families about psychosocial needs and coping strategies.
- By 2015, 50 percent of patients and their families will become active partners with their health care providers. [same as above]
- By 2010, reduce risk behavior, such as smoking, alcoholism, drug use (e.g., cocaine, marijuana), by 20 percent among adults and children with SCD.
- By 2010, increase to 100 percent the SCD patients who use at least one nonpharmacologic approach to pain management.
- By 2010, establish a central resource (e.g., an 800 number, Web site) that will provide patients and providers with educational information and material on SCD. Utilization of this service should be tracked along with user knowledge of SCD.
- By 2010, establish a program to teach and assess cultural competency in patient/provider interaction and communication with a focus on SCD.

## HEALTH SERVICES

Health services include primary and specialized health services, medical tests, access to health care, and ER services.

### Background Information

**Optimal and Suboptimal Treatment.** Evidence-based guidelines for treating SCD complications are needed. Optimal treatment requires utilizing appropriate partnerships between ER physicians, PCPs, neurologists, pulmonologists, and pain management specialists. Models of care do not typically exist for SCD.

Mental health and psychological support services for SCD patients are lacking in many areas of the United States. Qualified practitioners of non-pharmacologic pain management (massage, acupuncture, hypnosis, cognitive behavioral therapy, etc) are either not available or poorly compensated for services to SCD patients. In some geographic areas, the services of physicians knowledgeable about SCD are limited. This is usually not a problem in major cities, but in some

smaller cities or rural areas, patients must see general practitioners. Some patients need to travel hours to see specialists because the availability of services is poor in their area.

**Access to Services.** Even if services are available, reimbursement is a major issue. Health insurance or disability payments often will not pay for care such as neurocognitive assessment, magnetic resonance imaging (MRI), and neurologist visits. Without private health insurance, neurocognitive, dental, orthopedic, and eye care may not be accessible. Inadequate support is due to inadequate funding.

Most children receive recommended health care maintenance for SCD because they have better access to health insurance. While there have been improvements in access to care for children, the system for adults is fragmented and not focused on prevention and health maintenance. Children receive episodic care in the ER; accurate data are not available to characterize this problem.

Other barriers to accessing SCD services include lack of transportation to services, and barriers to trust.

**The “Medical Home.”** The concept of a knowledgeable provider in a “medical home” involves a primary care provider who coordinates a variety of subspecialty care for children with special health care needs. The “medical home” concept can be used for SCD. This concept differs between the American Academy of Pediatrics and the Health Resources and Services Administration (HRSA). While adults have no “medical homes,” the same concept (relying on PCPs and specialists) can apply to them.

**Transition Programs.** A study by Ballas, et al found an increased risk of mortality in patient who do not transition well. Transition from the pediatric to adult care is a major issue.

## Objectives

- By 2010, establish a telephone referral service for referrals to health care providers.
- By 2010, establish nationally accepted and implementable standards for blood transfusion in SCD that address antigen matching, transfusion targets, and indications.
- By 2015, establish evidence-based standards of care for pediatric and adult SCD patients using the National Cancer Institute approach, which is based on evidence, data, or practice, or the U.S. Preventive Task Force approach.
  - Routine health care maintenance and SCD-related health care maintenance (i.e., flu shot, pneumonia vaccine)
  - Mental health services
  - Pain protocols for SCD widely known and used
  - Palliative care
  - Referral to subspecialists
- By 2010, increase by 25 percent the proportion of SCD patients with public or private health insurance coverage. By 2015, increase this coverage to 100 percent of SCD patients.

## HEALTH WORKFORCE

### Background Information

**Hospital Visits.** SCD patients are a significant population in many urban hospitals; SCD is the second most common diagnosis in the inpatient population. It is also common in urban ER visits.

**Workforce Knowledge of SCD.** Although a few Centers of Excellence exist (there are only 10 NIH-funded Comprehensive Sickle Cell Centers), the vast majority of SCD patients do not have access to a workforce knowledgeable about SCD. Only about 1 percent of training and certification/recertification of ER and primary care physicians (PCPs) is related to SCD.

General practitioners need more knowledge of SCD. Other health care providers that need education about SCD include nurses, physical therapists, occupational therapists, neurologists, psychologists, neuropsychologists, dentists, ophthalmologists, optometrists, and obstetrician-gynecologists.

A general problem is that both the pool of PCPs and hematologists is shrinking. The lack of interest in hematology is part and parcel of inequalities in reimbursement.

**Training.** Professional organizations (e.g., for orthopedics and neurology) should be encouraged to provide education about SCD for physicians. A 1-day (4–8 hour) Continuing Medical Education (CME) session on “what you need to know about SCD” is a possible approach that might be tailored for many different audiences. Certification provides an incentive to attend these sessions, and certifying examinations can ensure that all providers are educated about SCD.

There is a perception that SCD is not an area covered in medical training. SCD is a model disease in teaching biochemistry and genetics—but not in other training. SCD is often cited in medical education as an example of translation (from genetics to medical condition).

The entire education system at all levels—public schools, colleges, graduate schools—is not knowledgeable about SCD. For example, school nurses need to know about SCD to help explain absences.

### Objectives

- By 2010, increase opportunities (scholarships, incentives) for SCD patients to join the health care delivery workforce for SCD.
- Beginning in 2010 and completed by 2015, establish that 100 percent of providers will have training and clinical exposure to SCD.
- Beginning in 2010 and completed by 2015, establish that provider training, certifying exams, and recertification exams will have content related to SCD.
- By 2015, increase the number of specialty health care providers who are trained and certified to manage SCD patients by 50 percent. These providers include nurses, PCPs and specialty physicians, psychologists, trained genetic counselors, social workers, and health educators.



- By 2010, increase knowledge of the attitudinal and structural determinants of health care access and utilization in SCD.
- By 2010, identify physicians, nurses, and social workers who provide care for patients with SCD and increase access to care by 35 percent.
- By 2015, establish that 90 percent of identified SCD patients will have a knowledgeable provider located within an hour driving distance from their homes. [An interim goal might target 60 percent by an earlier deadline.]
- By 2010, develop and disseminate curricula for training of specialists and subspecialists to develop knowledge about management of SCD and its complications.

## **TALKING POINTS**

This section identifies the important questions to answer and compelling talking points. Data are needed for many of these talking points. The following questions and talking points were raised during the breakout sessions and discussion and are grouped here by topic.

### ***A Unique Disease***

- Hemoglobin mutations are the most common mutations detected in newborns in the US and SCD is the most common genetic disease detected in newborns in the United States.
- SCD is the only disorder in which pain can start as early as 6 months of age. SCD-related pain can require narcotics/analgesic treatment throughout the lifespan.
- SCD patients use more pain medication in 1 month than the average person will need in a lifetime. .
- The unpredictability of SCD and the lack of knowledge among patients and providers present unique problems.

### ***Genetics***

- What is the percentage of persons with SCD who do not know their genotype? What is the percentage of family members of SCD patients who do not know that genetics is involved in SCD and were not informed they were carriers of the SC gene?
- What percentage of women are tested for the SC trait (and other abnormal hemoglobin traits) before or during pregnancy?

### ***Who Is Affected***

- What is the prevalence of SCD by age and geographic location? How many health care providers are available for these patients? A population-based research question (using a survey) is needed to learn *who* is affected and *where* SCD occurs. This would identify underserved populations and areas where service is needed.

- One in 400 African-American newborns has SCD. There is a mismatch between the ethnicity of providers and patients. Cultural competence should work across cultural differences.

### ***Health Status***

- All people with SCD need health education and health maintenance.
- Are *some* or *most* patients doing well? Those who are doing well can serve as a role model, and peer health educator.
- An unknown number of SCD patients are struggling because of complications and poor health services. How many people with SCD are not receiving care?
- To manage pain, all people with SCD are encouraged to use nonpharmacologic approaches—such as distraction techniques (e.g., talking on the telephone, walking).

### ***Impact on Families***

- Family relationships are impacted by any chronic disease. What percentage of SCD families experience social breakups, financial burdens, and burdens on children and other family members?
- All families with children's diseases are burdened. However, compared with families with childhood asthma, families with SCD take more unpaid days of leave, and they experience more depression among caretakers.

### ***Impact on Patients***

- What is the impact of SCD on the individual's personal identity, unemployment, lack of education, family roles, and interpersonal relationships?
- Socioeconomic factors related to SCD are understudied and poorly understood. What percentage of SCD patients are unemployed? What percentage of patients lack life insurance?
- SCD patients feel stigmatized in the medical system.

### ***Cost***

- Many SCD patients are ER users. Better preventive and support systems would decrease the cost of ER and inpatient services.
- In the U.S., people with SCD need more than 500,000 units of blood per year, at a cost of \$300 per year, per patient

### ***Research***

- Research funding for cystic fibrosis is about 15 times higher than for SCD.
- Data indicate that SCD is one of the most successful diseases in which translational research leads to clinical care, resulting in improvement in patient health [Peterson].
- Abstracts on SCD research are available from the Sickle Cell Information Center at [www.scinfo.org](http://www.scinfo.org). Individuals also can sign up for a monthly newsletter.

### **ADJOURN**

Dr. Werner thanked all the participants for their input. The meeting was then adjourned.

