Newborn Screening for Sickle Cell Disease and Other Hemoglobinopathies
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Hemoglobinopathies represent one of the major health problems in the United States and constitute the most common genetic disorders in some populations. Sickle cell disease (SS, SC, S-β-thalassemia) alone affects about one in 400 American black newborns, as well as persons of African, Mediterranean, Asian, Caribbean, Middle Eastern, and South and Central American origins.

For the past 20 years, the medical profession has known that children with sickle cell anemia have an increased susceptibility to severe bacterial infection, particularly due to *Streptococcus pneumoniae*. The risk of major infection and death posed by this organism is greatest in the first 3 years of life and can occur as early as 3 months of age. In fact, this infection may be the first clinical manifestation of disease. The infection can be fulminant, progressing from the onset of fever to death in a matter of hours, and the case fatality rate is reported as high as 30%.

In addition, acute splenic sequestration, another acute catastrophic event, contributes to early mortality in children with sickle cell anemia and may occur as early as 5 months of age.

It has been proposed that early diagnosis to identify infants with major sickle hemoglobinopathies, who have a high risk of early mortality and morbidity, is essential to institute appropriate ongoing care and effective measures of prophylaxis and intervention. Early diagnosis of hemoglobinopathies should be in the newborn period.

Even though the technology to screen infants in the newborn period has been available for the past 15 to 20 years, screening has not received widespread acceptance. This has been due to a number of factors, including doubt that accurate diagnosis was possible because of the high proportion of fetal hemoglobin, the perception that without an effective treatment early diagnosis would not decrease morbidity and mortality, uncertainty about whom to test, and questions about obligations to those identified as carriers.

Data are now available concerning the positive effects on morbidity and mortality of newborn screening, immediate entry into programs of comprehensive care, and institution of penicillin prophylaxis.

To address the issues of screening and to enhance understanding among scientists, those responsible for the health of children, and the public at large, the National Heart, Lung, and Blood Institute of the National Institute of Health (NIH), the National Institute of Child Health and Human Development, the Genetic Disease Services Branch of the Health Resources and Services Administration, and the NIH Office of Medical Applications of Research sponsored an NIH Consensus Development Conference on Newborn Screening for Sickle Cell Disease and Other Hemoglobinopathies.

The conference brought together approximately 400 biomedical investigators, clinicians, other health professionals, parents, and representatives of the public. Following two days of presentations by medical experts and discussion by the audience, a consensus panel representing the fields of biochemistry, genetics, pediatrics, obstetrics, hematology, public health, nursing, law, ethics, epidemiology, and counseling considered the evidence and developed the Consensus Statement. The conference was designed to answer the following questions: (1) Are programs for screening the newborn for sickle cell disease effective in decreasing morbidity and mortality? (2) What are the techniques of screening, and what is their efficacy? (3) What are the major factors to be considered, including benefits and risks, in conducting newborn screening programs? (4) What are the optimal follow-up and management of infants identified with hemoglobinopathies (disease and carriers)? and (5) What future research directions are indicated?

The answers are contained in the Consensus Development Conference Statement and were published. This supplement is a compilation of the scientific papers, according to their presentation to answer each specific question, and commentaries.
or opinions presented at the conference and represents the data upon which the panel based its conclusions and recommendations.

The sponsors are pleased that the conference provided a national forum for exploration of all issues concerning this important topic, and the conference has proven to be a major impetus for newborn screening throughout the country.

The papers are being published with the hope that the information will be useful as new programs for newborn screening are being initiated across the country and established programs reevaluate their process and initiate change.

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REFERENCE
1. Newborn screening for sickle cell disease and other hemoglobinopathies. *JAMA* 1987;258:1205-1209