



DEPARTMENT OF HEALTH AND HUMAN SERVICES

Secretary's Advisory Committee on Heritable
Disorders in Newborns and Children
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October 15, 2010

The Honorable Kathleen Sebelius
Secretary of Health and Human Services
200 Independence Avenue, S.W.
Washington, DC 20201

Dear Secretary Sebelius:

The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (the Committee) is charged with making systematic evidence-based and peer-reviewed recommendations that include the heritable disorders that have the potential to affect public health significantly, for which all newborns should be screened. Thus far, nine conditions have been sent to the Committee for consideration of an evidence review and for consideration for addition to the Committee's Recommended Uniform Screening Panel. In May 2010, Severe Combined Immunodeficiency (SCID) was added to the panel. During the May 13-14, 2010 Committee meeting, the Committee voted to not recommend the addition of Hemoglobin H to the Panel. At the Committee's most recent meeting on September 17, 2010, the Committee reviewed a final draft report of the evidence review for Critical Congenital Cyanotic Heart Disease and voted to add this disorder to the Panel.

Congenital Heart Disease is an overarching term describing a spectrum of clinical outcomes derived from any number of defects that are present in the structure of the heart at birth. Specific defects may involve the interior walls of the heart, valves inside the heart or the arteries and veins that carry blood to the heart or out to the body. These varied congenital defects change the normal flow of blood through the heart, leading to a range of conditions and symptoms. Congenital Heart Disease affects about 7 to 9 of every 1000 live births in the United States and Europe and is the most common cause of death in the first year of life, with defects accounting for 3% of all infant deaths and more than 40% of all deaths due to congenital malformations. **Critical** Congenital Heart Disease is a group of defects that cause severe and life-threatening symptoms and require intervention within the first days or first year of life.

Current methods for detecting Congenital Heart Disease generally include prenatal ultrasound screening and careful and repeated clinical examinations, both in the nursery and as part of routine well-child care. Critical Congenital Heart Disease is often missed during the routine clinical exam that generally is scheduled prior to a newborn's discharge and many cases of Critical Congenital **Cyanotic** Heart Disease are missed by

discharge and post-discharge clinical exams. A large epidemiological population-based study showed that 78% of cases with hypoplastic left heart syndrome (HLHS) were discharged from hospital before diagnosis. HLHS is universally fatal without surgical intervention, sometimes within the first days of life, and the vast majority of deaths in this patient population occur within the first months of life. Fetal ultrasound screening programs improve detection of major congenital heart defects; however, prenatal diagnosis alone picks up less than half of all cases.

Newborn screening using pulse oximetry for detecting Critical Congenital Cyanotic Heart Disease was examined by the Committee's evidence review workgroup. Pulse oximetry is a method to augment current approaches (clinical exam and prenatal ultrasound) for the detection of Critical Congenital Cyanotic Heart Disease. Newborn screening using pulse oximetry is a test that occurs at the bedside (in the nursery or otherwise) similar to newborn screening for congenital hearing impairment. Pulse oximetry is a non-invasive test that estimates the percentage of hemoglobin in blood that is saturated with oxygen. While some types of Critical Congenital Heart Disease may present with hypoxemia, they do so only some of the time and are therefore less likely to be detected by pulse oximetry screening. Neonates with abnormal pulse oximetry screening results need confirmatory testing for the cause of the cyanosis, and immediate intervention. Virtually every hospital, even small ones, frequently uses pulse oximetry as a standard of care in their newborn nurseries.

When developing its recommendations to the Secretary, the Committee considers the nature of the science itself underlying the potential additions of the technology and the heritable conditions to the Committee's Recommended Uniform Screening Panel as well as the public health implications of implementation. Although there are recognizable evidence gaps (for example, standardization of screening protocol) there are compelling reasons for recommending screening newborns for Critical Congenital Cyanotic Heart Disease.

The Committee therefore recommends the addition of Critical Congenital Cyanotic Heart Disease to the Committee's Recommended Uniform Screening Panel with the understanding that the following activities will also take place in a timely manner:

1. The National Institutes of Health shall fund research activities to determine the relationships among the screening technology, diagnostic processes, care provided, and the health outcomes of affected newborns with Critical Congenital Cyanotic Heart Disease as a result of prospective newborn screening;
2. The Centers for Disease Control and Prevention shall fund surveillance activities to monitor the Critical Congenital Cyanotic Heart Disease link to infant mortality and other health outcomes;
3. The Health Resources and Services Administration shall guide the development of screening standards and infrastructure needed for the implementation of a public health approach to point of service screening for Critical Congenital Cyanotic Heart Disease; and
4. The Health Resources and Services Administration shall fund the development of, in collaboration with public health and health care professional organizations and

families, appropriate education and training materials for families and public health and health care professionals relevant to the screening and treatment of Critical Congenital Cyanotic Heart Disease.

The Committee fully recognizes that the various Agencies within HHS determine and carry out their missions within their goals and the budgets which they have available.

Sincerely yours,

R. Rodney Howell, M.D.
Chairperson