

SEP - 9 2005

Advisory Committee on Heritable
Disorders and Genetic Diseases in
Newborns and Children
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The Honorable Michael O. Leavitt
Secretary of Health and Human Services
200 Independence Avenue, S.W.
Washington, DC 20201

Dear Secretary Leavitt:

In 2001, the Health Resources and Services Administration (HRSA), Bureau of Maternal and Child Health, contracted with the American College of Medical Genetics (ACMG) to convene an expert group to review available information on newborn screening and to make recommendations based on the best scientific evidence and analysis of that information to strengthen State-based newborn screening programs in this country. The report from ACMG contains a summary of the project, a review of the current and future landscape of newborn screening, the process used for the expert group's analysis and the expert group's recommendations. The report also describes the decision making algorithm used by the expert group and outlines recommendations for a minimum uniform panel of conditions for screening in State newborn screening programs.

In January 2005, the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (Committee) reviewed and discussed the final report and recommended the report be released to the public and agreed to review the public comments and provide advice to the Secretary on those comments and the report.

On March 8, 2005, HRSA announced a 60 day public comment period on the report through publication of a Federal Register notice. The 60-day public comment period has ended and the Committee has reviewed the public comments sent to HRSA.

The Committee notes that the majority of comments were favorable to the report and its findings. Almost all of the comments supported the notion of using a uniform panel in state newborn screening programs. Upon review of the final report and the public comments to that report, the Committee is satisfied with the work of the ACMG expert panel, its methodology, its findings and its recommendations and again endorses the report and its recommendations.

However, the Committee notes that there were a few but significant concerns raised by some of the commentators about the methodology used by the expert panel. The Committee has reviewed that methodology with experts and has found the methodology satisfactory and that the concerns about the methodology do not negate the findings of the ACMG expert panel. The Committee concludes that the report and its findings are based on the best methodology available at this time. The Committee recognizes that analysis

and evaluation is a dynamic process, and therefore, the methodology used by this Committee for its own work in recommending conditions, tests and technologies will evolve over time.

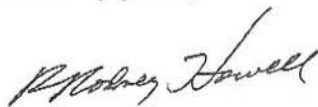
The Committee also realizes that there were some concerns from commentators about issues the ACMG expert panel did not address in detail although the report did note their importance. Many commentators, as did the ACMG expert panel, emphasized the importance of placing newborn screening within the context of our health care system and to view it as a system: that is, newborn screening is more than a test. Newborn screening includes not only the screening test, but importantly follow-up, diagnosis, management and treatment, evaluation, and education. In addition, commentators indicated:

- Access to health care services should be addressed, such as supporting systems to facilitate connection to the medical home for infants identified as screened positive,
- A need for organized information systems to allow for the collection and analysis of relevant health information about screen positive newborns.
- The definition and implementation of a process within the Committee for evaluating and recommending conditions, tests and technologies to be used in newborn screening programs.
- With expansion of newborn screening many financing issues need to be considered, including costs of care and treatments such as medical foods and formulas; cost of newborn screening expansion; and the cost of the newborn screening system.
- Newborn screening processes need to be standardized to bring more uniformity and equity to the newborn screening system.

The Committee has begun to examine and address many of these issues through the formation of subcommittees on laboratory standards and procedures, education and training, and follow-up and treatment.

In conclusion, the Committee strongly and unanimously recommends that the Secretary initiate appropriate action to facilitate adoption of the ACMG recommended screening panel by every State newborn screening program.

Sincerely yours,



R. Rodney Howell, M.D.
Chairperson
ACHDGDNC