

## **Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children**

National Society of Genetic Counselors  
Public Statement: September 23, 2004:

Good afternoon. I am Jennifer Sullivan, representative for the National Society of Genetic Counselors (NSGC). As you are aware, the NSGC represents genetic counselors worldwide and is the leading voice, authority and advocate for the genetic counseling profession. Over the years, the NSGC membership has contributed significant experience and expertise in implementation and coordination of state wide genetic services and clinical follow-up of positive newborn screen results. The NSGC strongly concurs that the status of newborn screening is at a critical juncture for re-evaluation. This reassessment is especially urgent because of the inequalities that can develop between states with new technology. For example the current discrepant implementation of tandem mass spectrometry has led to the inclusion of variable disorders between states. In the near future the availability of new treatments for genetic disease (i.e. enzyme replacement therapy for Pompe disease) will create a new disorders which would then become potential targets of newborn screening.

The NSGC endorses the regular and systematic review of newborn screening through a standardized mechanism. Regular review in such a manner will permit the natural inclusion of diseases as appropriate and effective. The NSGC also enthusiastically supports the rationale for and designation of the 30 “core disorders” for newborn screening recommended by the American College of Medical Genetics (ACMG). Further, we agree that the reporting of all abnormal newborn screening detected during the process of providing these “core disease” results would enhance overall medical knowledge and care. In turn, we support the call for comprehensive and timely reporting of screening statistics, short term follow up of screening results, and long term follow up of affected individuals. The NSGC agrees that such reporting will facilitate the generation of invaluable information to guide present and future newborn screening initiatives.

Our organization represents health care professionals closely affiliated with both the reporting of newborn screening results and the coordination of patient care and follow up through clinical appointments. Thus, the NSGC respectfully requests that in its recommendations this committee also address the need for careful evaluation of each state’s resources to support the ACMG suggestions. Existent state systems which have already incorporated expanded newborn screening have experienced increased demands for clinical follow-up services on already limited resources. We know first-hand the burden that genetic disease places on families, particularly when newly diagnosed, and therefore, we request the evaluation of each state’s clinical genetics resources consider how these resources will need to expand along with the newborn screening program. Further, the NSGC suggests that discussion of funding issues for anticipated services on all levels of the newborn screening process be included in any final recommendations related to expansion of newborn screening services.

NSGC also requests that any Federal policy regarding newborn screening include the stipulation that appropriate newborn screening requires the provision of comprehensive

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genetic services incorporating biochemical geneticists, clinical medical geneticists, genetic counselors, and metabolic dieticians. As the experts in genetic conditions, we want to ensure that high risk infants and their families receive the highest quality of medical care, regardless of geographical location or ability to pay.

In conclusion, the National Society of Genetic Counselors enthusiastically supports the efforts of the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children to address the issue of newborn screening. NSGC respectfully asks that this committee consider the points we have presented within the context of any recommendations the committee makes regarding the possible development of federal policy around newborn screening services. The NSGC continues to be at your disposal and will be pleased to work with you as the committee continues to consider these issues.