

national society
of genetic
counselors, inc.



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Good afternoon. I am Jennifer Sullivan, representing the National Society of Genetic Counselors (NSGC). The NSGC represents approximately 2000 genetic counselors worldwide and is the leading voice, authority and advocate for the genetic counseling profession.

For many years, the NSGC membership has contributed significant experience and expertise in the implementation and coordination of state wide genetic services and clinical follow-up of positive newborn screen results. The NSGC applauds this committee for spearheading the evaluation of the current newborn screening protocols in this country. This evaluation is especially important because of the service inequalities that can develop between states with the expansion of technology and knowledge.

The NSGC endorses the rationale for and designation of the core disorders for newborn screening as recommended by the American College of Medical Genetics (ACMG). Given the lack of long term follow up for many of the conditions endorsed by the screening recommendations, we commend the Committee dialogue yesterday regarding this aspect of newborn screening. We urge the Committee to consider that such evaluation include two important components: 1) a system for regular re-evaluation of the core panel of diseases for the addition or removal of diseases, as the depth and breadth of knowledge in newborn screening, genetics, and medicine in general expands; and 2) a mechanism by which researchers, state programs, and other interested parties can provide new data regarding a disorder or disease for possible inclusion in the revised core panel for newborn screening.

Further, the NSGC highly values the disclosure of all relevant medical information, and we agree that overall medical knowledge and care would be enhanced through the reporting of all abnormal newborn screening results for these core diseases, provided that adequate psychosocial and coping resources are also available. 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 collect critical information to guide present and future newborn screening initiatives.

Since the NSGC represents health care professionals closely affiliated with both the reporting of newborn screening results and the coordination of patient care and clinical follow-up, we respectfully requests that this committee recommend careful evaluation of each state's resources to support the clinical follow-up and necessary long-term monitoring of any national recommendation made in regards to standardization of newborn screening policies. State systems that have already expanded newborn screening have experienced increased demands for clinical follow-up services, stretching already limited resources. We know first-hand the burden that genetic disease places on families, particularly with the initial diagnosis. It is critical that the evaluation of each state's clinical genetics resources include how these resources will need to expand along with the newborn screening program. Further, the NSGC

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recommends 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.

Finally, NSGC also requests that any recommendations regarding a national policy for newborn screening include the stipulation that newborn screening requires the provision of comprehensive medical services incorporating primary care providers, genetic professionals, dietary professionals, and other disease specific medical specialists. It is essential to ensure that the high-risk infants and their families that are identified in newborn screening programs receive high quality and standardized medical care, regardless of geographical location or ability to pay.

In conclusion, the NSGC 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. The NSGC encourages the Committee to recommend periodic evaluation of the national and state directives regarding newborn screening to ensure the availability, accessibility and efficacy of such programs and their adjunct follow up services. Committee guidance regarding such reassessment may help avoid situations such as we have presently, with rapid disparities between state programs. The NSGC continues to be at your disposal and will be pleased to work with you as the Committee continues to consider these issues.