**Public Comments to the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children**

**Statement On Newborn Screening And Treatment Of Individuals With Inborn Errors Of Metabolism Detected By Newborn Screening by the Society for Inherited Metabolic Disorders**

**June 8, 2004**

**Newborn Screening**: Newborn screening, followed by appropriate long-term treatment, is a well-established strategy that effectively reduces disability and death from inborn errors of metabolism. As a result of many forces, including advances in technology and changes in public health systems, newborn screening and the systems of care for children identified by newborn screening are undergoing intense examination and change.

**The SIMD**: The Society for Inherited Metabolic Disorders (SIMD) is dedicated to improving scientific and public understanding about inborn errors of metabolism, and to promoting advances in the identification and care of those affected by inborn errors of metabolism. Members of the SIMD are professionals actively involved in clinical or basic research or patient care directly related to inherited metabolic disorders. SIMD members are scientists, physicians, nutritionists, nurses, genetic counselors, and other health professionals working in patient care and research, in the laboratory and in the clinic, in academia, in public health, in private medical systems and in the biotechnology industry.

**The SIMD and Newborn Screening**: SIMD membership includes world leaders in newborn screening and in the care of patients with inborn errors of metabolism. Members of the SIMD serve on newborn screening advisory committees and task forces at the local, state and national level.

In 1998, the SIMD urged state health departments to examine their newborn screening profiles and consider expanding them by appropriate technology to include disorders of amino acids, organic acids and fatty acid oxidation for which treatment is beneficial. In 2003, the membership of the SIMD was surveyed for opinions on selected key issues in newborn screening and in the long term care of individuals with inborn errors of metabolism detected by newborn screening. The results of that survey were used to develop this statement, which has been approved by the SIMD Board and membership.

**SIMD Survey Results:**

A full report of the survey is available on the SIMD website at www.simd.org. This SIMD statement is based on the following key points: Respondents overwhelmingly agreed that screening of newborn infants for inherited (genetic) metabolic diseases should be expanded in the United States to include medium-chain acyl-CoA dehydrogenase (MCAD) deficiency and other inborn errors of amino acid, fatty acid and organic acid metabolism detectable by analysis of amino acids and acylcarnitines using tandem mass spectrometry (MS/MS). However, SIMD respondents did not reach consensus about which criteria should be used to select conditions for an MS/MS screening panel. Most respondents agreed that each infant in the US should be tested for the same diseases, but a majority of respondents also noted that there are sometimes compelling reasons for variability in testing between populations (for example regional differences in incidence of conditions). Respondents strongly agreed that newborn screening should continue as a mandated state public health process, while envisioning flexibility in implementation including the possibility of regional newborn screening systems and the option to perform newborn screening testing in contracted laboratories that may be public or private. Respondents agreed that the director or medical director of the laboratory that is responsible for biochemical testing to confirm the diagnosis of an inborn error of metabolism should be certified in Biochemical Genetics by the American Board of Medical Genetics or have equivalent qualifications. Finally, while a majority of respondents agreed that newborn screening can be supported by private payers as well as by public funds, they also strongly agreed that there should be public assurance of support for immediate follow-up and long-term treatment to prevent death and disability.

**Recommendations:**

- Screening of newborn infants for inherited (genetic) metabolic diseases should be expanded in the United States to include MCAD deficiency and other inborn errors of amino acid, organic acid and fatty acid metabolism detectable by MS/MS.
- All infants in each state should be tested for the same panel of diseases, but programs should allow for flexibility when there are compelling reasons for variability in newborn screening testing between populations.
- Newborn screening should continue as a mandated state public health process, with ultimate responsibility for a successful program resting with the state public health department. Innovation through regional newborn screening networks and contracted public - private partnerships is likely to improve the quality and scope of newborn screening programs.
- The diagnosis of a biochemical genetic disease in an infant detected through newborn screening should be confirmed in a laboratory where the director or medical director is board certified in Biochemical Genetics or has equivalent qualifications.
- State public health departments should develop mechanisms to adequately fund newborn screening and the treatment of inborn errors of metabolism in those who are identified by newborn screening, including testing, reporting of results, confirmation of abnormal screening results, diagnosis, and comprehensive long- term treatment and evaluation.