Testimony for the Record on behalf of
Pediatrix Medical Group, Inc.
Submitted to the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

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Mr. Chairman and members of the Advisory Committee (on Heritable Disorders and Genetic Diseases in Newborns and Children): My name is Phil Vaughn. I am a neonatologist, and currently serve as Vice President of Pediatrix Screening, a state-of-the art laboratory that provides the most comprehensive program for newborn screening of genetic metabolic disorders in the world.

Since its founding in 1994, Pediatrix Screening has analyzed dried blood spots from over 2 million newborns. Before I discuss the important role that Pediatrix Screening plays in helping State laboratories and health personnel provide the highest-quality newborn screening and clinical testing, I would like to take a moment to briefly describe the services provided by Pediatrix Medical Group (Pediatrix), the parent company of Pediatrix Screening, to aid the Committee's understanding of how metabolic and genetic screening is a natural and complementary adjunct to the spectrum of newborn services we provide.

Pediatrix Medical Group

Pediatrix is the largest neonatal and perinatal physician group in the United States, with 700 physicians and 325 nurse specialists providing direct medical care in more than 200 hospitals across the country. We care for both premature and critically sick newborns, as well as women with high risk pregnancies. Last year, our physicians cared for around 3,000 newborns each day in hospitals in 30 states and Puerto Rico. In addition to neonatologists and maternal fetal-medicine specialists, other members of the Pediatrix team include pediatric intensivists, pediatric cardiologists, pediatric hospitalists, and neonatal nurse practitioners. Pediatrix screens over 225,000 newborns annually for hearing loss, and has been a national leader in developing newborn hearing screening programs across the country over the last decade.

Because Pediatrix is in the unique position of treating so many pregnant women and babies, we are a leader among private-sector companies in developing Best Practice standards, conducting clinical trials, engaging in collaborative research efforts, developing an interactive educational Web site ("Pediatrix University"), offering continuing medical education for physicians and nurses, and tracking the outcomes of more than 180,000 neonatal cases in a centralized database.

Understanding how traumatic a Neonatal Intensive Care Unit (NICU) experience can be for the families involved, we also work directly with parents and family members of critically sick newborns, providing brochures and other materials in easy to understand terminology in order to help prepare them to understand the workings of a NICU. We are a partner in the March of Dimes NICU Family Support program, actively working to ensure that parents are better equipped to handle the additional emotional stress of having their newborn cared for in a NICU and at home.

I mention the scope of Pediatrix' overall efforts in direct clinical patient care, as well as its background in research, expertise in physician education, and leadership in hearing screening to show the level of commitment we bring to health care quality, and the professionalism and responsiveness to our patients and the community at large.

Our goal is to ensure the best start on a healthy life for our patients. We know that our efforts are directly contributing to more advanced treatments and better outcomes, which help all women and babies.

Pediatrix Screening Services

Pediatrix Screening embodies the same philosophies inherent in Pediatrix Medical Group. Our laboratory delivers the highest quality metabolic and genetic screening services in the most efficient and cost effective manner.

Pediatrix Screening is able to detect more than 50 disorders in newborns. The program uses a unique combination of biochemical, tandem mass spectrometry, and DNA based analysis. This spectrum of technologies delivers results with a high positive predictive value. Just as important as the number of disorders screened is the fact that definitive test results are made available to the physician or state health department laboratory typically in less than 48 hours. Health care providers involved in the care of newborns understand that accurate identification and early detection of many disorders can dramatically improve the long-term health of affected newborns. For many of these babies, the timeliness and accuracy of test results is truly a matter of life or death.

In the tragic event that treatment is not available for a particular disorder, we still believe that comprehensive screening is extremely important. Not only can the data gained from screening for all known disorders help researchers study these rare occurrences with the goal of developing future treatments, but it can also provide answers for the parents and family members of a sick or dying newborn.

From 1996-2003, Pediatrix Screening conducted a metabolic autopsy survey of over 17,500 postmortem specimens. A metabolic autopsy looks for compounds that are markers of inherited disorders of metabolism associated with sudden and unexpected death. Pediatrix' Metabolic Autopsy Survey identified evidence of a genetic disorder in ~1 percent of all infant deaths, including some previously misclassified as infection, vaccine related deaths, SIDS, SUDS or Reye-like syndrome. Had comprehensive newborn screening been available in these cases, countless families may have been spared a measure of terror and guilt over these ostensibly "unexplained" deaths.

Supporting state public health efforts

Some of the Advisory Committee's key goals are to provide recommendations that will enhance the ability of state and local health agencies to provide newborn screening, as well as guide the Secretary of Health and Human Services in developing newborn screening tests, policies and technologies to reduce newborn deaths from metabolic or genetic disorders.

We would like to make the Committee aware that resources are currently available – and are being used effectively and efficiently – in the private sector that can help address the serious newborn screening problem. Pediatrix Screening currently partners with health care providers in 48 states and internationally, at hospitals in Latin America, South America, the Middle East and Asia, to provide a variety of newborn screening services. We also perform the state mandated newborn screening in Nebraska, Mississippi, Maryland, and Washington, D.C., and will soon resume mandated screening in Pennsylvania.

We believe that all newborns should have access to a metabolic and genetic screening program that provides for quality, comprehensive testing. Pediatrix Screening will continue to work in partnership with state and federal officials, healthcare professionals, and parents to promote this goal. We offer laboratory testing, educational support, and ongoing research in collaboration with existing state newborn screening programs.

We are aware that some believe newborn screening should only be conducted through a state-operated laboratory under the auspices of the state public health department. Concern has been expressed that overall quality of care, as well as needed documentation, follow up, and treatment for newborns found to have a genetic or metabolic disorder may suffer if states and hospitals are allowed to partner with a private laboratory to provide screening services. Some are also concerned that a state's public health role may be supplanted.

We respectfully, but strongly, disagree with these assertions. First, states where Pediatrix Screening performs state mandated testing, report exceptional quality indicators in their newborn screening programs. A rapid turn around of results allows for prompt clinical attention to infants at risk, and a low

"false-positive" rate avoids both emotional anguish for families and additional health care costs to unnecessarily evaluate healthy infants. We comply fully with all state reporting requirements, and tailor our services to the individual needs of our state clients.

Pediatrix Screening offers states, hospitals, and health providers the most advanced resources available for genetic metabolic screening, in a manner that works within the constructs of each state and local government administrative requirements. We do not supplant state public health efforts, but complement and serve as a partner for such efforts. As an organization, we bring to the table an exemplary track record grounded in the highest quality clinical care, coupled with the expertise needed to ensure that all babies are tested, diagnosed, and treated. Our goal is to work in partnership with all stakeholders to ensure a healthy start for our newest generation.

As well, it should be noted that a tandem mass spectrometer does not, in and of itself, guarantee a high quality comprehensive newborn screening program. The combination of experience and the use of a spectrum of technologies allow our laboratory to provide timely and accurate analysis for our clients and patients.

Parental notification and education: the cornerstone of any screening program

Today there is a disparity across state newborn screening programs in the number of disorders screened for. The number varies from 4 disorders to the state of the art 54 offered by Pediatrix Screening. One result of the current extreme disparity in the level of various state screening programs is that parents of newborns may not be informed that their baby can be tested for a comprehensive set of disorders that far exceeds what is provided by their own state health department. This is particularly disturbing, since most babies born with serious metabolic or genetic disorders rarely show any visible sign of disease immediately after birth. And, most parents do not even realize that screening occurs, or that options for more comprehensive screening are currently available through private laboratories. Sadly, it is only after the baby suffers an adverse consequence that the parent learns that the child could have been screened – and possibly treated – for the disease or defect.

For these reasons, we encourage the Committee to recommend that states be required to inform parents in writing of the option to test their newborn for a comprehensive set of disorders that exceeds the state mandated list. Continuing advances in medical technology will routinely identify new disorders, as well as new methods of treatment. Regardless of which specific disorders this Committee may recommend as part of a universal screening package, parents should be informed that they have the option to have their child tested for other metabolic genetic disorders that may be detected through an existing supplemental newborn screening program.

In closing, on behalf of Pediatrix Screening and Pediatrix Medical Group, we very much appreciate the opportunity to address the Committee today. We look forward to having the chance to discuss these issues in the future with you, and are available to serve as a resource on metabolic screening disorders or to provide any other information that may assist you in your work.