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The Role of Technology in the Neonatal Screening Laboratory

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In 1961, Dr. Robert Guthrie initiated the collection of blood as dried spots on filter paper for testing newborns for the detection of phenylketonuria (PKU) (1, 2). He analyzed these dried blood spot (DBS) specimens with a bacterial inhibition test that he developed to measure phenylalanine (2). This combination of easily transportable specimens and an inexpensive, simple test made large-scale testing for PKU possible. As a result of Guthrie's efforts, the successful introduction of DBS as a source for PKU screening eventually led to population-based screening for over 32 disorders for all newborns in the United States from only a few blood drops collected from a heel stick and absorbed into special filter paper.

Today, newborn screening (NBS) is the largest population-based genetic screening effort in the U.S. and is performed worldwide (3). The detection of treatable, inherited congenital disorders is a major public health responsibility. The U.S. Centers for Disease Control and Prevention has recognized newborn screening as one of the ten great public health achievements in the United States in the first decade on the 21st century (4). Newborn screening programs are designed to detect asymptomatic newborns that are at higher risk for certain diseases from those who may not, using DBS specimens collected 24–72 hours after birth. Babies that are screen-positive are rapidly followed-up with a diagnostic confirmation and appropriate treatment that helps to prevent mental retardation, premature death and other deleterious clinical outcomes. Improvements in technology and the expansion of the recommended uniform newborn screening panel of diseases have led to earlier life-saving treatment and intervention for at least 12,000 additional newborns each year in the United States with selected genetic, hearing and endocrine disorders (5).

NBS laboratories have traditionally been influenced by emerging technologies, and have adapted many clinical testing platforms for use. Examples include the use of tandem mass spectrometry (MS/MS) for amino acidopathies and organic acidurias, multi-marker HPLC testing for hemoglobinopathies, multi-analyte immunoassays for HIV, Hepatitis B and C antibodies, and also second-tier DNA-based assays that detect mutation panels and even next-generation sequencing technologies for cystic fibrosis and other disorders.

The aim of this Special Issue is to present newborn screening laboratory experiences where advances in technology and automation have enabled more efficient high-throughput laboratory screening, the detection of more congenital disorders, as well as the incorporation of molecular methods to the newborn screening laboratory workflow. The six articles

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presented in this Supplement provide a comprehensive look at how technological advances have facilitated increased population-based screening activities in the U.S. and worldwide. The authors provide examples of how different technologies are used in the modern newborn screening laboratory, with particular emphasis on the application of these technologies to improve sensitivity and specificity. Newborn screening laboratories must constantly work to reduce the number of false-positive results – and eliminate false-negative results – in a way that ensures no harm is done during the analytical process. The six articles provide unique approaches to accomplish this goal. Journal readers will gain an appreciation of the wide variety of technologies used by newborn screening laboratorians – a unique high throughput situation that merits close examination as a potential model for disease detection in other fields.

The role that technology plays in the newborn screening laboratory is unequivocally intertwined with the expansion of newborn screening programs worldwide. As such, we must consider how new technologies and their innovative applications can be utilized to further the goals of every newborn screening program in the world. I express my gratitude and appreciation for the work of all of the authors in this Supplement.

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