ISSUE BRIEF

NEWBORN METABOLIC SCREENING

History:

In 1961, Dr. Robert Guthrie of the University of Buffalo developed a laboratory test to detect Phenylketonuria (PKU) in newborns. PKU is an inherited metabolic disorder resulting from an enzyme deficiency; if left untreated, it results in profound mental retardation.

In 1963, Massachusetts became the first state to mandate screening for PKU, which can be treated by lifelong dietary management and counseling.

By 1966, testing for PKU was required by all 50 states and the District of Columbia.

Researchers continued to develop newborn metabolic screening tests to detect many more disorders, and each state decided what screens would be available for its newborns. Significant disparities resulted; in 1997, in Massachusetts, infants were tested for 10 or more disorders, while Utah's babies were screened for three. Some states, like West Virginia, had mandated tests and offered one other such as hemoglobinopathies/sickle cell, by physician request.

Each year four million infants born in the U.S. are screened shortly after birth to detect a variety of congenital conditions. These public health screening programs have become models for population-based screening. This public health activity is aimed at the early identification of infants who are affected by genetic, metabolic disorders. Early identification is critical, as timely identification can lead to a significant reduction in morbidity, mortality and disability in affected infants.

Present:

Over time ever smaller and more powerful microprocessors, technology and equipment have become significantly more sophisticated. This permitted testing that had not been feasible before. A process called tandem mass spectrometry (tandem MS, MS/MS) can now run tests for nearly 30 disorders at one time. None are curable, but all are treatable.

Newborn metabolic screening is a population-based public health activity housed in state public health agencies across the country. All states are now faced with the challenge of expanded newborn screening and are at different stages in approaching the issue.

How's West Virginia Doing?

- All screening specimens are sent to the Office of Laboratory Services (OLS).
- All abnormal tests are followed by the Office of Maternal, Child and Family Health (OMCFH) to assure treatment.
- The Pediatric Genetics Program at WVU offers five sub-specialty clinics across the State in addition to providing technical support for physicians statewide on the treatment/care of infants with metabolic disorder.
- The OMCFH has identified physicians who are serving as expert resources for their colleagues who have children in their practices with rare metabolic disorders.
- All infants, including those with special needs, require a "medical home."
- Treatment and other supports are provided every child who tests positive. This includes the distribution of special formulas, linkage to specialized medical care and community supports, etc.
- There is a data linkage between the Office of Laboratory Services and OMCFH to assure no child is lost to care.

Year	Number of Births	% Screened and Followed
2000	21,616	98.6
2001	21,001	99.8
2002	21,132	99.8
2003	21,480	99.8
2004	21,306	99.8
2005	21,150	99.5

System Expansion

- The U.S. Department of Health and Human Services convened an expert panel who subsequently issued recommendations about newborn metabolic screening.
- The recommendations specifically identified a core panel of tests that states across the country should perform.

- West Virginia's practice has been routinely and universally to screen for PKU, Hypothyroidism, Galactosemia, and hemoglobinopathies which include Sickle Cell disease. The State has also been screening for hearing loss since 2000.

- All cost associated with distribution of the test specimen kits, laboratory processing, repeat testing, short-term follow-up, diagnosis and treatment has been paid for by MCFH since the inception of the program.

- The OLS will be gradually expanding the number of tests performed on West Virginia newborns.

- Effective July 1, 2007, West Virginia expanded the screening panel to include Biotinidase Deficiency (BIOT), and in approximately 60 to 90 days, screening tests for Congenital Adrenal Hyperplasia (CAH) and Cystic Fibrosis (CF) will be added.

- The Bureau for Public Health will be charging birthing facilities for the cost of the newborn screening system. Birthing facilities will recover the cost incurred through appropriate third party payment process.

- West Virginia is participating on a Regional Resource Development Group to address medical capacity across multiple states.

- On or before July 2008, West Virginia will offer screening for all twenty-eight (28) metabolic disorders recommended by the Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, U.S. Department of Health and Human Services.