

FOR IMMEDIATE RELEASE

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WISCONSIN NEWBORNS SCREENED FOR 48 GENETIC DISORDERS

*Screening Panel Includes 29 Disorders Recommended by U.S. Department of Health and Human Services
and Endorsed by the March of Dimes*

MADISON—Families and healthcare providers of babies born in Wisconsin will now receive enhanced newborn screening information for 48 genetic disorders. This includes 29 disorders included on a uniform newborn screening panel being developed by the U.S. Department of Health and Human Services and endorsed by the March of Dimes.

“The change is being made to provide physicians and parents with more complete information regarding newborn screening in Wisconsin,” explains Gary Hoffman, Wisconsin’s Newborn Screening Laboratory Manager. “Our focus is always on doing what’s best for the health of babies born in our state.”

According to state law (WS 253.13), the newborn screening program tests every baby born in Wisconsin for a variety of inherited disorders, such as phenylketonuria (PKU), cystic fibrosis and sickle cell disease. If left untreated, these disorders can lead to poor growth, neurological impairment, brain damage, organ failure, and even death. However, if these conditions are detected early through a simple blood test and treated properly, most often with special diets and vitamin supplements, the affected individuals have a greater chance of survival and a significantly improved quality of life. Although not required by state law, 103 of 105 birthing hospitals in the state also provide voluntary newborn hearing screening. In 2004, 96.3% of newborns received hearing screening.

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The Newborn Screening Program is administered by the Wisconsin Department of Health and Family Services (DHFS), with all of the testing occurring at the Wisconsin State Laboratory of Hygiene (WSLH) located on the University of Wisconsin-Madison campus. About 70,000 newborns are tested each year, of which about 100 babies are found to have a serious inherited disorder. Discovering these disorders early often allows doctors to begin treatment before the disorder becomes life-threatening to the baby.

Although the newborn screening panel is now increasing the reportable disorders from 26 to 48, no additional testing is being done, and no additional blood or resources are required. The expansion represents a better definition of the types of disorders that can be identified using state-of-the-art technology called tandem mass spectrometry (MS/MS). Wisconsin was one of the first states to use MS/MS for newborn screening.

“In 2000 when we started using tandem mass spec, we defined many of the disorders as representing broad categories,” says Hoffman. “Now after five years of our own experience and the experience in other state screening programs that eventually implemented MS/MS technology, we understand that the original categories should be re-defined so that the specific disorders identified by the screening process are listed. In order to give physicians and parents more complete information, we are now changing the panel disorders to reflect this new reality. As technology evolves, we remain committed to helping protect the health of babies born in Wisconsin.”

Currently each state decides what disorders to include on their newborn screening panel. The upcoming national guidelines, with March of Dimes endorsement, should help reduce the disparity in testing panels that currently exists among the state screening programs. According to Murray L. Katcher, MD, PhD, Chief Medical Officer for Community Health Promotion at the Wisconsin Department of Health and Family Services, Wisconsin’s newborn screening program is one of the most complete and comprehensive in the country.

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“We rely on our Newborn Screening Advisory Committee, which includes laboratory experts, parents of affected children, primary and specialty medical care providers, nutritionists, genetic counselors, ethicists, and public health professionals, to review the science and make the best decisions for the babies,” Katcher says. “The advisory group uses established criteria for these decisions, including how often the disorder appears in the state’s population, the implications for the baby if the disorder goes undetected, whether treatments for the disorder are available, whether reliable technology exists to screen for the disorder, and whether screening is cost effective.”

For more information on newborn screening in Wisconsin, visit the WSLH’s Newborn Screening Laboratory website at <http://www.slh.wisc.edu/newborn> and the DHFS Newborn Screening Website at http://dhfs.wisconsin.gov/DPH_BFCH/Newborn_Screen/

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ATTACHMENT 1 – List of 48 disorders on Wisconsin’s Newborn Screening Panel

**WISCONSIN NEWBORNS SCREENED FOR 48 GENETIC DISORDERS
ATTACHMENT 1 (Page 4 of 4)**

**Wisconsin Newborn Screening Panel Disorders
August 1, 2005**

* U.S. Department of Health and Human Services proposed uniform panel that has been endorsed by March of Dimes

Argininosuccinic Acidemia*
Biotinidase Deficiency*
Carnitine/Acylcarnitine Translocase Deficiency
Carnitine Palmitoyltransferase Deficiency Type II
Carnitine Uptake Defect*
Citrullinemia*
Citrullinemia (type II)
Congenital Adrenal Hyperplasia*
Cystic Fibrosis*
Congenital Hypothyroidism*
2,4-Dienoyl-CoA Reductase Deficiency
Galactosemia*
Glutaryl CoA Dehydrogenase Deficiency Type I* (aka, Glutaric Acidemia Type I)
Glutaryl CoA Dehydrogenase Deficiency Type II (aka, Glutaric Acidemia Type II)
Hearing Screening*
Hemoglobin S-Beta Thalassemia*
Hemoglobin S/C Disease*
Hemoglobin Variants (HbE, HbD, HbC)
Homocystinuria*
Hypermethioninemia
Hyperphenylalaninemia
3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency* (aka, 3-Hydroxy-3-Methylglutaric Aciduria)
Isobutyryl-CoA Dehydrogenase Deficiency
Isovaleryl-CoA Dehydrogenase Deficiency* (aka, Isovaleric Acidemia)
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency*
Malonyl-CoA Decarboxylase Deficiency
Maple Syrup Urine Disease*
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)*
Medium Chain 3-Ketoacyl-CoA Thiolase Deficiency
Medium/Short Chain Hydroxyacyl-CoA Dehydrogenase Deficiency
2-Methylbutyryl-CoA Dehydrogenase Deficiency
3-Methylcrotonyl-CoA Carboxylase Deficiency*
3-Methylglutaconyl-CoA Hydratase Deficiency
2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency
Methylmalonic Acidemia, Cbl A and Cbl B forms*
Methylmalonic Acidemia, Cbl C and Cbl D forms
Methylmalonic Acidemia, Mutase Deficiency*
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency* (aka, Beta-Ketothiolase Deficiency)
Multiple CoA Carboxylase Deficiency*
Propionyl-CoA Carboxylase Deficiency* (aka, Propionic Acidemia)
Phenylketonuria*
Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)
Sickle Cell Disease*
Trifunctional Protein Deficiency*
Tyrosinemia Type I*
Tyrosinemia Type II
Tyrosinemia Type III
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)*