

ORDER OF THE  
DEPARTMENT OF HEALTH AND FAMILY SERVICES  
AMENDING RULES

To create HFS 115.04 (9) to (13), relating to screening of newborns' blood for congenital and metabolic disorders.

Analysis Prepared by the Department of Health and Family Services

The early identification of particular congenital and metabolic disorders that are harmful or fatal to persons with the disorders is critical to mitigating the negative effects of such disorders. Therefore, Wisconsin Statute 253.13 requires that every infant born be subjected to blood tests for congenital and metabolic disorders, as specified in administrative rules promulgated by the Department. Parents, however, may refuse to have their infants screened for religious reasons. The Department has issued ch. HFS 115, Screening of Newborns for Congenital and Metabolic Disorders, to administer this statutory requirement. Currently, s. HFS 115.04 lists eight congenital and metabolic disorders for which the state hygiene laboratory must test newborn blood samples.

In determining whether to add or delete disorders from the list under s. HFS 115.04, s. HFS 115.06 directs the Department to seek the advice of persons who have expertise and experience with congenital and metabolic disorders. For this purpose, the Department established the Wisconsin Newborn Screening Umbrella Advisory Group. Section HFS 115.06 also lists six criteria on which the Department must base its decision to add to or delete disorders from s. HFS 115.04. These criteria are:

1. Characteristics of the specific disorder, including disease incidence, morbidity and mortality.
2. The availability of effective therapy and potential for successful treatment.
3. Characteristics of the test, including sensitivity, specificity, feasibility for mass screening and cost.
4. The availability of mechanisms for determining the effectiveness of test procedures.
5. Characteristics of the screening program, including the ability to collect and analyze specimens reliably and promptly, the ability to report test results quickly and accurately and the existence of adequate follow-up and management programs.
6. The expected benefits to children and society in relation to the risks and costs associated with testing for the specific condition.

In consideration of these criteria, the Wisconsin Newborn Screening Umbrella Advisory Group recently recommended that the Department add five aminoacidopathies, i.e., amino acid-related disorders, to the eight disorders currently screened for and listed in s. HFS 115.04. These disorders are:

- Maple Syrup Urine Disease;
- Homocystinuria;
- Tyrosinemia;
- Citrullinemia; and
- Argininosuccinic Acidemia.

Persons with these disorders can experience serious medical consequences such as failure-to-thrive, developmental delays, seizures, mental retardation and death.

The additional costs associated with these five additional screening tests is less than a dollar per baby screened because the amino acids in the blood sample are measured

simultaneously with the acylcarnitines for Fatty Acid Oxidation and Organic Acidemias. In the absence of this screening, the Department estimates the annual Wisconsin costs for these disorders to be \$144,909. The Department also estimates the annual Wisconsin costs of this screening to be \$29,134. Therefore, the cost benefit from these five screening tests is \$115,775.

The Advisory Group also recommended that the Department immediately begin screening newborns for these additional disorders. Before this testing can begin, the Department must change its rules to add the five new disorders to the existing list under s. HFS 115.04. Therefore, the Department issued identical emergency rules that became effective on October 12, 2002. These proposed permanent rules are intended to replace the emergency rules currently in effect.

The Department's authority to create these rules is found in ss. 253.13 (1) and 227.11 (2) (a), Stats. The rules interpret s. 253.13 (1), Stats.

SECTION 1. HFS 115.04 (9) to (13) are created to read:

HFS 115.04 (9) Maple Syrup Urine Disease, ICD-9-CM 270.3.

(10) Homocystinuria, ICD-9-CM 270.4.

(11) Tyrosinemia, ICD-9-CM 270.2.

(12) Citrullinemia, ICD-9-CM 270.6.

(13) Argininosuccinic Acidemia, ICD-9-CM 270.6.

This rule shall take effect on the first day of the month following publication in the Wisconsin administrative register, as provided in s. 227.22 (2) (intro.), Stats.

Wisconsin Department of Health  
and Family Services

Dated: February 14, 2003

By: \_\_\_\_\_  
Helene Nelson  
Secretary

SEAL: