# STATEMENT OF SCOPE
# WISCONSIN DEPARTMENT OF HEALTH SERVICES

**CHAPTER:** DHS 115
**RELATING TO:** SCREENING OF NEWBORNS FOR CONGENITAL DISORDERS
**RULE TYPE:** PERMANENT
**SCOPE TYPE:** ORIGINAL

## SUMMARY

### 1. Description of rule objective/s

The objectives of the proposed rule are as follows:

- To increase the fee, which is provided in s. DHS 115.055 and authorized under s. 253.13 (2),Stats., for the cost of testing newborns for congenital and metabolic disorders and funding follow-up services and other activities under s. 253.13, Stats.
- To add X-Linked Adrenoleukodystrophy (X-ALD) and Mucopolysaccharidosis type 1 (MPS-1) to the panel of congenital and metabolic disorders for which newborns shall be tested in accordance with s. 253.13 (1). The addition of X-ALD and MPS-1 to the newborn screening panel were based on the consideration and recommendations of the Wisconsin Newborn Screening Program ("NBS") and the Secretary's Advisory Committee on Newborn Screening ("SACNBS"). Based upon those recommendations, former Secretary-designee Timberlake approved adding X-ALD and Secretary-designee Johnson approved adding MPS 1 to the newborn screening panel.
- To revise the list of conditions screened to include hypermethioninemia (MET) and trifunctional protein deficiency (TFP).
- To revise outdated or erroneous provisions in the current version of the rule.
- To correct spelling and grammatical errors in the current version of the rule.

### 2. Existing policies relevant to the rule

The Newborn Screening Program performs blood screening as well as hearing and critical congenital heart disease screening for newborns in Wisconsin. Section 253.13 (1), Stats., requires that every infant born in a hospital or maternity home, or other place be screened for congenital and metabolic disorders as specified in rules by the department. Section 253.13 (2), Stats., requires that the Wisconsin Department of Health Services ("the department") impose a fee, by rule, for tests performed under this section sufficient to fund all of the following:

- The cost of testing performed under s. 253.13 (1), Stats.
- The provision of diagnostic and counseling services, special dietary treatment, and periodic evaluation of infant screening programs.
- The cost of consulting with experts under s. 253.13 (5), Stats.
- The cost of administering the hearing screening program under s. 253.115, Stats.
- The cost of administering the congenital disorder program under s. 253.13, Stats.

All funds received are credited to appropriation accounts under s. 20.435 (1) (ja) and (jb), Stats.

Section DHS 115.055 currently imposes a fee of $109. This fee was established in 2010 and imposed by the Wisconsin State Laboratory of Hygiene Board, based on language in s. 253.13 (2) which, at the time, authorized the State Laboratory of Hygiene board to establish a fee "on behalf of the department." 2011 Wis. Act 32 amended s. 253.13 (2), Stats., to remove all references to the State Laboratory or Hygiene Board, and required the department to impose a fee by rule.

Section 253.13 (1), Stats., requires every infant born in a hospital or maternity home or other place to be screened for congenital and metabolic disorders as specified in rule by the department. In order to fulfill its

statutory duty to identify those disorders by rule, s, DHS 115.06 provides that the department seek "the advice and guidance of medical consultants, staff of the state laboratory and other persons who have expertise and experience in dealing with congenital and metabolic disorders" to determine whether to add or delete disorders to the newborn screening panel.

The NBS Umbrella Committee and its subcommittees—which are comprised of a variety of medical practitioners, experts on genetics, pediatrics and medical ethics, and various advocacy organizations—meet regularly to review and evaluate program processes and make recommendations to the department's secretary with respect to adding or deleting a condition from the newborn screening panel. The NBS Umbrella Committee recommendations are then forwarded to the SACNBS—comprised of experts on medicine, statistics, epidemiology, medical ethics, and legal, social, and policy—which advises the secretary on policy issues related to newborn screening panel of conditions and makes recommendations on additions to the newborn screening panel.

The SACNBS recommendations are then forwarded to the department's secretary, who makes a final determination based on the NBS Umbrella Committee and SACNBS recommendations.

### 3. Policies proposed to be included in the rule

Since the fee under s. 253.13 (2), Stats., was originally established, several additional conditions, such as Critical Congenital Heart Disease, Spinal Muscular Atrophy, Carnitine Palmitoyltransferase Deficiency Type IA (CPT IA), and Pompe Disease have been added to the newborn screening panel under s. 253.13 (1). The added conditions have increased the cost of administering the program, which thereby requires increasing the fee to account for the added costs for ordering and administering additional tests, and the increased number of patients needing follow-up care following the screen. The department proposes to revise the rule to include an initial adjustment of $223 and include ongoing increases at the time of the biennial budget based on the average three-year Medicare Economic Index. Additionally, the department seeks to revise outdated or erroneous provisions, and correct any spelling or grammatical errors in the current version of the rule.

X-ALD is a rare (approximately 1 in 15,000 births) inherited condition. X-ALD is caused by a change in a gene that makes a protein which helps the body break down certain types of fats. It is an X-linked disorder that affects both males and females, but females tend to develop symptoms in adulthood. Males with X-ALD are often normal in infancy, but they may go on to develop problems with their adrenal glands, brain and spinal cord. Without treatment, these boys may become seriously ill or develop irreversible neurologic injury during childhood which is ultimately fatal. Treatments for X-ALD include cortisol replacement for adrenal dysfunction and hematopoietic stem cell transplantation (HSCT) to arrest progressive brain abnormalities. There is no cure for X-ALD, but early diagnosis means that children with X-ALD can avoid serious adrenal insufficiency, degenerative brain disease, and death by having regular monitoring to detect endocrine and brain abnormalities at early stages when treatment is most likely to be effective.

X-ALD is caused by pathogenic variants in the *ABCD1* gene. X-ALD can take different clinical forms. These include:

•Adrenal Insufficiency: By the time they are adults, most affected men (~85%) will develop some degree of adrenal insufficiency, which usually begins in childhood.

•Childhood Cerebral ALD: Between 33-40% of males affected will develop a rapidly progressive cerebral demyelination in childhood, which is called childhood cerebral ALD (CCALD). CCALD can lead to cognitive loss, blindness, severe disability, and death. CCALD can be treated with HSCT, but treatment is only effective when CCALD is identified at an early stage, when there are brain changes seen on imaging studies but before the development of clinical symptoms. When boys are diagnosed with X-ALD because they are presenting with clinical symptoms related to underlying demyelination, such as inattention or cognitive loss, it is usually too late to treat. Therefore, early presymptomatic diagnosis can allow for close monitoring with MRI so as not to miss the best therapeutic window for providing HSCT.

•Adrenomyeloneuropathy: Nearly all males with X-ALD will develop stiffness in their legs (spasticity) and gait abnormality as adults due to X-ALD's spinal cord effects, causing adrenomyeloneuropathy (AMN).

•Females: the majority of adult women "carriers" will develop some central nervous system effects in adulthood, but they rarely get adrenal disease

X-ALD was nominated for addition to the NBS panel of conditions. The NBS Metabolic subcommittee and the NBS Umbrella Committee considered the nomination at meetings on September 17, 2021, and December 3, 2021, and recommended adding X-ALD to the newborn screening panel. The NBS Umbrella Committee recommendation was then forwarded to the SACNBS, which considered the nomination on March 4, 2022, and voted in support of adding X-ALD to the newborn screening panel. On May 12, 2022, former Secretary-designee Timberlake approved the SACNBS's recommendation to add X-ALD to the newborn screening panel.

MPS-1 is a rare progressive autosomal recessive lysosomal storage disorder also known as Hurler syndrome. The incidence of MPS-1 is estimated to be 1.50 - 1.85 cases per 100,000 newborns. Both males and females are equally affected. There are two forms of MPS-1 - severe and attenuated. Severe MPS-1 can lead to death in the first ten years of life without treatment. Attenuated MPS-1 presents between the ages of 3 and 10, commonly resulting in early death in the second to third decade when untreated, but can also lead to substantial physical or mental disability (e.g. progressive joint or cardiorespiratory issues and/or developmental delay) without an effect on life expectancy. Affected individuals have a deficiency of the lysosomal enzyme a-L-iduronidase ("IDUA") leading to build up of intracellular components, glycosaminoglycans ("GAGs"), eventually leading to tissue damage and organ dysfunction. Treatments for MPS-1 include enzyme replacement therapy and hematopoietic stem cell transplantation (HSCT). MPS-1 was added to the national Recommended Uniform Screening Panel ("RUSP") in 2016.

On March 3, 2023, the SACNBS met via Zoom to review the nomination to add MPS-1 to the Wisconsin mandatory newborn screening panel. MPS-1 was nominated by Dr. Donald Basel, MD (Children's Wisconsin) with co-sponsor Dr. Roberto Mendez, PhD (State of Wisconsin Newborn Screening Laboratory). The nomination was reviewed by Metabolic subcommittee which recommended acceptance, leading to presentation to the Umbrella Committee meeting, on December 2, 2022, which also recommended acceptance. The SACNBS voted unanimously in favor of adding MPS-1 to the Wisconsin NBS panel. On June 15, 2023, the recommendation to add MPS-1 to the newborn screening panel was sent to Secretary-designee Johnson and ultimately approved on June 28, 2023.

## 4. Analysis of policy alternative

Section 253.13 (2), the department is required to impose a fee, by rule, sufficient to cover the costs of tests performed under this section and other services specified. Rule promulgation is necessary to comply with the requirements under s. 253.13(2), Stats., to impose sufficient fees by rule.

The department pursued a funding increase through the 2023-25 biennial budget, but that provision was not adopted by the Legislature. Therefore, there are no reasonable alternatives to address these increases than through proposed rulemaking.

Section 253.13 (1), Stats. requires that every infant born in each hospital or maternity home, prior to the infant's discharge, be tested for congenital and metabolic disorders, as specified in rules promulgated by the department. Experts on the NBS Umbrella Committee and SACNBS recommended adding X-ALD and MPS-1 to the newborn screening panel. Former Secretary-designee Timberlake approved adding X-ALD to that panel on May 12, 2022 and promulgating a rule to add X-ALD to the newborn screening panel while Secretary-designee Johnson approved adding MPS-1 to that panel on June 28, 2023 and promulgating a rule to add MPS-1 to the newborn screening panel. Therefore, there are no reasonable alternatives to the proposed rulemaking.

## 5. Statutory authority for the rule

### a. Explanation of authority to promulgate the proposed rule

The department is authorized to promulgate the rule based upon explicit statutory language.

**b. Statute/s that authorize/s the promulgation of the proposed rule**

Section 227.11 (2), Stats.:

(2) Rule-making authority is expressly conferred on an agency as follows:

(a) Each agency may promulgate rules interpreting the provisions of any statute enforced or administered by the agency, if the agency considers it necessary to effectuate the purpose of the statute, but a rule is not valid if the rule exceeds the bounds of correct interpretation. All of the following apply to the promulgation of a rule interpreting the provisions of a statute enforced or administered by an agency:

1. A statutory or nonstatutory provision containing a statement or declaration of legislative intent, purpose, findings, or policy does not confer rule-making authority on the agency or augment the agency's rule-making authority beyond the rule-making authority that is explicitly conferred on the agency by the legislature.
2. A statutory provision describing the agency's general powers or duties does not confer rule-making authority on the agency or augment the agency's rule-making authority beyond the rule-making authority that is explicitly conferred on the agency by the legislature.
3. A statutory provision containing a specific standard, requirement, or threshold does not confer on the agency the authority to promulgate, enforce, or administer a rule that contains a standard, requirement, or threshold that is more restrictive than the standard, requirement, or threshold contained in the statutory provision.

(b) Each agency may prescribe forms and procedures in connection with any statute enforced or administered by it, if the agency considers it necessary to effectuate the purpose of the statute, but this paragraph does not authorize the imposition of a substantive requirement in connection with a form or procedure.

(c) Each agency authorized to exercise discretion in deciding individual cases may formalize the general policies evolving from its decisions by promulgating the policies as rules, which the agency shall follow until they are amended or repealed. A rule promulgated in accordance with this paragraph is valid only to the extent that the agency has discretion to base an individual decision on the policy expressed in the rule.

(d) An agency may promulgate rules implementing or interpreting a statute that it will enforce or administer after publication of the statute but prior to the statute's effective date. A rule promulgated under this paragraph may not take effect prior to the effective date of the statute that it implements or interprets.

(e) An agency may not inform a member of the public in writing that a rule is or will be in effect, unless the rule has been filed under s. 227.20 or unless the member of the public requests that information.

Section 253.13 (1), Stats.:

(1) TESTS; REQUIREMENTS. The attending physician or nurse licensed under s. 441.15 shall cause every infant born in each hospital or maternity home, prior to its discharge therefrom, to be subjected to tests for congenital and metabolic disorders, as specified in rules promulgated by the department. If the infant is born elsewhere than in a hospital or maternity home, the attending physician, nurse licensed under s. 441.15, or birth attendant who attended the birth shall cause the infant, within one week of birth, to be subjected to these tests.

Section 253.13 (2), Stats.:

(2) TESTS; DIAGNOSTIC, DIETARY AND FOLLOW−UP COUNSELING PROGRAM; FEES. The department shall contract with the state laboratory of hygiene to perform any tests under this section that are laboratory tests and to furnish materials for use in the tests. The department shall provide necessary diagnostic services, special dietary treatment as prescribed by a physician for a patient with a congenital disorder as identified by tests under this section, and follow−up counseling for the patient and his or her

family. The department shall impose a fee, by rule, for tests performed under this section sufficient to pay for services provided under the contract. The department shall include as part of the fee established by rule amounts to fund the provision of diagnostic and counseling services, special dietary treatment, and periodic evaluation of infant screening programs, the costs of consulting with experts under sub. (5), the costs of administering the hearing screening program under s. 253.115, and the costs of administering the congenital disorder program under this section and shall credit these amounts to the appropriation accounts under s. 20.435 (1) (ja) and (jb).

**c. Statute/s or rule/s that will affect the proposed rule or be affected by it**

Section 20.435 (1) (ja) and (jb), Stats.

DHS 115.06 Criteria for adding and deleting conditions.

In determining which disorders are to be added or deleted from s. DHS 115.04, the department shall seek the advice and guidance of medical consultants, staff of the state laboratory and other persons who have expertise and experience in dealing with congenital and metabolic disorders. Criteria to be considered in adding or deleting disorders shall include all of the following:

(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 addition, as a part of the nomination review process, the department seeks the advice and guidance of medical consultants and experts via the NBS Umbrella Committee and SACNBS. The SACNBS forwards recommendations for adding or deleting conditions to the newborn screening panel to the Secretary. Prior to the committee's recommendation and report submission to the Secretary of DHS, the following criteria are reviewed:

1) Mandated testing should be limited to conditions that cause serious health risks in childhood that are unlikely to be detected and prevented in the absence of newborn screening.
2) For each condition, there should be information about the incidence, morbidity and mortality, and the natural history of the disorder.
3) Conditions identified by newborn screening should be linked with interventions that have been shown in well-designed studies to be safe and effective in preventing serious health consequences.
4) The interventions should be reasonably available to affected newborns.
5) Appropriate follow-up should be available for newborns who have a false positive newborn screen.
6) The characteristics of mandated tests in the newborn population should be known, including specificity, sensitivity, and predictive value or other convincing medical evidence (experience, natural history, or literature).
7) If a new sample collection system is needed to add a disorder, reliability and timeliness of sample collection must be demonstrated.
8) Before a test is added to the panel, the details of reporting, follow-up, and management must be completely delineated, including development of standard instructions, identification of consultants, and identification of appropriate referral centers throughout the state/region.

9) Recommendations and decisions should include consideration of the costs of the screening test, confirmatory testing, accompanying treatment, counseling, and the consequences of false positives. The mechanism of funding those costs should be identified. Expertise in economic factors should be available to those responsible for recommendations and decisions.

**6. Estimates of the amount of time that state employees will spend to develop the rule and other necessary resources**

The department estimates that it will take approximately 160 hours to develop the proposed rules. This includes time required for research and analysis, creating the advisory committee list, and coordinating meetings, rule drafting, preparing any related documents, holding a public hearing, and communicating with affected persons and groups.

**7. Description of all of the entities that may be affected by the rule, including any local governmental units, businesses, economic sectors, or public utility ratepayers who may reasonably be anticipated to be affected by the rule**

- purchasers of the newborn screening blood collection cards
- physicians
- nurses
- midwives
- parents of newborns
- insurers
- the Wisconsin State Laboratory of Hygiene
- The Newborn Screening Program
- Wisconsin Hospitals
- Newborns and parents of newborns will benefit from early diagnosis through newborn screening and follow up treatment
- The Wisconsin State Laboratory of Hygiene will provide ongoing testing for additional conditions and the WI Newborn Screening Program will have additional newborn screening conditions to follow up and report on
- Expert consultants in the newborn screening field and health care providers will have a slight increase in patient care for those diagnosed through newborn screening

**8. Summary and preliminary comparison of any existing or proposed federal regulation that is intended to address the activities to be regulated by the rule**

There do not appear to be any existing or proposed federal regulations that address the activities of this rule.

**9. Anticipated economic impact, locally or statewide**

The proposed rule may have a moderate economic impact. An increase to the Newborn Screening card fee is needed by the Wisconsin State Laboratory of Hygiene to cover screening and testing costs and for the department to cover the provision of diagnostic and counseling services, special dietary treatment, and periodic evaluation of infant screening programs, costs of consulting with experts, and an integrated data system and program operation costs.

In FY 24, 25, and 26 it is estimated that there will be approximately 52,000 blood cards annually. Based on current fees, this is an annual projected revenue of $5,668,000 and with the proposed increased a projected annual revenue of $11,596,000. This will cover increased costs experienced by both WSLH and the Department.

**10. Agency contacts**

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