Status Report to the General Assembly

Adding Conditions to the Iowa Newborn Screening Panel

January 1, 2025 – December 31, 2025

Division of Public Health

Center for Congenital and Inherited Disorders

IOWA Health and Human Services

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Citation for Required Status Report

Iowa Code chapter 136A Section 136A.3A Subsection 4 (Link to subsection on the Iowa Legislature website, https://www.legis.iowa.gov/docs/code/136A.pdf) states:

4. The department shall submit a status report to the general assembly, annually, by December 31, regarding all of the following:

a. The current conditions included in the newborn screening.

b. Any new conditions currently under consideration or recommended by the advisory committee for inclusion in the newborn screening.

c. Any new conditions considered but not recommended by the advisory committee in the prior twelve-month period and the reason for not recommending any such conditions.

d. Any departmental request for additional program capacity or resources necessitated by the inclusion of a recommended new condition in the newborn screening.

e. Any delay and the reason for the delay by the advisory committee in complying with the specified twelve-month time frame in considering or recommending the inclusion of a new condition in the newborn screening to the department.

f. Any delay and the reason for the delay by the department in complying with the specified eighteen-month time frame in including a new condition in the newborn screening following receipt of a recommendation from the advisory committee recommending the inclusion of such condition.

Current Conditions on the Iowa Newborn Screening Panel

The Iowa newborn screening panel includes all conditions on the federal Recommended Uniform Screening Panel (RUSP).

The Iowa Newborn Screening Program (INSP) currently screens Iowa newborns for the following conditions:

Amino Acidemias and Urea Cycle Disorders

- (ASA) Argininosuccinic aciduria*
- (CIT) Citrullinemia, type 1 or ASA Synthetase Deficiency*
- (HCY) Homocystinuria (cystathionine beta synthetase)*
- (MSUD) Maple Syrup Urine Disease*
- (PKU) Classic Phenylketonuria*

- (TYR-1) Tyrosinemia, type I*
- (ARG) Argininemia**
- (BIOPT-BS) Defects of biopterin cofactor biosynthesis**
- (CIT-II) Citrullinemia, type II**
- (BIOPT-REG) Defects of biopterin cofactor regeneration**
- (H-PHE) Benign hyperphenylalaninemia**
- (MET) Hypermethioninemia**
- (TYR II) Tyrosinemia, type II**
- (TYR III) Tyrosinemia, type III**

Organic Acidemias

- (GA-1) Glutaric acidemia type I*
- (HMG) 3-Hydroxy 3-methylglutaric aciduria *
- (IVA) Isovaleric acidemia*
- (3-MCC) 3-Methylcrotonyl-CoA carboxylase*
- (Cbl-A,B) Methylmalonic acidemia (cobalamin disorders, vitamin B12 disorders)*
- (βKT) βeta-Ketothiolase*
- (MUT) Methylmalonic Acidemia (methylmalonyl-CoA mutase)*
- (PROP) Propionic acidemia*
- (MCD) Holocarboxylase synthetase*
- (2M3HBA) 2-Methyl-3-hydroxybutyric aciduria**
- (2MBG) 2-Methylbutyrylglycinuria**
- (3MGA) 3-Methylglutaconic aciduria**
- (Cbl-C, D) Methylmalonic acidemia with homocystinuria**
- (MAL) Malonic acidemia**

Fatty Acid Oxidation Disorders

- (CUD) Carnitine uptake defect (Carnitine transport defect)*
- (LCHAD) Long-chain L-3 hydroxyacyl-CoA dehydrogenase*
- (MCAD) Medium chain acyl-CoA dehydrogenase*
- (TFP) Trifunctional protein deficiency*

- (VLCAD) Very long-chain acyl-CoA dehydrogenase*
- (CACT) Carnitine acylcarnitine translocase**
- (CPT-Ia) Carnitine palmitoyltransferase type I**
- (CPT-II) Carnitine palmitoyltransferase type II**
- (GA2) Glutaric acidemia type II**
- (MCAT) Medium-chain ketoacyl-CoA thiolase**
- (M/SCHAD) Medium/Short chain L-3-hydroxyacyl-CoA dehydrogenase**

Endocrine

- (CAH) Congenital adrenal hyperplasia*
- (CH) Primary Congenital hypothyroidism*

Lysosomal Storage Disorders

- Pompe Disease (glycogen storage disease type II)*
- (MPSI) Mucopolysaccharidosis Type I*
- (MPS II) Mucopolysaccharidosis Type II*
- Krabbe disease (globoid leukodystrophy)*

Hemoglobinopathies

- (Hb SS) S,S Disease (Sickle Cell Anemia)*
- (Hb S/C) S,C Disease*
- (HB S/βTh) S, βeta-thalassemia*
- (Var Hb) Variant hemoglobinopathies**

Other

- (BIOT) Biotinidase deficiency*
- (CF) Cystic Fibrosis*
- (GALT) Classic Galactosemia*
- (SCID) Severe Combined Immunodeficiency*
- (HEAR) Hearing loss*
- (CCHD) Critical Congenital Heart Disease*
- (SMA) Spinal Muscular Atrophy*
- X-Linked Adrenoleukodystrophy (XALD)*

- (GAMT) Guanidinoacetate methyltransferase deficiency*

*US Department of Health and Human Services Recommended Uniform Screening Panel (RUSP) - Core Panel.

**US Department of Health and Human Services Recommended Uniform Screening Panel (RUSP) - Secondary Targets. Conditions on the secondary target list are not directly targeted by screening but are routinely identified through screening for disorders on the Core Panel (by-products of screening for the core conditions).

New conditions currently under consideration or recommended by the Congenital and Inherited Disorders Advisory Committee for inclusion in the Iowa Newborn Screening Panel

The Iowa HHS Council Ad Hoc Committee for Management of the Iowa Newborn Screening Panel (Committee) is beginning a review of Metachromatic Leukodystrophy (MLD), with the first meeting on November 19, 2025.

MLD is not on the RUSP, but advocacy organizations have made a recommendation to the Secretary of the US Department of Health and Human Services to have it added. In anticipation of this decision and based on an evidence-based review published in the Pediatrics journal, INSP leadership recommended advancing MLD for review by the Committee. It is anticipated that the Committee will have a recommendation regarding the addition of MLD to Iowa's newborn screening panel in February 2026.

New conditions considered but not recommended by the advisory committee in the prior twelve-month period and the reason for not recommending any such conditions

There are no conditions that have been considered but not recommended for inclusion in the newborn screening panel at this time.

Departmental request for additional program capacity or resources necessitated by the inclusion of a recommended new condition in the newborn screening

Department – Committee members and ad hoc reviewers serve on a volunteer basis and receive no compensation for their commitment. The average time commitment for a committee member to this effort was 20 hours over the last year.

State Hygienic Laboratory (SHL) – Due to an increasing number of tests, the SHL newborn screening laboratory purchased an additional instrument to increase its capacity to conduct tandem mass spectrometry (MS/MS) testing. There is an increase in costs for calibration and maintenance of the instruments utilized for the testing of new conditions. Screening for Krabbe disease used existing equipment and is conducted by existing staff but required re-validation of the existing lysosomal storage disorder testing panel. The addition of new conditions requires the purchase of new reagents and testing kits. INSP leadership made an adjustment to the newborn screening fee in 2025 to account for the increased costs.

Short-term Follow-up (STFU) – Physician experts for Krabbe disease and GAMT serve as the medical consultants for these disorders. The future addition of new conditions will require additional medical consultant expertise. There is a need to educate newborn screening stakeholders about new conditions. This requires developing educational resources (brochure inserts, notices to birthing facilities, newsletter articles, press releases, etc.) as well as additional staff time to carry out these tasks. While no additional STFU or genetic counselors were needed to staff the caseload work of adding Krabbe disease or GAMT, staff workload will need to be reallocated to complete the necessary educational/informational tasks for the future addition of new conditions.

Additional Resources Needed as New Conditions are Added

Fixed costs for implementation of newborn screening are increasing. The SHL is currently close to capacity in its physical space to accommodate the additional equipment, supporting instruments and staff for testing new conditions. As new conditions are added, SHL will need to purchase additional testing equipment, such as those used for tandem mass spectrometry (MS/MS); and 2nd tier testing to validate the newborn screening results for these conditions is often molecular-based and requires specific space, equipment and staff. The SHL will need building modifications to continue to expand.

Short-term follow-up staff will need to engage medical consultants with experience in caring for newborns with the new condition to advise the SHL on testing methodology and cut-offs, establish standard treatment algorithms, and manage the care of the newborn.

The addition of new conditions increases the volume of newborn screening testing so SHL and STFU will need to hire more staff accordingly.

Delay and the reason for the delay by the advisory committee in complying with the specified twelve-month time frame in considering or recommending the inclusion of a new condition in the newborn screening to the department

There has been no delay in complying with the twelve-month time frame in considering or recommending the inclusion of a new condition in the newborn screening panel to the Department.

Any delay and the reason for the delay by the department in complying with the specified eighteen-month time frame in including a new condition in the newborn screening following receipt of a recommendation from the advisory committee recommending the inclusion of such condition

It has taken a large effort to add six new conditions to Iowa's newborn screening panel since 2022, and SHL staff have needed to commit a considerable amount of time to building new test methodologies and screening protocols and validating the tests. Guanidinoacetate methyltransferase deficiency (GAMT) was added to the Iowa newborn screening panel in September 2023. Even though GAMT was added to the Iowa newborn screening panel before Krabbe disease, leadership of the INSP decided to move Krabbe disease ahead of GAMT in the test development queue. Reasons for this decision:

- The testing platform for Krabbe disease is the same as what is required for testing of conditions already screened, so test development for Krabbe disease could be expedited.
- Krabbe disease is considered a time-critical condition, meaning that the newborn will need to receive life-saving treatments within the first 30 days of life. GAMT is not a time-critical condition.
- The incidence of Krabbe disease is estimated to be 1 in 400,000 births. The incidence of GAMT is estimated to be 1 in 550,000 – 2 million births.

The INSP is currently conducting a screening pilot for Krabbe disease. With the implementation of the screening pilot for Krabbe disease, SHL and short-term follow-up staff can turn their full attention to GAMT. The test development for GAMT will require

re-calibration and validation of other conditions on the same testing platform as GAMT, which may take four to five months to complete.

Review of the Iowa Newborn Screening Panel

Leadership of INSP reviewed the current Iowa newborn screening panel in November 2025. INSP leadership made a recommendation to Committee members that no changes to the Iowa newborn screening panel are needed at this time. Committee members accepted the recommendation in November 2025.