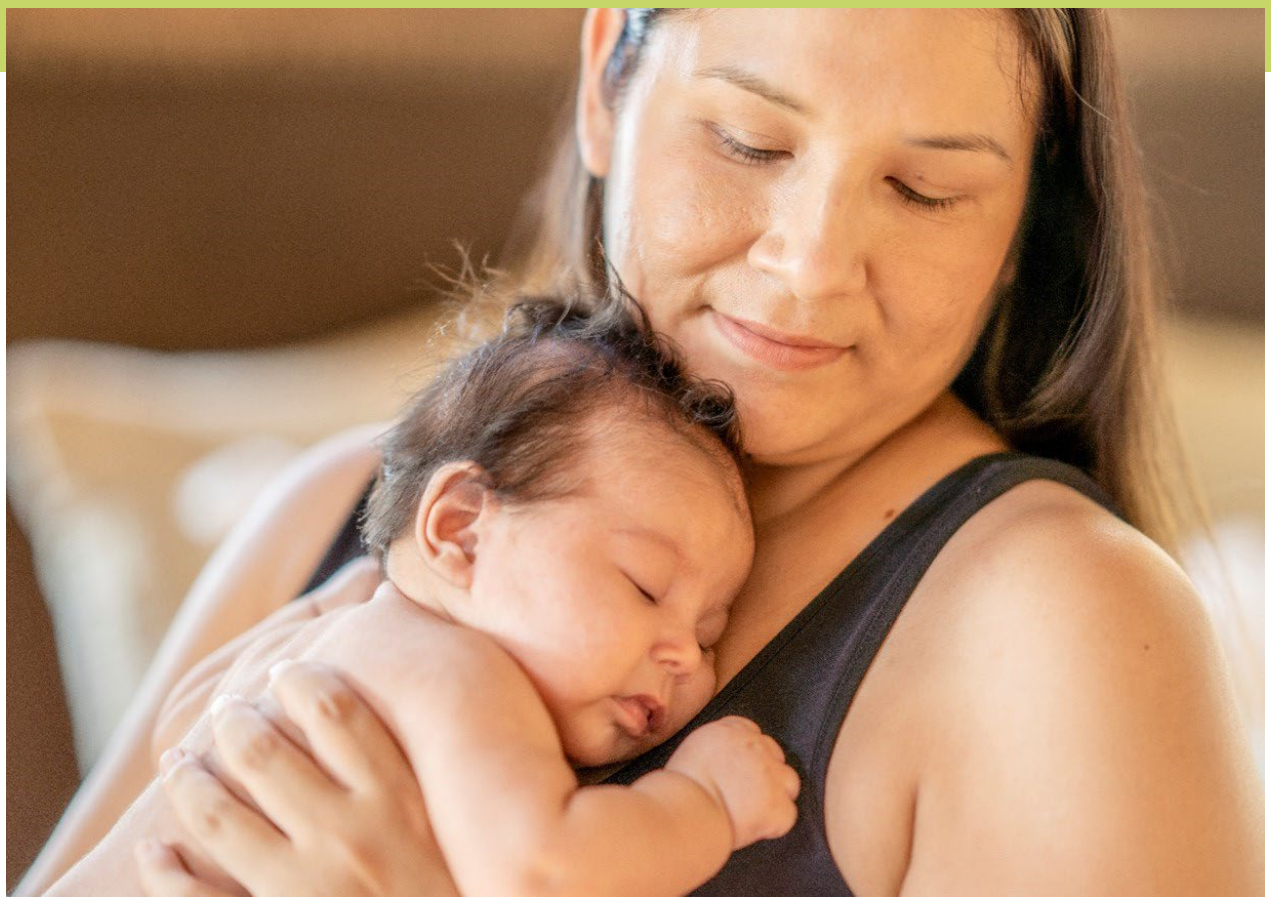

Quarterly Report to the Iowa Health and Human Services Council

Committee for Management of the Iowa Newborn Screening Panel

Division of Public Health

Center for Congenital and Inherited Disorders

May 2025

IOWA Health and Human Services

Mission Statement

The Iowa Health and Human Services Council Committee for the Management of the Iowa Newborn Screening Panel supports the Iowa Newborn Screening Program to provide comprehensive, high quality, relevant, and timely newborn screening services that ensure the optimal health of Iowa newborns and their families.

Committee Membership

Dr. Amy Calhoun – Medical Geneticist, Iowa Newborn Screening Program Medical Director
Department of Pediatrics, University of Iowa Health Care Stead Family

Carol Cross – Parent of children with an inherited condition

Ken Coursey – Manager, Newborn Screening Laboratory
State Hygienic Laboratory, University of Iowa

Dr. Amy Ferguson – Pediatric Hospitalist and Medical Director
UnityPoint Blank Children's Hospital and UnityPoint Ft. Dodge Newborn Nurseries

Jeanette Hasley – Certified Nurse Midwife
Cherokee Regional Medical Center

Kimberly Noble Piper – Director, Center for Congenital and Inherited Disorders
Bureau of Chronic, Inherited and Congenital Conditions, Division of Public Health, Iowa Department of Health and Human Services

Jeremy Penn, Ph.D. - Parent of child with an inherited condition
Director of Assessment and Continuous Improvement, University of Iowa College of Education

Jaclyn Zamzow – Genetic Counselor (licensed and certified)
Division of Medical Genetics and Genomics, University of Iowa Health Care Stead Family Department of Pediatrics

To be added - Additional ad hoc members with relevant experience and expertise to the specific condition being reviewed, e.g., physician with experience treating individuals with the condition, parent of child or individual with the condition being reviewed.

Committee Activity

The committee has not met since established, but communications have occurred by email where members re-affirmed the condition review process and decision matrix and

have considered the elimination of the Advisory Committee for Heritable Disorders in Newborns and Children.

At the federal level, the Advisory Committee for Heritable Disorders in Newborns and Children (ACHDNC) has been terminated. This is the body that was responsible for making recommendations to the US Health and Human Services (US HHS) Secretary regarding the addition of conditions to the Recommended Uniform Screening Panel (RUSP). An ACHDNC external evidence-based review group was building a recommendation to add metachromatic leukodystrophy (MLD) to the RUSP. Members of the evidence-based review group have stated they plan on finalizing the recommendations and forwarding them directly to the US HHS Secretary for his consideration. The status of RUSP as a US HHS supported newborn screening panel is unknown.

Current Conditions on the Iowa Newborn Screening Panel

The following conditions are on the Iowa newborn screening panel:

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* - pilot screening
- Krabbe disease (globoid leukodystrophy)* - pilot begins summer 2025

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* - pilot begins Fall 2025

Review of the Iowa Newborn Screening Panel

The Committee convenes at least annually to review the effectiveness and appropriateness of the current newborn screening panel.