Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)

Report to Congress (2022)

TABLE OF CONTENTS

EXECUTIVE SUMMARY

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC or the Committee) was formed to advise and provide evidence-based recommendations to the Secretary of the United States (US) Department of Health and Human Services (HHS Secretary) regarding the best applications of newborn screening tests, technologies, policies, guidelines, and standards.

The Committee's advice and recommendations are intended for use by the HHS Secretary toward the development of policies and priorities that enhance states' abilities to reduce morbidity and mortality in newborns and children who have, or who are at risk for, heritable conditions that can be present at birth. Left undetected, these conditions can cause significant harm to the child, including disability or death. Newborn screening saves lives and improves quality of life throughout the lifespan.

The Health Resources and Services Administration (HRSA) provides coordination, management, and operational services to the Committee. This review of the Committee's activities and accomplishments in calendar year 2022 fulfills a legislative requirement to submit an annual report to Congress and other federal and state stakeholders.

Selected highlights from the Committee's work in 2022 include:

- Recommending the inclusion of Mucopolysaccharidosis type II (MPS II) and Guanidinoacetate Methyltransferase (GAMT) deficiency on the Recommended Uniform Screening Panel (RUSP).
- Beginning its review of evidence for Krabbe disease to determine a recommendation for its inclusion to the RUSP.
- Receiving and initiating its review of two nominations, for Duchenne muscular dystrophy and Congenital Cytomegalovirus (cCMV).
- Considering its capacity for providing a comprehensive review of new nominations.
- Hearing multiple presentations on the facilitators and barriers to the implementation of newly added RUSP conditions.
- Developing an initial set of recommendations to strengthen the newborn screening system for the early and effective detection of genetic, congenital, and metabolic disorders.
- Deliberating on systemic issues that underlie public health crises, such as the formula shortage and chronic inequities and disparities in newborn screening, diagnosis, and treatment.

KEY ABBREVIATIONS AND TERMS

Term	Definition
AAP	American Academy of Pediatrics
ACHDNC	Advisory Committee on Heritable Disorders in Newborns and Children. Also referred to as "the Committee".
APHL	Association of Public Health Laboratories
CDC	Centers for Disease Control and Prevention
Congenital CMV	Congenital cytomegalovirus: a type of herpes virus that can be passed to a fetus during pregnancy.
DoD	Department of Defense
Duchenne muscular dystrophy	A genetic disorder characterized by muscle damage and weakness.
HHS	Department of Health and Human Services
Heritable disorders	A group of genetically inherited conditions present at birth that, undetected, can cause intellectual and physical disabilities and life-threatening illness.
Homocystinuria	A genetic condition that prevents the body from processing proteins correctly.
HRSA	Health Resources and Services Administration
GAMT deficiency	Guanidinoacetate Methyltransferase Deficiency: A genetic condition that prevents the body from making creatine, an essential protein.
Krabbe disease	A genetic condition that destroys myelin in nerve cells.
MCHB	Maternal and Child Health Bureau
MPS I	Mucopolysaccharidosis type I: a group of genetic conditions that prevent the body from processing sugars properly.
MPS II	Mucopolysaccharidosis type II: a group of genetic conditions that prevent the body from processing sugars properly.
Newborn Screening Family Education Program	A HRSA-funded program dedicated to ensuring all families have access to newborn screening education and training.
NewSTEPS	Newborn Screening Technical Assistance and Evaluation Program: a HRSA-funded program to provide data, technical assistance, and training to newborn screening programs.
Pompe disease	A genetic condition that results in the build-up of glycogen in the body.
RUSP	Recommended Uniform Screening Panel: a standard guideline for newborn screening of genetic conditions approved by the HHS Secretary.
SCID	Severe combined immunodeficiency: a group of genetic conditions resulting in a poorly working immune system at birth.

REPORT

Introduction

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC or the Committee) was formed to advise and provide evidence-based recommendations to the Secretary of the United States (US) Department of Health and Human Services (HHS Secretary) regarding the best applications of newborn screening tests, technologies, policies, guidelines, and standards. As part of this mission, the Committee provides the Secretary:

- Recommendations and advice regarding grants and projects funded, awarded, or authorized for the screening of heritable disorders in newborns and children.
- Technical information required to develop policies and priorities for the Heritable Disorders Program meant to enhance the screening, counseling, and health care services provided at the state and local levels for newborns and children who either have or are at risk for heritable disorders.
- Advice, recommendations, and information designed to enhance, expand, or improve the Secretary's ability to reduce mortality and morbidity from heritable disorders in newborns and children.

The Health Resources and Services Administration (HRSA) provides coordination, management, and operational services to the Committee. This report, which summarizes the Committee's activities for calendar year (CY) 2022, fulfills the legislative requirement for the submission of an annual report to Congress, the Secretary, the Interagency Coordinating Committee on Newborn and Child Screening, and state health departments.

The discussion of the Committee's activities in this report is subdivided into sections aligned with the Committee's legislatively mandated duties. For ease of reference, the specific legislation related to each activity is presented alongside descriptions of these activities.

ACHNDC Activities

Section 1. Advice, Technical Information, and Systematic Evidence-Based and Peer-Reviewed Recommendations

The Advisory Committee shall

(1) provide advice and recommendations to the Secretary concerning grants and projects awarded or funded under section 300b-8 of this title

(2) provide technical information to the Secretary for the development of policies and priorities for the administration of grants under section 300b-8 of this title

(3) make systematic evidence-based and peer-reviewed recommendations that include the heritable disorders that have the potential to significantly impact public health for which all newborns should be screened, including secondary conditions that may be identified as a result of the laboratory methods used for screening

In CY 2022, the Committee recommended the addition of two conditions to the Recommended Uniform Screening Panel (RUSP) and began or continued review of three other nominated conditions.

Section 1.1 Mucopolysaccharidoses Type II (MPS II)

In CY 2021, the Committee voted to move MPS II to a full evidence-based review by the external Evidence-Based Review Group (ERG), funded under a HRSA contract. MPS II is a genetic condition that leads to inadequate quantities of an enzyme called iduronate-2-sulfatase, which is needed to break down long-chain sugars called glycosaminoglycans. The accumulation of glycosaminoglycans results in progressive and permanent damage to organ function, physical abilities, and cognition. In its severe form, MPS II also leads to intellectual disability and significant behavioral problems. Approximately 60 percent of individuals with MPS II have the severe form of the disease.

In February 2022, the Committee evaluated the results and findings of the evidence review. They found that, although infants with MPS II did not show symptoms at birth, significant symptoms would appear between the ages of two to four years if left untreated. With the availability of a reliable screening test and an effective treatment to slow the progression of disease, the Committee voted to recommend the inclusion of MPS II as a core condition on the RUSP. HHS Secretary Becerra accepted the Committee's recommendation on August 2, 2022.

Section 1.2. Guanidinoacetate Methyltransferase (GAMT) Deficiency

In CY 2021, the Committee voted to move GAMT deficiency to a full evidence-based review by the ERG. GAMT is an enzyme that is important for the synthesis of a protein called creatine. GAMT deficiency is a genetic condition that is associated with elevated guanidinoacetate and leads to a deficiency in creatine, resulting in progressive neurological impairments such as significant intellectual disability, limited speech development, recurrent seizures, movement disorders, weakness, and behavioral problems.

In May 2022, the Committee determined that the availability of a reliable screening test and an accessible, low-cost treatment could support better neurological, cognitive, and functional outcomes in infants identified with GAMT deficiency. The Committee therefore voted to recommend the inclusion of GAMT deficiency as a core condition on the RUSP.

Section 1.3. Krabbe Disease

In CY 2021, the Committee's Nomination and Prioritization Workgroup reviewed a nomination for Krabbe disease. In February 2022, the Committee voted to move Krabbe disease forward for a full evidence-based review. Krabbe disease, or globoid cell leukodystrophy, is a genetic disorder that causes reduced activity in an enzyme called galactocerebrosidase, which leads to an accumulation of a lipid called psychosine and

results in demyelination of neural cells. Without treatment, most children with Krabbe disease will die within two years of symptom onset.

The ERG presented interim findings from its review of evidence to the Committee in August and November 2022. The ERG will present its final review at the February 2023 Committee meeting for Committee deliberation and vote on the Krabbe disease nomination.

Section 1.4. Congenital Cytomegalovirus (cCMV)

The Nomination and Prioritization Workgroup conducted a review of the cCMV nomination that was submitted in October 2021. CCMV occurs when an infant is born with cytomegalovirus (CMV), a common herpes virus that often has no symptoms. Although most infants do not develop health problems as a result of cCMV, some will be born with or later develop damage to the brain, ears, and other organs.

In August 2022, the Committee determined that additional information was required from the nominators in order to make a decision whether to advance the nomination for full evidence review.

Section 1.5. Duchenne Muscular Dystrophy (DMD)

The Committee's Nomination and Prioritization Workgroup began its review of the DMD nomination that was submitted in June 2022. DMD is a genetic disorder characterized by muscle damage and weakness.

It is anticipated that the Committee will vote on whether to advance the DMD nomination for full evidence review in February 2023.

Section 1.6. RUSP Condition Nomination and Evidence Review Process

Capacity and Prioritization Workgroup

In February 2022, the Committee recognized the possibility that the number of nominated conditions could outpace the Committee's capacity to conduct a thorough evidence-based review. The Committee's current capacity is to conduct two full evidence-based reviews annually. The Committee considered prioritizing nominations for review as a potential strategy to mitigate this challenge. In August 2022, the Committee initiated a Capacity and Prioritization Workgroup, comprised of current and former Committee members, to discuss and develop prioritization criteria. In November 2022, the Chair provided a brief update on this workgroup's activities. The workgroup will provide an update in February 2023.

Section 2. Technical Assistance and Nomination Review

The Advisory Committee shall

(4) provide technical assistance, as appropriate, to individuals and organizations regarding the submission of nominations to the uniform screening panel, including prior to the submission of such nominations

(5) take appropriate steps, at its discretion, to prepare for the review of nominations prior to their submission, including for conditions for which a screening method has been validated but other nomination criteria are not yet met, in order to facilitate timely action by the Advisory Committee once such submission has been received by the Committee

Section 2.1. Technical Assistance to Nominators

The Committee provides technical assistance to nominators. If potential nominators need guidance in their development of a complete nomination package or if a submitted nomination package is missing information, the Committee will support the nominators with ongoing technical assistance by responding to questions and clarifying the types of data and information that should be included in the nomination. The Committee also provides technical assistance for nomination packages that are not moved forward for evidence review to assist in future resubmission. In CY 2022, the Committee provided direct technical assistance to the nominators who submitted nomination packages for cCMV, Krabbe disease, and DMD.

In November 2021, the Committee approved new consumer-friendly guidance and resources for RUSP nominators. The updated content included easy-to-follow information and graphics, sample key questions addressed in a full evidence review, and a description of the Committee's evaluation process. The updated guidance resources were made available to the public on the ACHDNC website in January 2022.

Section 2.2. Updated Nomination Form

In November 2021, the Committee approved an update to its Nomination Form to ensure that it was more consumer-friendly. The updates included requesting more information about the condition to strengthen the case definition, including current standards of care for treatment and availability of follow-up treatment; more detail about confirmatory testing; and more data to assist with evidence review. The updated fillable PDF form was made available to the public on the ACHDNC website in January 2022.

Section 3. Decision Matrix

The Advisory Committee shall

(6) develop a model decision-matrix for newborn screening expansion, including an evaluation of the potential public health impact, including the cost of such expansion, and periodically update the recommended uniform screening panel, as appropriate, based on such decision matrix

In November 2021, the Committee provided input on an updated Decision Matrix that offered more guidance on the criteria for assessing net benefit and implementation feasibility. The updated Decision Matrix was utilized by the Committee in CY 2022. A description of the updated Decision Matrix was also made available to the public on the ACHDNC website in January 2022.

Section 4. State Capacity to Screen

The Advisory Committee shall

(7) consider ways to ensure that all States attain the capacity to screen for the conditions described in paragraph (3), and include in such consideration the results of grant funding under section 300b-8 of this title

The Committee recognized that variation across states in the implementation of RUSP conditions could create inequities among families in their access to early identification and treatment. Several factors are involved in a state decision to initiate newborn screening for a RUSP condition. Differences in laboratory capacity, technology, available funding and staff, and access to long-term follow-up services and treatment can impact both the state decision to initiate a new screen and its successful implementation within the state's existing newborn screening program.

To address these systemic challenges, the Committee discussed the facilitators and barriers of successful implementation, the multiple components and stakeholders involved in implementation, and the need for standardization across states toward reducing inequities.

Section 4.1 Strengthening Newborn Screening Systems

Diagnosis and Treatment of RUSP Disorders

After an infant screens positive for a RUSP condition, there is a need for collaboration and coordination across newborn screening programs, genetic counselors, follow-up coordinators, and clinicians in order to support a confirmatory diagnosis and to initiate appropriate treatment.

To better understand challenges experienced by states in providing short- and long-term follow-up, the Committee heard a presentation from a genetic counselor and follow-up coordinator for Colorado and Wyoming. The Committee learned that confirmatory diagnostics have become more complex, leading to delays and uncertainty among families. States differ in their ability to conduct confirmatory diagnostics and there can be considerable financial and travel burdens placed on families in their journey towards a confirmed diagnosis, particularly if the family must travel to another state. Solutions include increased access to multidisciplinary teams, state monitoring to verify families are engaged in follow-up, and better coordination between state and clinical providers.

System Factors Impacting Implementation

The Director of the North Carolina State Public Health Laboratory provided a presentation to the Committee describing the myriad of systemic, interlinking factors involved in the state implementation of a RUSP condition. Newborn screening exists within the intersection of public health and medicine and is influenced by stakeholders such as legislators, payers, vendors, and consultants. The successful implementation of newborn screening relies on the coordination of systems such as state leadership, legislative mandates and regulations, funding, supply chains, and the availability of a sufficient public health and clinical workforce. These systemic factors impact the ability to hire, train, and sustain personnel and to procure the equipment and technologies necessary for successful implementation of a newborn screen.

As a result of these complex systemic factors and the lack of a mandate to implement RUSP conditions, states may choose to delay implementation of a condition indefinitely. Early adopters of a new RUSP condition typically require state Department of Health approvals, compliance with state legislative requirements for screening, secured funding, collaboration with other states, and active stakeholder engagement—all of which support staff hiring and training, laboratory equipment and assay procurement, and the development of short- and long-term guidelines.

Recommendations from Committee Workgroups

In CY 2022, Committee workgroups offered suggestions to the Committee for strengthening the newborn screening system. These suggestions included developing protocols and guidelines for states, receiving technical assistance from federal agencies such as the Centers for Disease Control and Prevention (CDC) to help with screening test validation, holding national training events to allow states to share and learn best practices, conducting grant writing workshops, and modifying the condition nomination package to include a plan for follow-up and treatment.

State Implementation of Recently Added RUSP Conditions

A roundtable discussion with representatives from the California, Florida, Oklahoma, Texas, and Washington newborn screening systems presented on the state-specific facilitators and barriers involved in the implementation of newly added RUSP conditions. Highlighted facilitators included legislative and programmatic factors (i.e., online newborn screening program and established contracts for confirmatory testing), advisory councils to assess a condition for implementation on a state's newborn screening panel, and feasibility and assessment processes for adding new conditions. Highlighted barriers included workforce challenges, funding required to implement a newly added condition, limited timeframes for laboratory preparation in states with a statutory requirement to adopt a new condition, and state-specific legislative requirements. The state representatives reported that it typically takes between 18 months and two years depending on availability of funding and capacity to implement screening for a new condition.

Section 4.2 Lessons Learned from the Department of Defense Newborn Screening Program

The Department of Defense (DoD) presented on its policies for newborn screening, the availability and coverage of genetic testing, and strategic plans to expand testing and laboratory services.

DoD's Defense Health Agency is an organization mandated by Congress to lead military health care delivery, including the TRICARE Health Plan as a payer. Currently, each military service has its own newborn screening policy. Military treatment facilities within the continental US provide genetic testing and counseling, send most genetics tests to a single laboratory, and are encouraged to follow their state's newborn screening program. Military treatment facilities outside the continental US typically send newborn screens to different laboratories across the US, but may provide diagnosis and treatment locally.

Ongoing challenges in newborn screening services across DoD include conditional TRICARE coverage for genetic counseling, state disparities that are aligned with disparities in civilian newborn screening, and challenges in providing services to areas without a military treatment facility or that are outside of the continental US. The Defense Health Agency is currently developing strategies to expand coverage and availability of genetic testing and counseling, address workforce challenges, adopt best practices to reduce variation and disparities, and promote data sharing through its electronic health records system.

Section 5. Recommendations, Advice, or Information to Reduce Morbidity and Mortality

The Advisory Committee shall

(8) provide such recommendations, advice or information as may be necessary to enhance, expand or improve the ability of the Secretary to reduce the mortality or morbidity from heritable disorders, which may include recommendations, advice, or information dealing with the following.

The Committee provides the Secretary with recommendations, advice, and information on a broad range of topics related to newborn screening to reduce newborn and child mortality from heritable disorders. Sections 6 to 17 describe activities that fall under this charge that were undertaken or overseen by the ACHDNC in CY 2022.

Section 6. Follow-up Activities

(A) follow-up activities, including those necessary to achieve best practices in rapid diagnosis and appropriate treatment in the short-term, and those that ascertain long-term case management outcomes and appropriate access to related services.

Long-Term Follow-Up

Long-term follow-up in newborn screening is the process of monitoring identified children to ensure they have access to the resources and services necessary for optimal outcomes. To better understand best practices and potential challenges, the New York and Connecticut newborn screening programs presented on their HRSA-funded long-term follow-up projects for severe combined immunodeficiency (SCID) and other newborn screening conditions.

New York's long-term follow-up program began with a study to demonstrate the effectiveness of a long-term follow-up care model. The model included standardized protocols and integrated data collection aimed to support the provision of coordinated care and ongoing quality improvement in the state's newborn screening program. Individuals enrolled in the program were diagnosed with an inherited metabolic disorder and will be monitored until age 18 to determine long-term outcomes. After initial success, the program expanded its sites and staff, developed educational resources for families, and launched a social media campaign to promote awareness of the program. One of their major challenges was a lack of existing long-term follow-up datasets and standardized common data elements, which necessitated the development and ongoing updates of standard data collection protocols.

Connecticut's long-term follow-up program was developed to improve family-centered care, increase community collaboration, and improve chronic disease management through a focus on continual monitoring, preventive care, long-term follow-up data collection, and the transition from pediatric to adult care. To achieve these objectives, the program engages a family advisory group to support a family-centered workflow, a program evaluator to identify needs and quality improvement areas, and community partners to improve care coordination and advocacy. Additionally, the program developed a registry dashboard to help monitor patient care and document performance and outcome metrics. The registry dashboard is also integrated with state health data to allow providers to view primary care metrics.

Section 7. Implementation, Monitoring, and Evaluation

(B) implementation, monitoring, and evaluation of newborn screening activities, including diagnosis, screening, follow-up, and treatment activities.

The Committee did not undertake activities related to implementation, monitoring, and evaluation in CY 2022.

Section 8. Diagnostic and Other Technology

(C) diagnostic and other technology used in screening.

Diagnostic Tests for Homocystinuria

Researchers from the University of Utah, the Mayo Clinic, and CDC presented on the status of newborn screening for homocystinuria which is a group of amino acid metabolism disorders that can result in visual, skeletal, and neurological impairments. Although newborn screening tests for homocystinuria are available and effective, there are some challenges with the use of these tests, including lower sensitivity and specificity and increased rates of false positives.

One available solution for these challenges is second tier testing for total homocystinuria, which is a sensitive, accessible, cost-effective approach that can be multiplexed with other biomarkers and can reduce the number of false positives. However, second tier testing has not been widely adopted by state newborn screening programs. To overcome this barrier, a universal screening assay or a regional second tier testing program could streamline second tier testing and increase its adoption. Committee members suggested further discussion on a potential recommendation for second tier testing of total homocystinuria, which could help improve innovative homocystinuria screening approaches in the future.

Section 9. Availability and Report of Testing

(D) the availability and reporting of testing for conditions for which there is no existing treatment, including information on cost and incidence.

The Committee did not undertake activities related to the availability and report of testing in CY 2022.

Section 10. Conditions Not Included in the RUSP

(E) conditions not included in the recommended uniform screening panel that are treatable with Food and Drug Administration-approved products or other safe and effective treatments, as determined by scientific evidence and peer review.

The Committee did not undertake activities related to conditions not included in the RUSP in CY 2022.

Section 11. Minimum Standards and Related Policies and Procedures

(F) minimum standards and related policies and procedures used by State newborn screening programs, such as language and terminology used by State newborn screening programs to include standardization of case definitions and names of disorders for which newborn screening tests are performed.

The Committee did not undertake activities related to minimum standards and related policies and procedures in CY 2022.

Section 12. Quality Assurance, Oversight, and Evaluation

(G) quality assurance, oversight, and evaluation of State newborn screening programs, including ensuring that tests and technologies used by each State meet established standards for detecting and reporting positive screening results.

The Committee did not undertake activities related to quality assurance, oversight, and evaluation in CY 2022.

Section 13. Public and Provider Awareness and Education

(H) public and provider awareness and education.

The Newborn Screening Family Education Program

The Newborn Screening Family Education Program, a HRSA-funded initiative to promote equitable access to newborn screening education and information, presented an overview of their program and research initiatives to the Committee. The program aims to ensure that all families, especially medically underserved families, have access to the knowledge they need about newborn screening when and where they need it.

The program conducts studies to better understand family needs, preferences, and understanding of educational materials. One study indicated that many families prefer to receive information about newborn screening prior to labor and delivery. Another study conducted in collaboration with community-based prenatal groups in different communities found that there was a need to understand the community preferences for receiving educational materials, to develop culturally-relevant and relatable material, and to integrate the provision of educational materials into existing workflows. With these considerations, the program found that the information they provided to families helped increase their awareness, knowledge, and confidence in talking to their doctors.

Section 14. Cost Effectiveness

(I) the cost and effectiveness of newborn screening and medical evaluation systems and intervention programs conducted by State-based programs.

The Committee did not undertake activities related to cost and effectiveness in CY 2022.

Section 15. Causes, Public Health Impacts, and Risk Factors

(J) identification of the causes of, public health impacts of, and risk factors for heritable disorders.

Health Equity in Newborn Screening

NewSTEPs, a newborn screening technical assistance center that is a program of the Association of Public Health Laboratories, funded by HRSA via a cooperative agreement, presented on results of preliminary analysis of quality indicators on health equity. The Committee reviewed results of an analysis of nearly 30,000 cases entered into the NewSTEPS data repository by 46 state newborn screening programs. The data highlighted racial disparities in time-sensitive conditions related to the time between birth to diagnosis and between birth to intervention. The Committee discussed the implications of these findings and suggested that further research is needed to better understand the disparities and to target interventions that bridge access to diagnosis and treatment.

Infant Formula Shortage

The Committee heard presentations on the CY 2022 infant formula shortage and discussed the negative outcomes, underlying factors, and future considerations for the equitable access to medical foods. A representative from the American Academy of Pediatrics (AAP) provided information on the background of the infant formula shortage, which began with a recall of specialty formulas in one manufacturing plant and overwhelmed families, pediatricians, metabolic dieticians, and hospitals who struggled to find substitutions. AAP responded to the public health crisis with guidance for families and pediatricians on appropriate substitutions, safety warnings, and updates to initiatives such as Operation Fly Formula. The Committee also heard a presentation from a metabolic dietician, who provided an overview of the successes and challenges from the Oregon Formula Program's response to the shortage.

Family members talked about their lived experiences with the infant formula shortage, sharing stories about the high levels of anxiety and stress created by the crisis as a result of limited or no access to necessary medical foods and formulas. As a result of the shortage, some individuals were unable to access their required formulas, some had to switch formulas, and some limited their formula intake to extend their supply. The Committee discussed the systemic factors underlying the formula shortage, such as the limited number of specialty formula manufacturers and manufacturing facilities. They highlighted that the shortage was a result of ongoing systemic issues that required planning and preparedness to avoid another formula crisis in the future.

Blueprint for Change: A National Framework for a System of Services for Children and Youth with Special Health Care Needs

The HRSA Maternal and Child Health Bureau presented the "Blueprint for Change: A National Framework for a System of Services for Children and Youth with Special Health Care Needs." The objective of the framework is to advance a vision that children and youth with special health care needs enjoy a full life from childhood through adulthood; thrive in a system that supports families and their social, health, and emotional needs; and are assured dignity, autonomy, independence, and active participation in their communities.

Four key focus areas serve as the framework's foundation: health equity, quality of life and well-being, access to services, and financing of services. The framework emphasizes care integration, health equity, family-defined outcomes, human-centered design, and consistent approaches across HRSA. The framework also calls for outcome measures that are meaningful to families, a system built around the needs of children and their families, consideration for the upstream factors that contribute to inequities, and a system that supports equitable and affordable access to services. The Committee reviewed case examples of the framework in action across both state and federal initiatives. They also discussed how to frame their deliberations using the framework to guide a holistic view of specialty care needs and access to care.

Section 16. Coordination of Surveillance Activities

(K) coordination of surveillance activities, including standardized data collection and reporting, harmonization of laboratory definitions for heritable disorders and testing results, and confirmatory testing and verification of positive results, in order to assess and enhance monitoring of newborn diseases.

The Committee did not undertake activities related to the coordination of surveillance activities in CY 2022.

Section 17. Timeliness of Collection, Delivery, Receipt, and Screening

(L) the timeliness of collection, delivery, receipt, and screening of specimens to be tested for heritable disorders in newborns in order to ensure rapid diagnosis and follow-up.

The Committee did not undertake activities related to the timeliness of collection, delivery, receipt, and screening in CY 2022.

FUTURE DIRECTIONS

In CY 23, the Committee expects to:

- Continue the full evidence review of Krabbe disease.
- Continue review of the nomination package for DMD.
- Address potential challenges in its capacity to provide timely and comprehensive reviews of new condition nominations through the Capacity and Prioritization Workgroup discussion and development of prioritization criteria.

- Continue its discussion on the challenges in state implementation of newborn screening and pursue recommendations to improve and reduce barriers to newborn screening systems.

CONCLUSIONS

This report was prepared to summarize the Committee's activities and outcomes for CY 2022 in order to fulfill the legislative requirement for the submission of an annual report to Congress, the HHS Secretary, the Interagency Coordinating Committee on Newborn and Child Screening, and state health departments. The mission of the Committee is to reduce morbidity and mortality in newborns and children who have, or who are at risk for, genetic disorders. The Committee accomplishes this mission by providing advice, recommendations, and technical information to the HHS Secretary to support the development of policies and priorities that enhance services at state and local levels. The Committee also invites public comments as an important approach to understanding lived and professional experiences, issues, and concerns related to newborn screening.

In CY 2022, the Committee recommended the inclusion of MPS II and GAMT deficiency to the RUSP, began its evidence review of Krabbe disease, requested more information on review of the nomination package for cCMV, and initiated review of the nomination package for Duchenne muscular dystrophy. The Committee also heard several presentations from the DoD, and nine state newborn screening and long-term follow-up programs to better understand the facilitators and barriers related to the implementation of newborn screening, treatment, and follow-up. In response, the Committee developed an initial set of recommendations aimed at strengthening the newborn screening system. Throughout its deliberations, the Committee focused on ongoing systemic challenges that create inequitable access to care and result in outcome disparities.

The coordinated efforts of the Committee and its stakeholders—including policymakers, state public health agencies, providers, advocates, and the public—will continue to ensure that newborns and children have universal access to high-quality screening, diagnosis, follow-up, disease management and treatment, evaluation, and education. These efforts will support state newborn screening programs and continue to reduce or prevent the potentially devastating consequences of disabilities, life-threatening disease, or death.

APPENDICES

Appendix A. Membership of the Advisory Committee on Heritable Disorders in Newborns and Children

Committee Members

Kyle Brothers, MD, PhD
Endowed Chair of Pediatric Clinical and Translational Research
Associate Professor of Pediatrics
University of Louisville School of Medicine

Ned Calonge, MD, MPH (Chairperson)
Associate Professor of Family Medicine
University of Colorado School of Medicine
Term End Date: June 30, 2026

Jannine D. Cody, PhD
Professor, Department of Pediatrics
Director, Chromosome 18 Clinical Research Center
University of Texas Health Science Center
Founder and President
The Chromosome 18 Registry & Research Society

Jane M. DeLuca PhD RN
Associate Professor
Clemson University School of Nursing
Metabolic Nurse Practitioner
The Greenwood Genetic Center

Ashutosh Lal, MD
Professor of Clinical Pediatrics
University of California San Francisco School of Medicine
Benioff Children's Hospital

Jennifer M. Kwon, MD, MPH, FAAN
Director, Pediatric Neuromuscular Program
American Family Children's Hospital
Professor of Child Neurology
University of Wisconsin School of Medicine & Public Health

Shawn E. McCandless, MD
Professor, Department of Pediatrics
Head, Section of Genetics and Metabolism
University of Colorado
Anschutz Medical Campus
Children's Hospital Colorado

Chanika Phornphutkul, MD, FACMG
Professor of Pediatrics and Pathology and Laboratory Medicine and Genetics
Director, Division of Human Genetics
Department of Pediatrics, Brown University
Hasbro Children's Hospital
Rhode Island Hospital

Ex-Officio Members

Agency for Healthcare Research & Quality
Kamila B. Mistry, PhD, MPH
Senior Advisor
Child Health and Quality Improvement

Centers for Disease Control & Prevention
Carla Cuthbert, PhD
Chief
Newborn Screening and Molecular Biology Branch
Division of Laboratory Sciences
National Center for Environmental Health

Food & Drug Administration
Kellie B. Kelm, PhD
Director
Division of Chemistry and Toxicology Devices
Office of In Vitro Diagnostics and Radiological Health

Health Resources & Services Administration
Michael Warren, MD, MPH, FAAP
Associate Administrator
Maternal and Child Health Bureau

National Institutes of Health
Diana W. Bianchi, MD
Director
Eunice Kennedy Shriver National Institute of Child Health and Human Development

Acting Designated Federal Official

Soohyun Kim, MPH, CPH
Health Resources and Services Administration
Genetic Services Branch
Maternal and Child Health Bureau

Appendix B. ACHDNC Charter can be accessed online.

Appendix C. Recommended Uniform Screening Panel (RUSP) can be accessed online.

Appendix D. Recommendations to HHS Secretary can be accessed online.

Appendix E. Previously Nominated Conditions can be accessed online.