2025 Current Fiscal Year Report: Advisory Committee on Heritable Disorders in Newborns and Children

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1. Department or Agency	2. Fiscal Year
Department of Health and Human Services	2025

3. Committee or Subcommittee	3b. GSA Committee No.
Advisory Committee on Heritable Disorders in Newborns and Children	13817

4. Is this New During Fiscal Year?	5. Current Charter	6. Expected Renewal Date	7. Expected Term Date
No	11/10/2024	11/10/2026	

8a. Was Terminated During FiscalYear?	8b. Specific Termination Authority	8c. Actual Term Date
Yes	2025 Secretary Directive	04/01/2025

9. Agency Recommendation for Next FiscalYear	10a. Legislation Req to Terminate?	10b. Legislation Pending?
Terminate	No	Enacted

11. Establishment Authority Authorized by Law

12. Specific Establishment Authority	13. Effective Date	14. Commitee Type	14c. Presidential?
Public Health Service Act (PHS), Title XI Section 1111, 42 U.S.C 300b-10	12/18/2014	Continuing	No

15. Description of Committee Scientific Technical Program Advisory Board

16a. Total Number of Reports	No Reports for this FiscalYear

17a. Open	17b. Closed	17c. Partially Closed	Other Activities	17d. Total
1	0	0	0	1

Meetings and Dates

Purpose	Start	End
ACHDNC provides advice and recommendations to the Secretary of Health and Human Services concerning genetic disorders and newborn and childhood screening practices for these disorders.	11/14/2024	11/14/2024

Number of Committee Meetings Listed: 1

	Current FY	Next FY
18a(1). Personnel Pmts to Non-Federal Members	$0.00	$0.00
18a(2). Personnel Pmts to Federal Members	$1,250.00	$0.00
18a(3). Personnel Pmts to Federal Staff	$104,000.00	$0.00
18a(4). Personnel Pmts to Non-Member Consultants	$0.00	$0.00
18b(1). Travel and Per Diem to Non-Federal Members	$0.00	$0.00
18b(2). Travel and Per Diem to Federal Members	$0.00	$0.00
18b(3). Travel and Per Diem to Federal Staff	$0.00	$0.00
18b(4). Travel and Per Diem to Non-member Consultants	$0.00	$0.00
18c. Administrative Costs (FRNs, contractor support, In-person/hybrid/virtual meetings)	$0.00	$0.00
18d. Other (all other funds not captured by any other cost category)	$0.00	$0.00
18e. Total Costs	$105,250.00	$0.00
19. Federal Staff Support Years (FTE)	1.60	0.00

20a. How does the Committee accomplish its purpose?

The Advisory Committee provides advice and

recommendations concerning the grants and projects authorized under the Heritable Disorders Program and technical information to develop policies and priorities for this program that will enhance the ability of the state and local health agencies to provide for newborn and child screening, counseling and health care services for newborns and children having or at risk for heritable disorders. Specifically, the Committee provides advice to the Secretary of HHS regarding the most appropriate timelines of universal newborn screening that each newborn screening program should work towards to ensure that infants are not missed and which conditions should be on the Recommended Uniform Screening Panel. The Committee bases their advice on data and information collected by the Committee through stakeholder interviews, data collection, and evidence reviews.

20b. How does the Committee balance its membership?

The Committee structure shall not exceed 15 members, including the Chair and the ex-officio members. The total membership of the Committee shall be an odd number. The Secretary or his designee shall appoint, as ex-officio members to the Committee, the Administrator of the Health Resources and Services Administration; the Directors of the Centers for Disease Control and Prevention, the National Institutes of Health, and the Agency for Healthcare Research and Quality; or their designees; and representatives of such federal agencies as the Secretary determines are necessary for the Committee to effectively carry out its functions. Other members and the Chair shall be selected by the Secretary or their designee from medical, technical, public health, or scientific professionals with special expertise in

the field of heritable disorders or in providing screening, counseling, testing or specialty services for newborns and children at risk for heritable disorders and from members of the public having special expertise about or concern with heritable disorders. To the extent practicable, Committee members should represent minority, gender, and geographical diversity of newborns served by the state newborn screening programs. The Department will give close attention to equitable geographical distribution and to minority and female distribution so long as the effectiveness of the Committee is not impaired.

20c. How frequent and relevant are the Committee Meetings?

There are approximately four meetings in a calendar year and each meeting is relevant to newborn and child screening for genetic conditions.

20d. Why can't the advice or information this committee provides be obtained elsewhere?

The Committee makes recommendations to the Secretary of HHS on grants and projects to help states and local public health agencies improve screening, counseling, and health care services to newborns and children who have or are at risk for heritable disorders. Committee members also advise the Secretary on policies and priorities to help agencies provide these services. Of importance, the Committee provides recommendations to the Secretary regarding which conditions should be on the Recommended Uniform Screening Panel (RUSP). The RUSP is a list of disorders that are screened at birth and recommended by the Secretary of HHS for states to screen as part of their state universal newborn screening (NBS) programs. Disorders on the

RUSP are chosen based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments. It is recommended that every newborn be screened for all disorders on the RUSP. Most states screen for the majority of disorders on the RUSP; newer conditions are still in process of adoption. Some states also screen for additional disorders. Although states ultimately determine what disorders their NBS program will screen for, the RUSP establishes a standardized list of disorders that have been supported by the Committee and the Secretary of HHS.

20e. Why is it necessary to close and/or partially closed committee meetings?

N/A. To date, all Committee meetings are public.

21. Remarks

2025 Secretary Directive-Elimination of Federal Advisory Committees Within the Department of Health and Human Services, Terminated effect April 1, 2025.

Designated Federal Officer

Leticia Manning Public Health Analyst

Committee Members	Start	End	Occupation	Member Designation
Bianchi, Diana	11/10/2020	11/10/2024	National Institutes of Health	Ex Officio Member
Brosco, Jeffrey	08/01/2024	04/01/2025	Health Resources & Services Administration	Ex Officio Member
Caggana, Michele	04/20/2022	04/01/2025	Wadsworth Center	Special Government Employee (SGE) Member
Calonge, Bruce	04/20/2022	04/01/2025	The Colorado Trust	Special Government Employee (SGE) Member

Caposino, Paula	08/01/2023	04/01/2025	Food and Drug Administration	Ex Officio Member
Cody, Jannine	07/03/2022	04/01/2025	Chromosome 18 Registry and Research Society	Special Government Employee (SGE) Member
Cuthbert, Carla	11/10/2020	11/10/2024	Centers for Disease Control and Prevention	Ex Officio Member
Dorley, M.	03/26/2023	04/01/2025	Newborn Screening Assistant Director at the Tennessee Department of Health	Special Government Employee (SGE) Member
Lal, Ashutosh	07/03/2022	04/01/2025	University of California San Francisco Benioff Children's Hospital	Special Government Employee (SGE) Member
Mistry, Kamila	11/10/2020	11/10/2024	Agency for Health Care Research and Quality	Ex Officio Member
Sagatov, Robyn	08/01/2024	04/01/2025	Agency for Health Care Research and Quality	Ex Officio Member
Warren, Michael	11/10/2020	11/10/2024	Associate Administrator MCHB, HRSA	Ex Officio Member

Number of Committee Members Listed: 12

Narrative Description

Purpose of the Committee: To provide the Secretary with advice and recommendations that will enhance the ability of the state and local health agencies to provide for screening, counseling, and health care services for newborns and children having or at risk for heritable disorders, and to advise and guide the Secretary regarding the most appropriate application of universal newborn screening tests, technologies, policies, guidelines and programs. The Committee supports HRSA's mission and strategic plan by the following: Goal III: Build healthy communities. Sub-goal a – Lead and collaborate with others to help communities strengthen resources that improve health for the population. HRSA’s Principle - Partner with stakeholders at all levels - from individuals, families, and communities to organizations, states and tribal organizations. The Advisory Committee members represent consumers, families, grassroots organizations, advocacy organizations, medical

providers, researchers, and state public health entities and provides opportunities for the various stakeholders to work together on policies that can reduce mortality/morbidity of genetic disorders. Goal IV: Improve health equity Sub-goal b - Monitor, identify, and advance evidence-based and promising practices to achieve health equity. HRSA's Principle - Focus on results across the population, by using the best available evidence, monitoring impact and adapting programs to improve outcomes. One of the charges for the Advisory Committee is to make systematic evidence-based recommendations that have the potential to significantly impact public health as well as health outcomes for all newborns and children screened in the United States.

What are the most significant program outcomes associated with this committee?

	Checked if Applies
Improvements to health or safety	✓
Trust in government	✓
Major policy changes	✓
Advance in scientific research	✓
Effective grant making	✓
Improved service delivery	✓
Increased customer satisfaction	✓
Implementation of laws or regulatory requirements	✓
Other	✓

Outcome Comments

The Advisory Committee shall review and report regularly on newborn and childhood screening practices, recommend improvements in the national newborn and childhood screening programs, and shall engage in the following activities: 1) Provide advice and recommendations to the Secretary of HHS concerning grants and projects awarded or funded under Section 1109 of the PHS Act; 2) Provide technical information to the Secretary for the development of policies and priorities for the administration of grants under Section 1109 of the PHS Act; 3) Provide recommendations, advice, or information on certain diagnostic and screening activities; 4) 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 in newborns and children from heritable

disorders; 5) 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; 6) Develop a model decision-matrix for newborn screening expansion, including an evaluation of the potential public health impact of such expansion and periodically update the recommended uniform screening panel, as appropriate, based on such decision-matrix; and 7) Consider ways to ensure that all states attain the capacity to screen for the conditions designated in the uniform screening panel and include in such consideration the results of grant funding under section 1109. To address this task, the Committee may make recommendations, advice, or information dealing with: follow-up activities, including those necessary to achieve rapid diagnosis in the short-term, and those that ascertain long-term case management outcomes and appropriate access to related services; implementation, monitoring, and evaluation of newborn screening activities, including diagnosis, screening, follow-up, and treatment activities; diagnostic and other technology used in screening; the availability and reporting of testing for conditions for which there is no existing treatment; 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; 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; 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; public and provider awareness and education; the cost and effectiveness of newborn screening and medical evaluation systems and intervention programs conducted by state-based programs; identification of the causes of, public health impacts of, and risk factors for heritable disorders; and 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. In addition, the Committee provides consultation to the Secretary of HHS, acting through the Director of the Centers for Disease Control and Prevention on laboratory quality to provide for: 1) quality assurance for laboratories involved in screening newborns and children for heritable disorders, including quality assurance for newborn-screening tests, performance evaluation services, and technical assistance and technology transfer to newborn screening laboratories to ensure analytic validity and utility of screening tests; and 2) appropriate quality control and other performance test materials to evaluate the performance of new screening tools. The

Advisory Committee will support the purpose and activities of the Interagency Coordinating Committee to 1) assess existing activities and infrastructure, including activities on birth defects and developmental disabilities authorized under Section 317C, in order to make recommendations to programs to collect, analyze, and make available data on the heritable disorders recommended by the Advisory Committee under Section 1111, including data on the incidence and prevalence of, as well as poor health outcomes resulting from, such disorders; and 2) make recommendations for the establishment of regional centers for the conduct of applied epidemiological research on effective interventions to promote the prevention of poor health outcomes resulting from such disorders as well as providing information and education to the public on such effective interventions.

What are the cost savings associated with this committee?

	Checked if Applies
None	☐
Unable to Determine	☑
Under $100,000	☐
$100,000 - $500,000	☐
$500,001 - $1,000,000	☐
$1,000,001 - $5,000,000	☐
$5,000,001 - $10,000,000	☐
Over $10,000,000	☐
Cost Savings Other	☐

Cost Savings Comments

Committee's cost savings have not been determined.

What is the approximate Number of recommendations produced by this committee for the life of the committee?

13

Number of Recommendations Comments

In FY 2025 the Committee did not submit any recommendations to the Secretary for approval.

What is the approximate Percentage of these recommendations that have been or will be Fully implemented by the agency?

80%

% of Recommendations Fully Implemented Comments

In November 2015, the Secretary of HHS adopted the Committee's recommendations and added Mucopolysaccharidosis 1 (MPS 1) and X-Linked Adrenoleukodystrophy (ALD) to the Recommended Uniform Screening Panel (RUSP). The Secretary also asked federal agencies to consider ways within their existing resources to support state programs as they begin to implement population-based screening for MPS 1 and Adrenoleukodystrophy (X-ALD) . As a result, in FY 2016 HRSA issued a $2 million funding opportunity announcement with the purpose being to implement new conditions added to the RUSP including MPS I and X-ALD. In FY 2017 the Newborn Screening Implementation Program Regarding Conditions Added to the RUSP was awarded to the Association of Public Health Laboratories (APHL), worked to increase the number of newborns with Pompe disease, MPS I, and X-ALD identified through newborn screening that receive early treatment. States continue to work on adding conditions recently added to the RUSP. In FY 2018 the Committee provided the following recommendation to the Secretary: Expand the Recommended Uniform Screening Panel (RUSP) to include the addition of spinal muscular atrophy (SMA) due to homozygous deletion of exon 7 in SMN1. In July 2018 the Secretary accepted the recommendation and SMA due to homozygous deletion of exon 7 in SMN1 was added to the RUSP. In addition, the Secretary requested a report, "…describing the status of implementing newborn screening for SMA and clinical outcomes of early treatment, including any potential harms, for infants diagnosed with SMA." In December 2020, the Committee submitted the requested report, titled "Review of Newborn Screening Implementation for Spinal Muscular Atrophy" to the Secretary. The report noted that there has been relatively quick adoption of newborn screening for SMA by states and that the current available evidence supports the benefit of early detection. From FY 2018-2021 HRSA investments such as the Quality Improvement in Newborn Screening program continued to support state-level implementation of newly added RUSP conditions and 37 states are now screening for SMA. In March 2022, the Committee provided the following recommendation to the Secretary: Expand the RUSP to include the addition of Mucopolysaccharidosis Type II (MPS II). In August 2022, the Secretary accepted the recommendation to add MPS II to the RUSP and the condition has been added. In addition, the Secretary requested a report, "...describing the status of state implementation of MPS II screening, access, and cost of treatment for infants diagnosed with MPS II and the impact on families due to the treatment periodicity." In June 2022, the Committee provided the following recommendation to the Secretary: Expand the RUSP to include the addition of Guanidinoacetate Methyltransferase (GAMT) Deficiency. In FY 2022, this recommendation was submitted to the Secretary for review. In January 2023, the Secretary accepted the recommendation to add GAMT to the RUSP. In addition, the Secretary requested a report in 5 years, "…describing the status of implementation of

GAMT deficiency screening, potential barriers to treatment and to long-term follow up, and health outcomes.

What is the approximate Percentage of these recommendations that have been or will be Partially implemented by the agency?
20%

% of Recommendations Partially Implemented Comments
Not Applicable

Does the agency provide the committee with feedback regarding actions taken to implement recommendations or advice offered?
Yes ☑ No ☐ Not Applicable ☐

Agency Feedback Comments
Through correspondence from the Secretary. The public can obtain information of agency feedback via the committee website at https://www.hrsa.gov/advisory-committees/heritable-disorders/index.html

What other actions has the agency taken as a result of the committee's advice or recommendation?

	Checked if Applies
Reorganized Priorities	☐
Reallocated resources	☐
Issued new regulation	☐
Proposed legislation	☐
Approved grants or other payments	☐
Other	☐

Action Comments
N/A, no comments

Is the Committee engaged in the review of applications for grants?
No

Grant Review Comments
The committee does not review applications for grants.

How is access provided to the information for the Committee's documentation?

	Checked if Applies
Contact DFO	☑
Online Agency Web Site	☑
Online Committee Web Site	☑
Online GSA FACA Web Site	☑
Publications	☑
Other	☐

Access Comments

https://www.hrsa.gov/advisory-committees/heritable-disorders/index.html