2018 Current Fiscal Year Report: Advisory Committee on Heritable Disorders in Newborns and Children

Report Run Date: 06/26/2019 02:11:06 PM

1. Department or Agency	2. Fiscal Year
Department of Health and Human Services	2018

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	05/07/2015	09/30/2019	

8a. Was Terminated During FiscalYear?	8b. Specific Termination Authority	8c. Actual Term Date
No		

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

11. Establishment Authority Statutory (Congress Created)

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 3

16b. Report Date	Report Title
02/08/2018	The Role of Quality Measures to Promote Long-Term Follow-up of Children Identified by Newborn Screening Programs
03/31/2018	Evidence-based Review of NBS for SMA
04/01/2018	Annual Report to Congress

Number of Committee Reports Listed: 3

17a. Open 4 **17b. Closed** 0 **17c. Partially Closed** 0 **Other Activities** 0 **17d. Total** 4

Meetings and Dates

Purpose	Start	End

The Committee heard updates on states' progress toward the Committee's newborn screening timeliness goals and the evidence review of spinal muscular atrophy (SMA).The Committee heard presentations on, and discussed: the implications of detecting carriers through newborn screening; the clinical and public health impact of Severe Combined Immunodeficiency (SCID), and long term follow up in newborn screening. The Committee also heard updates from, and provided direction to, the Laboratory Standards and Procedures workgroup, Follow-up and Treatment workgroup, and Education and Training workgroup.	11/08/2017 - 11/09/2017
The Committee considered the nomination of spinal muscular atrophy (SMA) to the Recommended Uniform Screening Panel (RUSP) and voted to recommend to the HHS Secretary to expand the RUSP to include SMA due to homozygous deletion of exon 7 in SMN1. The Committee also discussed a cutoffs and risk assessment methods used for dried bloodspot newborn screening. The Committee finalized a report on Quality Measures in Newborn Screening from the Follow up and Treatment workgroup.	02/08/2018 - 02/08/2018
The Committee discussed and finalized two education and training tools (a communication guide for health care providers and an educational planning guide). The Committee heard presentations from experts in the field and discussed the following topics: quality assurance, risk assessment in newborn screening, timeliness in newborn screening, assessing the public health impact of adding conditions to the RUSP, and the status of newborn screening pilot studies for Guanidinoacetate Methyltransferase (GAMT) deficiency. The Committee also heard updates from, and provided direction to, the Laboratory Standards and Procedures workgroup, Follow-up and Treatment workgroup, and Education and Training workgroup.	05/09/2018 - 05/10/2018
The Committee heard from experts in the field and discussed the following topics: risk assessment in newborn screening, improving timeliness, long-term follow up in newborn screening, and technology in newborn screening. The Committee also heard updates from, and provided direction to, the Laboratory Standards and Procedures workgroup, Follow-up and Treatment workgroup, and Education and Training workgroup.	08/02/2018 - 08/02/2018

Number of Committee Meetings Listed: 4

	Current FY	Next FY
18a(1). Personnel Pmts to Non-Federal Members	$13,500.00	$14,750.00
18a(2). Personnel Pmts to Federal Members	$0.00	$0.00
18a(3). Personnel Pmts to Federal Staff	$283,679.00	$293,788.00
18a(4). Personnel Pmts to Non-Member Consultants	$0.00	$0.00
18b(1). Travel and Per Diem to Non-Federal Members	$32,175.00	$40,000.00
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. Other(rents,user charges, graphics, printing, mail, etc.)	$144,368.00	$170,246.00
18d. Total	$473,722.00	$518,784.00
19. Federal Staff Support Years (FTE)	1.80	1.80

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 has provided advise to the Secretary regarding the most appropriate timelines of universal newborn screening that each newborn screening

program should work towards to ensure that infants are not missed. The Committee based their advise on data and information collected by the Committee through stakeholder interviews and data collection.

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 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 his 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 4 meetings a year and each meeting is relevant to newborn and child screening for genetic conditions. The Committee met four times in FY2018.

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

The Committee makes recommendations to the Secretary 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 will 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 the Department of Health and Human Services (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?
To date, all Committee meetings are public.

21. Remarks
The Advisory Committee on Heritable Disorders in Newborns and Children (Committee) legislative authority is the Public Health Service Act (PHS), Title XI, § 1111 (42 U.S.C. 300b-10). The Committee also is governed by the provisions of the Federal Advisory Committee Act (FACA), as amended (5 U.S.C. App.), which sets forth standards for the formation and use of advisory committees. Objective and Scope of Activities: The Committee provides advice, recommendations, and technical information about aspects of heritable disorders and newborn and childhood screening to the Secretary of Health and Human Services (HHS) for the development of policies and priorities that will enhance the ability of the State and local health agencies to provide for such screening, counseling and health care services for newborns and children having, or at risk for, heritable disorders. The Committee met four times in FY2018. The Committee posted an evidence review report for spinal muscular atrophy (SMA) in February 2018, a Report to Congress in May 2018, and a report on "The Role of Quality Measures to Promote Long-Term Follow-up of Children Identified by Newborn Screening Programs" in September 2018.

Designated Federal Officer
Catharine Riley Public Health Analyst, Genetic Services, Maternal and Child Health Bureau, Health Resources and Services Administration, 5600 Fishers Ln, Rm. 18A-19, Rockville, MD 20857

Committee Members	Start	End	Occupation	Member Designation
Baker, Mei	07/01/2016	06/30/2020	Co-Director, Newborn Screening Laboratory, Wisconsin State Laboratory	Special Government Employee (SGE) Member
Berry, Susan	10/29/2017	06/30/2021	Professor and Director, University of Minnesota	Special Government Employee (SGE) Member
Bianchi, Diana	01/01/2017	09/30/2019	National Institutes of Health	Ex Officio Member
Bocchini, Joseph	05/07/2015	04/24/2019	Louisiana State University Health Sciences Center in Shreveport	Special Government Employee (SGE) Member
Brosco, Jeffrey	07/01/2016	06/30/2020	Professor of Clinical Pediatrics	Special Government Employee (SGE) Member
Cuthbert, Carla	07/01/2017	09/30/2019	Centers for Disease Control & Prevention	Ex Officio Member
Kavanagh, Laura	01/01/2018	02/28/2019	Health Resources & Services Administration	Ex Officio Member

Kelm, Kellie	05/07/2015	09/30/2019	Food and Drug Administration	Ex Officio Member
Lorey, Fred	05/07/2015	01/12/2018	California Department of Public Health	Special Government Employee (SGE) Member
Matern, Dietrich	05/07/2015	06/30/2018	Mayo Clinic	Special Government Employee (SGE) Member
Mistry, Kamila	05/07/2015	09/30/2019	Agency for Healthcare Research and Quality	Ex Officio Member
Powell, Cynthia	10/29/2017	06/30/2021	The University of North Carolina at Chapel Hill	Special Government Employee (SGE) Member
Saarinen, Annamarie	07/01/2016	06/30/2020	Co-founder Newborn Coalition	Special Government Employee (SGE) Member
Shone, Scott	10/29/2017	06/30/2021	RTI International	Special Government Employee (SGE) Member
Tarini, Beth	07/01/2016	06/30/2020	Associate Professor and Division Director	Special Government Employee (SGE) Member
Wicklund, Catherine	05/07/2015	06/30/2018	Northwestern University	Special Government Employee (SGE) Member

Number of Committee Members Listed: 16

Narrative Description

Purpose of the Committee: To provide the Secretary 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 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;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 diseasesIn addition, the Committee will provide consultation to the Secretary, 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; 1) to 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?
11

Number of Recommendations Comments
In FY18 there was one recommendation sent to the Secretary for approval. The total number of recommendations to date increased by one since FY17.

What is the approximate Percentage of these recommendations that have been or will be Fully implemented by the agency?
73%

% of Recommendations Fully Implemented Comments
In November 2015, the Secretary of Health and Human Services 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 stated in her response letter that she 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 X-ALD. As a result, in FY16 HRSA issued a $2 million funding opportunity announcement with the purpose being to implement new conditions added to the RUSP including. In FY17 the Newborn Screening Implementation Program Regarding Conditions Added to the Recommended Uniform Screening Panel (RUSP), awarded to the Association of Public Health Laboratories, worked to increase the number of newborns with Pompe disease, Mucopolysaccharidosis I (MPS I), and X-linked Adrenoleukodystrophy (X-ALD) identified through newborn screening that receive early treatment. States continue to work on adding conditions recently added to the RUSP. In FY18 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.

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

18%

% 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.

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

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