Secretary's Advisory Committee on Heritable Disorders in Newborns and Children

2012 Annual Report

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Mission of the SACHDNC

The U.S. Department of Health and Human Services (HHS) Secretary's Advisory Committee on Heritable Disorders in Newborns and Children[I] (SACHDNC) was chartered in February 2003. The SACHDNC provides the Secretary with recommendations, advice, and technical information regarding the most appropriate application of technologies, policies, guidelines, and standards for: (a) effectively reducing morbidity and mortality in newborns and children having, or at risk for, heritable disorders; and (b) enhancing 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.

This report fulfills the legislative requirement to submit an annual report to Congress, the Secretary, the Interagency Coordinating Committee on Newborn and Child Screening, and State Health Departments.

[I] Authority: Public Health Service Act, Title XI, §1111 as amended (42 U.S.C. 300b-10)

Executive Summary

The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) advises the Secretary of the U.S. Department of Health and Human Services (HHS). The Committee's guidance and information serves to enhance the states' ability to apply the most current technologies, policies, guidelines, and standards for effectively reducing morbidity and mortality in newborns and children having, or at risk of, heritable disorders. Heritable disorders can be present at birth and comprise a group of conditions that, undetected, can cause intellectual disabilities, physical disabilities, life-threatening diseases, and/or death. Newborn screening for heritable disorders is a longstanding public health program that provides early identification and follow-up for the treatment of infants affected by certain genetic, metabolic, hormonal, infectious, and/or functional conditions. The intent of newborn screening is to improve and save the lives of infants who may otherwise suffer or die.

During 2011, the SACHDNC:

- facilitated the expansion of screening activities with the addition of Critical Congenital Heart Disease to the Recommended Uniform Screening Panel (RUSP);
- examined and developed a strategy for revising the review process for conditions under consideration for the RUSP;
- considered the implications of point-of-care screening for public health;
- submitted a report to the Secretary for Severe Combined Immunodeficiency, containing pilot studies that reported over 60 infants received life-saving treatment;
- submitted a report to the Secretary on risk factors for sickle cell disease carriers, resulting in the Secretarial approval of three SACHDNC recommendations pertaining to screening access, evaluation/testing, and the education of safe practices in sports;
- compiled a report on the critical elements of long-term follow-up of individuals with heritable disorders, that will facilitate standardization/quality improvement of screening, data collection, and follow-up;
- supported endorsement of quality measures submitted by the Health Resources and Services Administration (HRSA), Centers for Disease Control and Prevention (CDC), and the National Committee for Quality Assurance (NCQA) to the National Quality Forum (NQF), advancing the view of quality assurance in newborn screening as a medical standard; and
- requested information and facts to increase the SACHDNC's capacity to provide technical information, advice, and recommendations to the Secretary regarding diagnostic and other technology used in screening.

Report

This report discusses activities undertaken by the SACHDNC from January to December 2011, including peer-reviewed newborn screening (NBS) guidelines, follow-up, and treatment in the United States. The discussion portion of this report is subdivided into sections that are aligned with the Committee's legislative duties.

1. Advice and Recommendations: Grants and Projects

> 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;

Following the SACHDNC's recommendation to add Critical Congenital Heart Disease (CCHD) to the RUSP in 2010, the Committee issued a 2011 report on *Strategies for Implementing Screening for CCHD*. The report concluded that pulse oximetry, a measurement for blood oxygen saturation, is a low-cost, non-invasive, point-of-care medical intervention. The report noted that public health agencies will have an important role in quality assurance and surveillance (Kemper et al., 2011).

In September 2011, the Secretary adopted the SACHDNC's recommendation to add CCHD to the RUSP, and requested that the Committee collaborate with the National Institutes of Health (NIH), the CDC, and HRSA in the development of research, surveillance, screening standards/infrastructure, and education/training materials for the NBS of CCHD[II] (Sebelius, September 2011).

2. Technical Information

> The Advisory Committee shall—
>
> (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;

To enable the SACHDNC to provide technical information to the Secretary for the development of policies and priorities, the Committee requested information and facts regarding: (a) the role of the Food and Drug Administration (FDA) in the treatment/screening of rare disease disorders, (b) procedures by the FDA for the management of drug shortages, and (c) findings of the Institute of Medicine (IOM) report on Rare Diseases and Orphan Products. This addressed the Committee's ongoing concerns about:

- rare diseases, new therapies/testing, and changes in FDA regulations;
- medication shortages within the rare disease community and its impact on individuals suffering from heritable disorders; and

[II] Refer to the topic *Critical Congenital Heart Disease* in Appendix B.

- the impact of the IOM report's findings (e.g., insufficient resources, inconsistent review of applications) on individuals with heritable disorders.

3. Systematic Evidence-Based and Peer-Reviewed Recommendations

> The Advisory Committee shall—
>
> (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;

The SACHDNC makes recommendations regarding conditions to be included on the RUSP based on independent scientific evidence that supports the potential net benefit of screening. Nominated conditions are assigned to the External Evidence Review Workgroup (EERW) for an independent, evidence-based review. The evidence review process determines the suitability and potential net benefit of screening for nominated inherited disorders, based upon the systematic evaluation of results of controlled trials, observational studies, case studies, expert opinion, focus groups, cost-effectiveness analysis, policy analyses, and ethical analysis.

Throughout 2011, the EERW continued their review for the nominated screening condition of Neonatal Hyperbilirubinemia. As of December 2011, the review was still in progress.

During 2011, the EERW began revising the process for preparing systematic evidence-based and peer-reviewed recommendations. This revised independent evidence review process, referred to as a *condition review*, will include a more robust analysis of the public health impact of conditions under nomination.[III]

As of December 2011, the RUSP listed 31 core conditions for universal NBS.[IV] States determine their respective list of screened conditions, and almost all States have adopted or are in the process of adopting the core conditions listed on the RUSP.

4. Decision Matrix

> The Advisory Committee shall—
>
> (4) 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;

There were no updates to the Decision Matrix during 2011. As noted in Section 3, the evidence review process is currently under revision and, once completed, this revised process will influence the framework of the Decision Matrix.[V]

[III] Refer to the *Future Forecast* section in this report.
[IV] For more information about the RUSP, refer to Appendix A.
[V] For more information about the Decision Matrix, refer to the SACHDNC 2011 Annual Report.

5. State Capacity to Screen

The Advisory Committee shall—

(5) 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;

In May 2011, the SACHDNC submitted a report, *Newborn Screening for Severe Combined Immunodeficiency* (SCID), to the Secretary. The report conveyed SCID pilot study results from seven states and territories throughout the U.S. that (a) over 900,000 infants were screened for SCID, (b) the infants screened comprised 25 percent of live births in the U.S., and (c) over 60 infants identified with immunodeficiencies were receiving life-saving treatments (SACHDNC, 2011).[VI] Based on these pilot studies, many additional state programs continue to actively work towards implementation of NBS for SCID.

6. Recommendations, Advice, or Information

The Advisory Committee shall—

(6) 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—

A. Follow-Up Activities

(A) 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;

The SACHDNC has been engaged with identifying and addressing the challenges of ensuring short-term and long-term follow-up (LTFU) activities, including care coordination, evidence-based treatment, and continuous quality improvement.

To assure quality outcomes and ensure standardization, the SACHDNC developed a report entitled, *What Questions Should Newborn Screening Long-Term Follow-Up Be Able To Answer?* (Hinton et al., 2011). These questions encompassed the key components of care coordination, evidence-based practice, continuous quality improvement, and new knowledge discovery, arranged from the perspectives of three important stakeholders: (a) families; (b) the medical home, primary care provider, specialists, and investigators; and the (c) nation/state. The SACHDNC compiled these crucial questions to assist in future evaluation of NBS systems and promote quality measures for LTFU programs (Hinton et al., 2011).

[VI] Refer to the topic *Severe Combined Immunodeficiency* in Appendix B.

B. Implementation, Monitoring, and Evaluation

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

During 2011, the SACHDNC initiated a study on the impact of implementing point-of-care (POC) screening for newborns. POC screening encompasses testing at or near the site of care, to facilitate the timely receipt of testing results and administration of needed care. CCHD, a heritable condition with results available at the bedside and requiring immediate diagnosis and follow-up, illustrates how POC screening can save lives.

C. Diagnostic and Other Technology

> (C) diagnostic and other technology used in screening;

To enable the SACHDNC to provide diagnostic and technology information to the Secretary, the Committee requested information and facts from the CDC.

In January 2011, the Newborn Screening and Molecular Biology Branch of the CDC provided updates to the SACHDNC regarding screening for SCID, a recent addition to the RUSP. As noted in a May 2011 report,[VII] states who conducted screening for SCID had adopted use of a highly sensitive and specific screening tool, a T-cell Receptor Excision Circle (TREC) assay (SACHDNC, 2011). As with other newborn blood spot screens, the CDC provides the appropriate reference materials for the TREC assay.

In September, 2011 the CDC provided the SACHDNC with an update about a newly formed Molecular Quality Improvement Program that provides support to public health laboratories as molecular testing is introduced into practice. This program was developed in response to the Secretary's decision to add SCID to the RUSP in May 2011.

D. Availability and Reporting of Testing

> (D) the availability and reporting of testing for conditions for which there is no existing treatment;

No updates occurred, during 2011, regarding the availability and reporting of testing for conditions for which there is no existing treatment.

E. Conditions Not Included in 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 SACHDNC is specifically charged with developing a knowledge base about conditions not included in the RUSP that are treatable with FDA-approved products.

[VII] Refer to section 5 for more information about the May 2011 report.

During the Committee's May 2011 meeting, the FDA presented information about programs related to the Orphan Drug Act as well as the Expanded Access Program (EAP) on investigational drugs for patients who have a: (a) serious or immediately life-threatening condition, or (b) condition for which there is no other alternate or satisfactory treatment.

F. 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;

In January 2011, the SACHDNC supported the addition of collecting quality assurance information related to state data on the measurement and recording of total state births receiving appropriate, state-mandated blood spot newborn screening. This measure was one of several submitted by HRSA, CDC, and NCQA to the National Quality Forum (NQF). In addition, the NQF also endorsed several hearing screening measures, also supported by the Committee. These endorsements provide a standard way of measurement that may lead to a national standard for the evaluation of public health NBS programs.

G. 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;

In September 2011, the Newborn Screening and Molecular Biology Branch of the CDC provided the SACHDNC with a presentation on their quality assurance activities to sustain vital NBS laboratory competence and capacity in the states. The CDC reported that quality assurance programs on SCID, cystic fibrosis, and hemoglobinopathy have evolved during 2011. As part of their mandated program, the CDC reported that they are engaged in improving NBS quality and detection of heritable disorders by responding to state program-identified needs and developing high-quality laboratory methods.

H. Public and Provider Awareness

> (H) public and provider awareness and education;

During 2011, the SACHDNC focused on NBS awareness and education issues including NBS efficacy, engagement of parents and the public, implementation of an awareness campaign, and outreach efforts through the Newborn Screening Clearinghouse (NBSC). The NBSC is required by the authorizing legislation and charged with increasing NBS awareness and improving the informed decision-making of new/expectant parents, health professionals, industry representatives, and the public. The NBSC is intended to connect parents and healthcare providers with

information about conditions screened, screening programs, and other available resources through a cooperative agreement with HRSA.

The Committee requested information on NBS efficacy from a family perspective. Specifically, the committee received information from a survey conducted that measured public knowledge/understanding; assessed public support; assessed NBS information the public had, or would like to have, received; examined disease type(s) the public would like to have screened; and determined whether disease severity, age of onset, and positive predictive value influenced public support for NBS.

To increase public awareness of NBS, the SACHDNC continued plans for a Newborn Screening Awareness Campaign, beginning with a media scan that examined what NBS information was currently available to the public. In September 2011, the Committee reviewed the results of a completed media scan, which revealed that (a) basic NBS information, though limited, was available on the Internet; and (b) additional research was needed to determine what NBS information was provided by pediatricians and hospitals to prospective mothers. Next steps for the campaign include convening a strategy session to determine the goals, objectives, audiences, and approach to most effectively increase NBS awareness.

In September 2011, the NBSC reported on their newly launched website (www.babysfirsttest.org). This website facilitates extensive networking and resource sharing through the improvement of ongoing activities, as well as the creation of new avenues for information exchange, providing a central connection to informational resources and NBS quality indicator data.

The Committee continues to: (a) engage parents, consumers, and the public through participation in SACHDNC activities; and (b) educate parents, consumers, the public, and medical providers[VIII] about NBS and heritable disorders.

I. Cost and Effectiveness

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

The SACHDNC acknowledges the need to include cost measurements of NBS, medical evaluation systems, and intervention programs, and will include a more robust consideration of cost-effectiveness as part of the revised independent condition review process.[IX]

J. Causes, Public Health Impacts, and Risk Factors

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

In June 2011, the Secretary agreed to support three SACHDNC recommendations regarding sickle cell disease carriers that support uniform access to screening,

[VIII] Refer to discussion of ACT sheets in section 6.K.
[IX] Refer to the *Future Forecast* section in this report .

completion of evaluation/testing within the medical home, and education of all sports participants on safe practices in sports.[X]

As discussed, identifying the cause, public health impact, and risk factors for heritable disorders will continue to be a focus of the SACHDNC. The revised independent condition review process will include a more robust examination of the potential public health impacts of heritable disorders under consideration for addition to the RUSP.

K. 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.

During 2011, the SACHDNC supported surveillance activities including: (a) development of LTFU data sets to capture disease specific/public health data; (b) creation of Newborn Screening ACTion (ACT) sheets, by the American College of Medical Genetics and Genomics (ACMG), to support primary care providers nationwide in the clinical decisions about newborn screening disorders; (c) improvement of lab quality through comparison/clinical validation of tandem mass spectrometry cutoff values; and (d) completion of emergency preparedness exercises with state NBS and confirmatory laboratories.

[X] Refer to the topic *Sickle Cell Disease Carrier Status* in Appendix B.

Future Forecast

Future plans by the SACHDNC have been grouped into four main categories: laboratory standards, education and training, follow-up and treatment, and the review process.

Laboratory Standards

The SACHDNC would like to establish a methodology for ongoing evaluation of heritable conditions listed on the RUSP, to ensure that future panels continue to reflect the latest advancements in clinical research and laboratory technology. Recently, there has been a shift to the use of molecular-based testing as a primary test, superseding its previous role as a follow-up procedure to a metabolite or analyte test. This will require further investigation. In addition, the Committee will continue to facilitate development of new medical language codes (e.g., LOINC) with the National Library of Medicine as well as monitor new health information technologies as they emerge.

Education and Training

The SACHDNC will facilitate a strategy summit for the Newborn Screening Awareness Campaign, as noted in section 6.H., and will continue to address education and training issues for parents, providers, and the public regarding NBS, follow-up, and treatment. The Committee will also encourage the incorporation of NBS information into genetic training initiatives for providers.

Follow-up and Treatment

The SACHDNC will continue to: (a) examine the needs for implementation and monitoring of NBS follow-up practices, (b) facilitate the interface of public health in short and long-term care, (c) examine the needs of states, and (d) examine needed mechanisms for improving quality assurance programs. The Committee will also examine issues regarding gaps in care and coverage for affected families.

Review Process

Through the improved condition review process, the EERW will be able to systematically analyze and report on the potential public health impact of additional conditions under consideration for addition to the RUSP. In 2012, an expert panel will convene public health officials, providers, and NBS experts to examine factors including cost-effectiveness, health economics, laboratory capacity, provider capacity, and impact on state programs. This expert panel will be responsible for creating the process to evaluate the public health impact within the condition review process.

Conclusion

During 2011, the SACHDNC continued to provide recommendations, advice, and technical information to assist the Secretary in efforts to reduce the morbidity and mortality in newborns and children having, or at risk of, heritable disorders.

The recent addition of CCHD to the RUSP has prompted examination of POC screening, and the revised condition review process will include a more robust examination of the potential public health impacts of heritable disorders under consideration for addition to the RUSP. The Committee has supported NQF-endorsed measures to assure that all infants receive appropriate NBS. The additions to the RUSP illustrate the importance and value of NBS, as evidenced by the additional lives saved through the implementation of screening for SCID and CCHD and awareness and knowledge of sickle cell disease and trait.

Identifying the causes, public health impacts, and risk factors for heritable disorders will continue to be a focus of the SACHDNC. Ten years ago, the majority of states were screening for seven conditions or less, supplemental screening information was not provided to families, a high number of children were at risk for morbidity and mortality related to heritable disorders,[XI] and there were limited opportunities for the public to provide input during policy discussions. Due to the ongoing efforts of the SACHDNC, all states now have expanded NBS to thirty or more conditions, more information is available to families and providers, a greater number of newborns are tested, and more newborns are receiving needed care.

Ultimately, through the information and guidance of the SACHDNC, states will continue to be updated with the most current application of technologies, policies, guidelines, and standards for effectively reducing morbidity and mortality in newborns and children having, or at risk of, heritable disorders. The coordinated efforts of the SACHDNC and stakeholders—including policymakers, state public health agencies, providers, and the public—will continue to ensure that newborns and children have universal access to high-quality screening, follow-up, diagnosis, disease management and treatment, evaluation, and education, which may prevent the potentially devastating consequences of disabilities, life-threatening diseases, or death.

[XI] Diagnosis did not occur until the infant or child was in the emergency room or intensive care.

References

Hinton, C.F., Feuchtbaum, L., Kus, C.A., Kemper, A.R., Berry, S.A., Levy-Fisch, J., Luedtke, J., Kaye, C., & Boyle, C.A. (2011). What questions should newborn screening long-term follow-up be able to answer? A statement from the U. S. Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. *Genetics in Medicine, 13*(10), 861–865.

Kemper, A.R., Mahle, W.T., Martin, G.R., Cooley, W.C., Kumar, P., Morrow, W.R., Kelm, K., Pearson, G.D., Glidewell, J., Grosse, S.D., & Howell, R.R. (2011). Strategies for implementing screening for critical congenital heart disease. *Pediatrics, 128*(5), e1259–1267.

Mayo Foundation for Medical Education and Research (Mayo). (2012). *Infant jaundice: Definition.* Retrieved from http://www.mayoclinic.com/health/infant-jaundice/DS00107

SACHDNC. (2011). *Newborn screening for Severe Combined Immunodeficiency Disorder.* Retrieved from http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendations/correspondence/combinedimmunodeficiency.pdf

Sebelius, K. (2011, September 21). [Letter to Dr. Rodney Howell]. Retrieved from http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendations/correspondence/cyanoticheartsecre09212011.pdf

Severe Combined Immunodeficiency (SCID). (2012). *Missing body defense systems.* Retrieved from http://www.scid.net/about.htm

Appendix A: Recommended Uniform Screening Panel[1]

Core [2] Conditions [3]

(as of December 2011)

ACMG Code	Core Condition	Metabolic Disorder			Endocrine Disorder	Hemoglobin Disorder	Other Disorder
		Organic acid condition	Fatty acid oxidation disorders	Amino acid disorders			
PROP	**Propionic acidemia**	X					
MUT	**Methylmalonic acidemia (methylmalonyl-CoA mutase)**	X					
Cbl A,B	**Methylmalonic acidemia (cobalamin disorders)**	X					
IVA	**Isovaleric acidemia**	X					
3-MCC	**3-Methylcrotonyl-CoA carboxylase deficiency**	X					
HMG	**3-Hydroxy-3-methyglutaric aciduria**	X					
MCD	**Holocarboxylase synthase deficiency**	X					
ßKT	**ß-Ketothiolase deficiency**	X					
GA1	**Glutaric acidemia type I**	X					
CUD	**Carnitine uptake defect/carnitine transport defect**		X				
MCAD	**Medium-chain acyl-CoA dehydrogenase deficiency**		X				
VLCAD	**Very long-chain acyl-CoA dehydrogenase deficiency**		X				
LCHAD	**Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency**		X				
TFP	**Trifunctional protein deficiency**		X				
ASA	**Argininosuccinic aciduria**			X			
CIT	**Citrullinemia, type I**			X			
MSUD	**Maple syrup urine disease**			X			
HCY	**Homocystinuria**			X			
PKU	**Classic phenylketonuria**			X			
TYR I	**Tyrosinemia, type I**			X			
CH	**Primary congenital hypothyroidism**				X		
CAH	**Congenital adrenal hyperplasia**				X		
Hb SS	**S,S disease (Sickle cell anemia)**					X	
Hb S/ßTh	**S, βeta-thalassemia**					X	
Hb S/C	**S,C disease**					X	
BIOT	**Biotinidase deficiency**						X
CCHD	**Critical congenital heart disease**						X
CF	**Cystic fibrosis**						X
GALT	**Classic galactosemia**						X
HEAR	**Hearing loss**						X
SCID	**Severe combined immunodeficiences**						X

1. Selection of conditions based upon “Newborn Screening: Towards a Uniform Screening Panel and System.” *Genetic Med*. 2006; 8(5) Suppl: S12-S252” as authored by the American College of Medical Genetics (ACMG) and commissioned by the Health Resources and Services Administration (HRSA).
2. Disorders that should be included in every Newborn Screening Program.
3. Nomenclature for Conditions based upon “Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels.” *Pediatrics*. 2006; 117 (5) Suppl: S308-S314.

SACHDNC Recommended Uniform Screening Panel[1]
SECONDARY[2] CONDITIONS[3]
(as of December 2011)

ACMG Code	Secondary Condition	Metabolic Disorder			Hemoglobin Disorder	Other Disorder
		Organic acid condition	Fatty acid oxidation disorders	Amino acid disorders		
Cbl C,D	Methylmalonic acidemia with homocystinuria	X				
MAL	Malonic acidemia	X				
IBG	Isobutyrylglycinuria	X				
2MBG	2-Methylbutyrylglycinuria	X				
3MGA	3-Methylglutaconic aciduria	X				
2M3HBA	2-Methyl-3-hydroxybutyric aciduria	X				
SCAD	Short-chain acyl-CoA dehydrogenase deficiency		X			
M/SCHAD	Medium/short-chain L-3-hydroxyacl-CoA dehydrogenase deficiency		X			
GA2	Glutaric acidemia type II		X			
MCAT	Medium-chain ketoacyl-CoA thiolase deficiency		X			
DE RED	2,4 Dienoyl-CoA reductase deficiency		X			
CPT IA	Carnitine palmitoyltransferase type I deficiency		X			
CPT II	Carnitine palmitoyltransferase type II deficiency		X			
CACT	Carnitine acylcarnitine translocase deficiency		X			
ARG	Argininemia			X		
CIT II	Citrullinemia, type II			X		
MET	Hypermethioninemia			X		
H-PHE	Benign hyperphenylalaninemia			X		
BIOPT (BS)	Biopterin defect in cofactor biosynthesis			X		
BIOPT (REG)	Biopterin defect in cofactor regeneration			X		
TYR II	Tyrosinemia, type II			X		
TYR III	Tyrosinemia, type III			X		
Var Hb	Various other hemoglobinopathies				X	
GALE	Galactoepimerase deficiency					X
GALK	Galactokinase deficiency					X
	T-cell related lymphocyte deficiencies					X

1. Selection of conditions based upon “Newborn Screening: Towards a Uniform Screening Panel and System.” *Genetic Med*. 2006; 8(5) Suppl: S12-S252” as authored by the American College of Medical Genetics (ACMG) and commissioned by the Health Resources and Services Administration (HRSA).
2. Disorders that can be detected in the differential diagnosis of a core disorder.
3. Nomenclature for Conditions based upon “Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels.” *Pediatrics*. 2006; 117 (5) Suppl: S308-S314.

Appendix B: SACHDNC Recommendations and Secretary Response, 2011

TOPIC	SACHDNC RECOMMENDATION	SECRETARY'S RESPONSE/OUTCOME
Critical Congenital Heart Disease (CCHD)* * Also known as critical congenital cyanotic heart disease, or CCCHD.	On October 15, 2010, SACHDNC recommended to the HHS Secretary the addition of "critical congenital cyanotic heart disease (CCCHD) to the recommended uniform screening panel with the understanding that the following activities will also take place in a timely manner: 1) The National Institutes of Health (NIH) shall fund research activities to determine the care provided and the health outcomes of affected newborns with critical congenital cyanotic heart disease as a result of prospective newborn screening; (2) the Centers for Disease Control and Prevention (CDC) shall fund surveillance activities to monitor the critical congenital cyanotic heart disease link to infant mortality and other health outcomes; (3) the Health Resources and Services Administration (HRSA) shall guide the development of screening standards and infrastructure needed for the implementation of a public health approach to point of service screening for critical congenital cyanotic heart disease; and (4) HRSA shall fund the development of, in collaboration with public health and health care professional organizations and families, appropriate education and training materials for families and public health and health care professionals relevant to the screening and treatment of CCCHD."	On **September 21, 2011**, the Secretary decided to adopt the SACHDNC's first recommendation to add CCHD to the RUSP. Additionally, the Secretary requested that the SACHDNC collaborate with HRSA to complete a thorough evaluation of the potential public health impact of universal screening for CCHD, as required by the authorizing statute. The Secretary stated that it would be beneficial to have the SACHDNC partner with HRSA and other public health experts to provide information to States, health care facilities, and individual clinicians about the impact of CCHD screening implementation and about the capabilities to ensure universal screening, as well as follow-up. The Secretary also decided to adopt the recommended activities directed towards NIH, CDC and HRSA, the Secretary, stating, *"I will direct the named agencies, as well as other relevant HHS agencies, to proceed expeditiously with implementation, as described [in the ICC Federal Agency Plan of Action], as feasible."* On **April 20, 2011**, the Secretary rejected SACHDNC recommendations, stating, *"As you noted in your letter, there are 'recognizable evidence gaps' regarding screening for Critical Congenital Cyanotic Heart Disease. After consultation with HHS agency leadership, I have determined that the Advisory Committee's recommendations are not ready for adoption. However, because this is such an important issue, I am referring these recommendations to the newly established Interagency Coordinating Committee on Newborn and Child Screening (ICC) for additional review and input regarding implementation.* *The ICC includes the National Institutes of Health, Centers for Disease Control and Prevention, Health Resources and Services Administration, Agency for Healthcare Research and Quality, and the Food and Drug Administration. ICC leadership will examine the evidence gaps described by the Advisory Committee, and propose a plan of action to address identification of effective screening technologies, development of diagnostic processes and protocols, education of providers and the public, and strengthening service infrastructure needs for follow-up and surveillance. The ICC will report this plan to me within 90 days, and I will keep you and the Advisory Committee informed."*

TOPIC	SACHDNC RECOMMENDATION	SECRETARY'S RESPONSE/OUTCOME
National Contingency Plan for NBS	On August 6, 2010, SACHDNC recommended to the Secretary, "In order to establish a comprehensive national all hazards approach to newborn screening incident response, the SACHDNC recommends that the Secretary of HHS coordinate newborn screening emergency preparedness activities, as defined in the CONPLAN, within HHS's National Response Framework."	On **September 2, 2011**, the Secretary responded as follows: *"I recognize the importance of federal interagency coordination of implementation of the CONPLAN, […], and I accept your recommendation.* *CDC, with the support from HRSA, will lead our efforts to coordinate implementation of the plan together with the Assistant Secretary for Preparedness and Response, […]."*
Retention/Use of Dried Blood Spot Specimens after NBS	On October 13, 2010, in order to address the potential to advance science and clinical care for newborns, children, their families and society through the use of residual newborn screening blood specimens and to protect those valuable resource for the public good SACHDNC made the following recommendations to the Secretary: "(1) All State newborn screening programs should have a policy in place that has been reviewed by the State Attorney General or other appropriate legal authority that specifies who may access and use dried blood specimens once they arrive at the state-designated newborn screening laboratory, including further access after newborn screening tests are completed. (2) All State newborn screening programs should have a policy in place that has been reviewed by the State Attorney General or other appropriate legal authority addressing the disposition of dried blood specimens remaining after newborn screening. Policymakers should consider the value of the specimens as a promising resource for research, the protection of the privacy and confidentiality of families and the necessity of ensuring the public's trust. (3) All State newborn screening programs should develop a well-defined strategy to educate health care professionals who provide patients with prenatal and postnatal care about newborn screening and the potential uses of residual dried blood specimens. (4) All State newborn screening programs should create policies that are in compliance with Federal research regulations, ensure that parents are aware of these activities, and consider whether documentation of parents' wishes and	On **April 13, 2011**, the Secretary rejected SACHDNC recommendations, stating, *"At this time, the Committee recommendations are not ready for adoption. Therefore, I am referring the Committee's report […] to the Interagency Coordinating Committee on Newborn and Child Screening (ICC) for their review and input regarding possible future implementation of the recommendations. The use of the ICC allows a more formal engagement of the Office of Human Research Protections and Office of Civil Rights, along with the Federal agencies assigned to the ICC by its authorizing legislation. I will encourage the ICC to submit a report with recommendations for appropriate HHS action by June 1, 2012."*

TOPIC	SACHDNC RECOMMENDATION	SECRETARY'S RESPONSE/OUTCOME
	willingness to participate are required. (5) All State newborn screening programs should work proactively to ensure that all families of newborns are educated about newborn screening as a part of prenatal and postnatal care. (6) The Secretary of Health and Human Services should help improve efforts to educate the public and health care providers about newborn screening and the retention and use of specimens. (7) The Secretary of Health and Human Services should facilitate a national dialogue among Federal and State stakeholders about policies for the retention and use of residual newborn screening specimens, including model consent and dissent processes. (8)The Secretary of Health and Human Services should explore the feasibility of establishing a voluntary national repository of residual dried blood specimens, in which families may choose to participate.	
Severe Combined Immunodeficiency (SCID)	Per the Request of the Secretary (dated May 21, 2010) to *"submit a report in May 2011 on the status of States' implementation of this recommendation, including the surveillance activities to be conducted through the Newborn Screening Translational Research Network,"* on May 19, 2011 the SACHDNC submitted a report entitled "Newborn Screening for Severe Combined Immunodeficiency." The report summarizes the current status of screening newborns for SCID in state-based newborn screening programs and surveillance updates from the Newborn Screening Translational Research Network (NBSTRN). It also includes the impact, successes, lessons learned, and next steps developed by expanding SCID pilot studies.	On **July 27, 2011**, the Secretary responded as follows: *"The report provides helpful information on current state and federal efforts to address the recommendation of the Secretary's Advisory Committee on Newborns and Children (SACHDNC) to add SCID to the Recommended Uniform Screening Panel.* *The collaboration among the Department of Health and Human Services' agencies to facilitate state and territory implementation of newborn screening for SCID, as well as the partnership demonstrated among state and territory public health newborn screening programs, has been influential in furthering improvement and expansion of screening for this life-threatening disorder. The pilot projects described in the report continue to generate new knowledge that will serve to improve early disease detection and treatment and provide valuable information to other states considering the addition of SCID to their state newborn screening panel. The report provides a valuable example of the importance of the SACHDNC in providing guidance to assist states and territories in implementing the most up-to-date research, technology, laboratory, and public health standards and practices as part of their newborn screening programs."*

TOPIC	SACHDNC RECOMMENDATION	SECRETARY'S RESPONSE/OUTCOME
Sickle Cell Disease Carrier Status, screening U.S. college athletes	On October 11, 2010, SACHDNC recommended to the Secretary the following revisions to recommendations made to the June 14, 2010 letter to the Secretary. The revisions were considered refinements to add clarity to the original SACHDNC recommendations, rather than changes in the content of the recommendations: Revised Recommendation 1: All individuals should have the opportunity to find out their risk for various medical disorders, including their carrier status for genetic conditions such as sickle cell disease. Revised Recommendation 2 (combining original Recommendations 2 and 3): Evaluation and testing for sickle cell disease and other genetic conditions should take place within the individual's medical home. That evaluation should include counseling regarding the implications of the information for the individual and assurance of the privacy of genetic information. Genetic testing should not be a prerequisite for participation in sports, unless deemed medically necessary. Revised Recommendation 3 (originally Recommendation 4): As part of the individual's annual medical evaluation for participation in sports, all potential athletes should receive education on safe practices for prevention of exercise and heat related illnesses. Revised Recommendation 4 (originally Recommendation 5): The Secretary, HHS, instruct SACHDNC to work with the SCDAA, relevant Federal HHS agencies, athletic associations, community based and health care professional organizations to develop guidelines and educational resources about screening for sickle cell trait in all persons, including athletes. Revised Recommendation 5 (originally Recommendation 6): The National institutes of Health and the Centers for Disease Control and Prevention conduct research to ascertain if some athletes with sickle cell trait are at increased risk of exercise-related sudden death."	On **June 27, 2011**, the Secretary decided to "support" revised recommendations 1-3, but determined that *"at this time the Committee's remaining two recommendations [revised recommendations 4 and 5] are not ready for adoption."* The Secretary also stated, "*I recently unveiled a Department-wide initiative to improve care for individuals with sickle cell disease. Multiple components of HHS are already engaged in sickle cell disease research and care, and this initiative builds upon ongoing activities by enhancing coordination and integration of these activities. In addition, this initiative has led to the identification of and commitment to pursue new and promising opportunities. For example, NIH will collaborate with CDC to conduct research on the clinical implications for individuals with sickle cell trait. Research topics are expected to include renal disease, risk of thrombosis, impact on stress related situations such as dehydration, athletic participation and vigorous physical activity, and access to genetic counseling. I am hopeful that this interagency effort will improve the knowledge base related to the health impacts of sickle cell trait and inform future efforts related to your remaining two recommendations that could not be adopted at this time."*

Appendix C: SACHDNC Members

SACHDNC members are appointed by the Secretary or designee, and shall not exceed 15 voting members, including the Chair and Federal Ex-Officio members. The Committee may also include up to 12 non-voting organizational representatives, as the Secretary determines necessary.

The Designated Federal Official from HRSA's Maternal and Child Health Bureau serves as the government's agent for matters related to the management of the SACHDNC's activities and ensures all procedures are within applicable statutory, regulatory, and HHS General Administration Manual directives.

For 2011, the following individuals were members of the SACHDNC.

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Appendix D: Glossary

Critical Congenital Heart Disease (CCHD)	Group of defects present in the heart structure at birth, causing severe and life-threatening symptoms and requiring intervention within the first year of life.
Heritable Disorders	Group of genetically inherited conditions present at birth that, undetected, can cause intellectual/physical disabilities and life-threatening illnesses.
Neonatal Hyperbilirubinemia	Condition distinguished by a build-up of bilirubin in an infant that, untreated, results in severe jaundice and serious health complications (Mayo, 2012).
Newborn Screening (NBS)	Practice of testing babies for heritable disorders and conditions that can hinder their normal development, to enable early detection/treatment and prevent intellectual/physical disabilities and life-threatening illnesses.
Recommended Uniform Screening Panel (RUSP)	Standard guideline for the newborn screening of heritable conditions, consisting of a list of conditions referred to as a screening panel. This panel provides guidance to the states regarding the latest evidence-based medical recommendations for newborn screening.
Severe Combined Immunodeficiency (SCID)	Primary immune deficiency disease distinguished by the lack of T-cells and B-cell function that, untreated, results in vulnerability to opportunistic infections and early death (SCID, 2012).
Sickle Cell Disease Carrier	Individual who "carries" sickle cell mutation (i.e., sickle cell trait), is clinically benign under ordinary circumstances, and often asymptomatic. This is due to the carrier possessing a recessive, disease-causing allele on one chromosome and a normal allele on the other chromosome.

Appendix E: Acronyms

ACMG	American College of Medical Genetics and Genomics
APHL	Association of Public Health Laboratories
CCCHD	Critical Congenital Cyanotic Heart Disease
CCHD	Critical Congenital Heart Disease
CDC	Centers for Disease Control and Prevention
EERW	External Evidence Review Workgroup
FDA	Food and Drug Administration
HHS	Health and Human Services
HRSA	Health Resources and Services Administration
ICC	Interagency Coordinating Committee on Newborn and Child Screening
IOM	Institute of Medicine
LOINC	Logical Observation Identifiers Names and Codes
LTFU	Long-Term Follow-Up
NBS	Newborn Screening
NBSC	Newborn Screening Clearinghouse
NBSTRN	Newborn Screening Translational Research Network
NCQA	National Committee for Quality Assurance
NIH	National Institutes of Health
NQF	National Quality Forum
POC	Point-of-Care
RUSP	Recommended Uniform Screening Panel
SACHDNC	Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
SCID	Severe Combined Immunodeficiency
TREC	T-cell Receptor Excision Circle