



## Issue Brief: Newborn Screening

*AAFP State Government Relations  
AAFP Commission on Science*

### Issue

The Maternal and Child Health Bureau of the federal Health Resources and Services Administration (HRSA) has recommended (2004) that states implement a uniform set of newborn screening tests through legislation or regulation. The report outlining the recommendations, *Newborn Screening: Toward a Uniform Screening Panel and System*, was developed by the American College of Medical Genetics (ACMG) under a contract from HRSA. A myriad of new genetic tests are available to detect genetic conditions in newborns. The report calls for states to screen for twenty-nine (29) genetic conditions (see the Appendix I) using a new technology called tandem mass spectrometry (MSMS). The screening requires blood from a single heel stick in the newborn nursery.

The report and recommendations, which were provided to AAFP and others for comment in mid-2005, have created some controversy. The two main concerns raised by some in the primary care community are that: (1) the report shows relatively little recognition of the role of primary care clinicians, who care for newborns and their mothers and to whom families will turn to sort out positive test results; and (2) the evidence basis for the recommendations is less rigorous than that used by the federal government through the US Preventive Services Task Force (see Appendix V).

The AAFP review of this report resulted in the following findings:

- The panel experts who developed these recommendations were composed primarily of specialists in the field of genetics and newborn screening. This composition may have introduced a bias towards screening absent due consideration of the primary patient-physician relationship.
- The report reflects the position that conditions detectable as a by-product of screening for other conditions should be included in the screening panel, whether or not treatments are available.
- The report gives comparatively little weight to the potential negative effects of screening, including the prevalence of false positives and the deleterious effects they can have, such as added cost and patient anxiety.
- The report pays insufficient attention to the effects on patients and families of abnormal test results for conditions that have no treatment or may represent only carrier states.

### State Activity

States are already introducing legislation and/or regulatory changes to current state laws governing newborn screening (see Appendix III). As of January 18, 2006, there were sixteen

bills pending in nine states addressing newborn screening. The National Conference of State Legislatures (NCSL) held a Newborn Screening Conference for state legislators and legislative staff on September 28th, 2005, to discuss advances in technology and their implications for states. The conference suggests strongly that the issue will generate activity at the state level. In 2005, thirty-eight (38) screening bills were filed, with thirteen (13) enacted and one resolution adopted (see Appendix IV for summaries of enacted and pending bills).

AAFP constituent chapters may have a role in defining such legislation by engaging in collaborative discussions with members and other organizations to ensure that state screening programs are evidence based.

## **AAFP Policy**

The AAFP supported the Genetic Nondiscrimination in Health Insurance and Employment Act (HR 2457; see <http://www.aafp.org/x6831.xml>). The AAFP currently recommends newborn screening for congenital hypothyroidism, hemoglobinopathies and phenylketonuria.

The AAFP Commission on Science has proposed the following recommendations to guide chapters in participating in decisions around newborn screening in the states:

*1. AAFP constituent chapters and family physicians should become involved, early in 2006, in legislative and administrative procedures in their states, to contribute to the states' response to the federal communication recommending a uniform panel of 29 newborn screening tests using Tandem Mass Spectrometry;*

*2. Chapters should consider recommending that their states give consideration to mandatory newborn screening for those tests for which the evidence is most rigorously supportive, and that families be appropriately informed for consent, that addresses both benefits and potential harms, to reporting the remaining tests in the Tandem Mass Spectrometry panel (i.e. the model used for screening and follow-up in Massachusetts, Rhode Island, Vermont, New Hampshire and Maine through the New England Newborn Screening Program as conducted by the University of Massachusetts Medical School (<http://www.umassmed.edu/nbs/>), See Appendix II);*

*3. Family physicians and their office staffs should prepare to educate families concerning newborn screening, and to respond to questions from families concerning positive tests. (To assist with such preparation, please see AAFP Web-based module on Newborn Screening through the Annual Clinical Focus 2005: Genomics at <http://www.aafp.org/x25023.xml>. Patient education materials developed by the Maternal and Child Health Bureau included in the program's Web tour - <http://www.aafp.org/x37688.xml> - address the significance of newborn screening tests, why newborns should be tested, how they will be tested, how test results can be obtained, and retesting concerns.)*

Additionally, the AAFP Commission on Science recommends that:

*If states adopt the panel of 29 tests for newborn screening, state health departments should collect data on the benefits and harms associated with each test, and then tests without practical*

*clinical benefits, or that have harms that outweigh their benefits, should be eliminated from the panel;*

*An additional panel of 28 tests under consideration in the HRSA report should not be added until adequate evidence is gathered on how the first 29 perform; and*

*Tests considered for addition to the screening program should be evaluated individually, not as a panel.*

Please direct questions related to this Issue Brief to:

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## **Appendix I**

### **The 29 Conditions Included in the Recommended Screening Panel**

- 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
- 3-OH 3-CH<sub>3</sub> glutaric aciduria (HMG)
- Argininosuccinic acidemia (ASA)
- Biotinidase deficiency (BIOT)
- Carnitine uptake defect (CUD)
- Citrullinemia (CIT)
- Classical galactosemia (GALT)
- Congenital adrenal hyperplasia (21-hydroxylase deficiency) (CAH)
- Congenital hypothyroidism (CH)
- Cystic fibrosis (CF)
- Glutaric acidemia type I (GA I)
- Hb S/C disease (Hb S/C)
- Hb S/ $\beta$ -thalassemia (Hb S/ $\beta$ Th)
- Hearing loss (HEAR)
- Homocystinuria (due to CBS deficiency) (HCY)
- Isovaleric acidemia (IVA)
- Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD)
- Maple syrup disease (MSUD)
- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- Methylmalonic acidemia (Cbl A,B) (Cbl A,B)
- Methylmalonic acidemia (mutase deficiency) (MUT)
- Multiple carboxylase deficiency (MCD)
- Phenylketonuria (PKU)
- Propionic acidemia (PROP)
- Sickle Cell Anemia (Hb SS)
- $\beta$ -Ketothiolase deficiency (BKT)
- Trifunctional protein deficiency (TFP)
- Tyrosinemia type I (TYR I)
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

**Appendix II**  
**New England New Born Screening Program**  
**(Maine, New Hampshire, Rhode Island, Vermont)**

*Routine Disorders:*

- Congenital Hypothyroidism
- Phenylketonuria (PKU)
- Hemoglobin Disorders
- Congenital Toxoplasmosis
- Biotinidase Deficiency
- Galactosemia
- “Maple Syrup” Urine Disease
- Homocystinuria
- Congenital Adrenal Hyperplasia
- Medium-chain acyl Co-A dehydrogenase deficiency (MCAD)

*Optional Disorders*

- Cystic Fibrosis
- Tyrosinemia I and II
- HMG Lyase Deficiency
- Argininosuccinic Aciduria
- Isovaleric Acidemia
- HHH Syndrome
- Glutaric Acidemia I and II
- Citrullinemia
- Argininemia
- CPT Deficiency
- Propionic Acidemia
- Methylmalonic Aciduria
- B-Methyl Crotonyl Carboxylase
- LCHAD – long-chain hydroxyacyl-CoA dehydrogenase deficiency
- VLCAD – very-long-chain acyl-CoA dehydrogenase deficiency
- SCAD – short-chain acyl-CoA dehydrogenase deficiency
- LCAD – long-chain acyl-CoA dehydrogenase deficiency
- B-Ketothiolase Deficiency – 2-methylacetoacetyl-CoA thiolase deficiency

**Appendix III  
Newborn Screening Program Statutes and 2005 Enacted Legislation**

<a href="#">Alabama</a>	Ala. Code §22-2-03
<a href="#">Alaska</a>	Alaska Stat. §18-15-200, 210
<a href="#">Arizona</a>	Ariz. Rev. Stat. §36-694
<a href="#">Arkansas</a>	Ark. Code Ann. §20-15-301 to 304, <a href="#">2005 HB 2428</a>
<a href="#">California</a>	Cal. Health and Safety Code §124975 to 125001
<a href="#">Colorado</a>	Colo. Rev. Stat. §25-4-1001 to 1006
<a href="#">Connecticut</a>	Conn. Gen. Stat. Ann. §19a-55
<a href="#">Delaware</a>	Del. Code §16.2.201 to 206
<a href="#">District of Columbia</a>	D.C. Code Ann. § 7-831 to 840
<a href="#">Florida</a>	Fla. Stat. Ann. 29 §383.14
<a href="#">Georgia</a>	Ga. Code §31-12-5 to 7
<a href="#">Hawaii</a>	Hawaii Rev. Stat. §-321-291
<a href="#">Idaho</a>	Idaho Code §39-909 to 912
<a href="#">Illinois</a>	410 ILCS 240/.01 to .03
<a href="#">Indiana</a>	Ind. Code §16-41-7
<a href="#">Iowa</a>	Iowa Code §136A.1 to 7
<a href="#">Kansas</a>	Kan. Stat. Ann. §65-180 to 183
<a href="#">Kentucky</a>	Ky. Rev. Stat. §214.155, <a href="#">2005 SB 24</a>
<a href="#">Louisiana</a>	40 §1299
<a href="#">Maine</a>	Me. Rev. Stat. Ann. tit. 22 §1531 to 1533
<a href="#">Maryland</a>	Md. Health Code Ann. §13-101 to 110
<a href="#">Massachusetts</a>	Mass. Gen. Laws 111 §3, 4E, 5, 6, 24A, 110A
<a href="#">Michigan</a>	Mich. Comp. Laws §333.5431
<a href="#">Minnesota</a>	Minn. Stat. §144.125 to 128
<a href="#">Mississippi</a>	Miss. Code Ann. §41-21-201 to 205; §41-24-1 to 5
<a href="#">Missouri</a>	Mo. Rev. Stat. §191.331, 332
<a href="#">Montana</a>	Mont. Code Ann. §50-19-201 to 211, <a href="#">2005 SB 275</a>
<a href="#">Nebraska</a>	Neb. Rev. Stat. §71-519 to 24, <a href="#">2005 LB 301</a>
<a href="#">Nevada</a>	Nev. Rv. Rev. Stat. §442.008
<a href="#">New Hampshire</a>	N.H. Rev. Stat. Ann. §132:10-a to d
<a href="#">New Jersey</a>	N.J. Stat. Ann. §26:2-510 and 511
<a href="#">New Mexico</a>	N.M. Stat. Ann. §24-1-6, <a href="#">2005 HB 479</a>
<a href="#">New York</a>	N.Y. Public Health Law §2500-a
<a href="#">North Carolina</a>	N.C. Gen. Stat. §130A-125
<a href="#">North Dakota</a>	N.D. Cent. Code §23-01-03.1; 25-17-00.1 to .5
<a href="#">Ohio</a>	Ohio Rev. Code Ann. §3701.50.1 to .9
<a href="#">Oklahoma</a>	Okla. Stat. §63-1-533, 534, <a href="#">2005 HB 1419</a>
<a href="#">Oregon</a>	Or. Rev. Stat. §433.285 to .295, §192.531 to .549
<a href="#">Pennsylvania</a>	Penn. Statutes 35 §621-625
<a href="#">Rhode Island</a>	R.I. Gen. Laws §23-13-14
<a href="#">South Carolina</a>	S.C. Code Ann. §44-37-30

<a href="#">South Dakota</a>	S.D. Codified Laws Ann. §34-24-17 to 25
<a href="#">Tennessee</a>	Tenn. Code Ann. §68-5-401 to 506
<a href="#">Texas</a>	Tex. Code Ann. Health and Safety §33.001 to .038, <a href="#">2005 HB 790</a>
<a href="#">Utah</a>	Utah Code Ann. §26-10-6
<a href="#">Vermont</a>	18 V.S.A. §115
<a href="#">Virginia</a>	Va. Code §32.1-65-69, <a href="#">2005 HB 1824/2005 SB 1184</a>
<a href="#">Washington</a>	Wash. Rev. Code §70.83.020 to .050
<a href="#">West Virginia</a>	W.V. Code §16-22-1 to 6
<a href="#">Wisconsin</a>	Wis. Stat. Ann. §253.13
<a href="#">Wyoming</a>	Wyo. Stat. §35-4-801 to 802

**Source:** NCSL, West Group, StateNet (June 2005),  
<http://www.ncsl.org/programs/health/genetics/newborn.htm>

**Appendix IV  
Enacted and Pending 2005 and 2006 Legislation**

*Arkansas*

**HB 2428**

**Sponsor:** Harrelson

**Status:** **Enacted**

**Summary:** As introduced: This bill requires newborn screening for cystic fibrosis, allows the health department to expand newborn screening if reliable and efficient testing techniques are available, and addresses follow up and privacy issues.

*Kentucky*

**2005 SB 24**

**Sponsor:** Denton

**Status:** **Enacted**

**Summary:** As signed by the Governor: This bill requires the Cabinet for Health Services to operate a newborn screening program that includes screening tests and definitive diagnostic evaluations for positive tests; requires administrative regulations to include the manner of testing, recording, and reporting of screening tests; establishes fees for the screening program; specifies the conditions included in a screening test; requires information to be provided to a parent about the screening tests and require an institution or health care provider to arrange for appropriate follow-up; requires information about tests not specified to be provided to a parent and specify the parent is responsible for costs associated with additional screening tests; defines qualified laboratory; requires the cabinet to contract with qualified laboratory if the cabinet is unable to screen for the specified conditions; permits the cabinet to contract with qualified laboratories for conditions not specified; specifies that cabinet contracts for unspecified conditions does not preclude other institutions or providers from utilizing other qualified laboratories for tests for unspecified conditions; permits the cabinet to solicit and accept private funds to expand, improve or evaluate the newborn screening program; provides a short title the James William Lazzaro and Madison Leigh Heflin Newborn Screening Act.

**2006 SB 18**

**Sponsor:** Denton

**Status:** **Pending**

**Summary:** As introduced: Amends statute pertaining to the Medical Assistance Program to include low-protein modified food products for the treatment of genetic and metabolic diseases for which the cabinet conducts newborn screening tests as required under KRS 214.155

*Michigan*

**SB 794**

**Sponsor:** George

**Status:** **Pending**

**Summary:** As introduced: This bill creates a newborn screening quality assurance advisory committee at the public health department.

*Mississippi*

**2005 SB 2511**

**Sponsor:** Nunnelee

**Status:** **Enacted**

**Summary:** As enacted: This bill extends the term of the Infant Mortality Task Force and direct the group to conduct a study of oxygen saturation screening of newborns for congenital heart disease.

**2006 S 2375**

**Sponsor:** Dawkins

**Status:** Pending

**Summary:** Screening for newborn disorders; add additional tests and authorize Medicaid and private insurance reimbursement.

*Montana*

**SB 275**

**Sponsor:** Schmidt

**Status:** Enacted

**Summary:** Introduced version: This bill raises the fee for the stateside genetics program to \$1 until July 1, 2007, and makes changes to the genetics program pertaining to the comprehensive nature of the program and contracting genetic services.

**SB 275**

**Sponsor:** Schmidt

**Status:** Enacted

**Summary:** As signed by the Governor: This bill amends the state statute for the state voluntary genetics program, including increasing fees.

*Nebraska*

**LB 301**

**Sponsor:** Jensen

**Status:** Enacted

**Summary:** Introduced version: This bill amends section 71-1,104.01 informed consent requirements related to genetic testing and newborn screening.

*New Hampshire*

**SB 108**

**Sponsor:** Martel

**Status:** Pending

**Summary:** Introduced version: This bill establishes criteria for adding disorders to the newborn screening panel. Carryover to 2006

*New Mexico*

**H479**

**Sponsor:** King

**Status:** Enacted

**Summary:** As signed by the Governor: This bill expands the state newborn screening panel.

*New York*

**A2768**

**Sponsor:** Brodsky

**Status:** Pending

**Summary:** Introduced version: This bill requires hospitals to provide parents of newborns with the option to have a DNA test performed on their child or have a blood sample taken from the child and preserved for future DNA testing.

*North Carolina*

**SB622**

**Sponsor:** Garrou, Dalton, Hagan

**Status:** Enacted

**Summary:** As enacted: This bill imposes a \$14 fee for newborn screening.

*Oklahoma*

**HB 1419**

**Sponsor:** Kiesel

**Status:** Enacted

**Summary:** Introduced version: This bill defines phenylketonuria and authorizes the state board of health and the Oklahoma health care authority board to promulgate certain rules.

**HB 1828**

**Sponsor:** Miller, R.

**Status:** **Pending**

**Summary:** As introduced: Clarifies language in newborn screening statute pertaining to parent education.

*Pennsylvania*

**HB 755**

**Sponsor:** Kenney

**Status:** **Pending**

**Summary:** As introduced: This bill provides for additional newborn screening.

**SB 819**

**Sponsor:** Orié

**Status:** **Pending**

**Summary:** As introduced: This bill expands the state newborn screening program and appropriates \$2,000,000 to carry out the changes.

**SB 901**

**Sponsor:** Boscola

**Status:** **Pending**

**Summary:** As introduced: This bill adds Severe Combined Immunodeficiency (SCID) to the list of newborn screening conditions screened for by the state.

*Tennessee*

**HJR 373**

**Sponsor:** DeBerry

**Status:** **Adopted**

**Summary:** As introduced: This resolution urges the Department of Health to coordinate its genetic services with its early intervention services and to continue to provide exemplary care for young Tennesseans.

*Texas*

**HB 790**

**Sponsor:** Crownover

**Status:** **Enacted**

**Summary:** Introduced version: This bill relates to the equipment and employees necessary for the conduct of newborn screening by the Department of State Health Services.

*Virginia*

**HB 1824**

**Sponsor:** Frederick

**Status:** **Enacted**

**Summary:** As signed by the Governor: This bill broadens the Commonwealth's newborn screening program for genetic disorders to include approximately 30 or more conditions that cause mental retardation, serious disability, or death if left untreated.

**SB 1184**

**Sponsor:** Puller

**Status:** **Enacted**

**Summary:** As signed by the Governor: This bill broadens the Commonwealth's newborn screening program for genetic disorders to include approximately 30 or more conditions that cause mental retardation, serious disability, or death if left untreated.

*Washington*

**HB 1537**

**Sponsor:** Schual-Berke

**Status:** Pending

**Summary:** Introduced version: This bill clarifies when the department of health may collect a fee for newborn screening services.

**SB 5491**

**Sponsor:** Poulsen

**Status:** Pending

**Summary:** Introduced version: This bill clarifies when the department of health may collect a fee for newborn screening services.

*West Virginia*

**HB 2447**

**Sponsor:** Staton

**Status:** Pending

**Summary:** Introduced version: This bill adds conditions to the list of disorders screened for under the newborn screening program.

**SB 231**

**Sponsor:** Hunter

**Status:** Pending

**Summary:** Introduced version: This bill expands newborn screening to include screening for sickle cell disease.

**HB 2607**

**Sponsor:** Leach

**Status:** Pending

**Summary:** As introduced: This bill adds several conditions to the state newborn screening panel.

**HB 2766**

**Sponsor:** Webster

**Status:** Pending

**Summary:** As introduced: This bill adds sickle cell anemia to the panel of newborn screening tests.

**HB 2969**

**Sponsor:** Marshall

**Status:** Pending

**Summary:** As introduced: This bill adds several conditions to the panel of newborn screening tests.

**Source:** NCSL, StateNet (June 2005)

## **Appendix V Methods for Development of Evidence-based Recommendations**

The AAFP develops evidence-based clinical practice recommendations for family physicians. The development process includes review of the evidence which then becomes the basis for specific recommendations. For its clinical practice guideline development process, the AAFP nominates a clinical topic to the Agency for Healthcare Research and Quality (AHRQ) for an evidence report by one of the Evidence-based Practice Centers (EPC). The AAFP then uses the evidence report as the basis for development of clinical practice guideline recommendations.

For clinical preventive services recommendations, the AAFP uses the evidence reports and summaries of the U.S. Preventive Services Task Force (USPSTF) conducted by the Oregon Health Science University (EPC). The USPSTF conducts rigorous, impartial assessments of the scientific evidence for the effectiveness of a broad range of clinical preventive services, including screening, counseling, and preventive medications. Its recommendations are considered the "gold standard" for clinical preventive services. For the majority of their clinical preventive services recommendations, the AAFP agrees with the USPSTF. However, there are some instances where the Academy differs from the USPSTF. These are where the AAFP develops recommendations which are more pertinent to the family medicine.

Current methods of the U.S. Preventive Services Task Force for development of recommendations can be found at: <http://www.ahrq.gov/clinic/ajpmsuppl/harris1.htm>

The AAFP Summary of Recommendations for Clinical Preventive Services can be found at: <http://www.aafp.org/exam.xml>