



Summary:  
Washington State Newborn Screening Advisory Committee Meeting  
September 24, 2007

The Washington State Newborn Screening Advisory Committee met for a full day at the Department of Health's Public Health Laboratories facility in Shoreline on September 24, 2007. The committee members represented a broad spectrum of individuals and groups who have specific interest in our state's newborn screening program. The membership includes representatives of parents, children's advocacy groups, professional associations, medical/clinical specialties, principal payers of medical costs, medical ethics, and public health. A complete list is included at the end of this summary.

The meeting was the first of two that will review seventeen conditions as potential candidates for addition to Washington's required screening panel. Sixteen of the conditions were recommended for inclusion in all states' newborn screening programs in a report commissioned by the federal Health and Human Resources Administration (HRSA) from the American College of Medical Genetics (ACMG). The seventeenth condition was added during a meeting of Washington medical and public health experts that was convened earlier to provide technical input to the advisory committee. All of the conditions are detectable by adjusting the tandem mass spectrometry equipment already in use by the program.

The morning was spent providing background and context for committee members. Co-chairs Diana Yu, MD, MSPH, Washington State Board of Health member, and State Health Officer Maxine Hayes, MD, MPH, Department of Health, explained that the State Board of Health has the responsibility for making any changes to the screening panel. They charged the committee to provide advice to the board based on an evaluation of each of the conditions against the board's established criteria for making additions to the screening panel.

The five criteria were reviewed and the committee was informed that because of complexity, the final criterion regarding costs and benefits would not be evaluated until the committee determines that the other four criteria are met. The co-chairs also shared the findings of an earlier committee of medical and public health experts who reviewed the conditions in light of medical/scientific evidence for two of the criteria: a) availability of appropriate and effective screening, diagnosis, treatment, and systems for evaluation and care; and b) availability of sensitive, specific, and timely tests that can be adapted to mass screening.

The committee was provided a brief historical overview of Washington State's newborn screening program and a description of its current operations. The medical, technical, and political environment that led to the ACMG recommendations was presented and there was discussion of the differing perspectives on expanding screening as reflected in two articles published in the medical journal *Pediatrics*\*. A summary prepared by Washington's March of Dimes chapter was shared which indicates that most states have added the recommended disorders to their screening panels. It was pointed out that while some groups\*\* have supported

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\* Botkin JR, Clayton EW, Fost NC, et al, Newborn screening technology: proceed with caution [commentary]. *Pediatrics*. 2006;117:1793-1799. & Howell RR, We need expanded newborn screening [commentary]. *Pediatrics*. 2006;117:1800-1805.

\*\* American Academy of Pediatrics, March of Dimes, and the Secretary of Health and Human Services' Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns' and Children.

the ACMG recommendations, the federal Department of Health and Human Services (parent agency of HRSA which commissioned the study), has yet to provide official response, despite having solicited and closed public input over two years ago (May 2005).

Following the introductions and background presentations, the committee began reviewing individual disorders. Following discussion and review of each disorder, members were asked to complete a ballot with check boxes for each of the four criteria: prevention potential and medical rationale, treatment available, public health rationale, and available technology. The check boxes provided the options “yes,” “no,” and unsure.” The committee was able to complete reviews of ten of the seventeen disorders. The Table on the next page shows the outcome of these reviews. Although most disorders received affirmative scoring for most of the criteria, only isovaleric acidemia and glutaric acidemia type 1 received unanimous support on all four criteria. The most frequent non-affirmative scores were “unsure.”

Discussion common to many of the reviews included questions about the potential negative aspects of screening—such as possible unintended harm caused by rigorous diagnostic and treatment regimens, the overall effectiveness of early detection and intervention for rare conditions, and the impact of disorders not screened for and identified at birth. The wide spectrum of clinical manifestations from mild to severe reported in the literature among both screened and unscreened populations was important to the discussion for several of the conditions such as the methylmalonic acidemias.

There was considerable discussion of the potential impacts of positive screening results for children who are found to be negative during diagnostic evaluation. These false positive results impact both affected families and the diagnostic workload. Questions were also raised about the overall impact of additional screening on our state’s clinical care and treatment systems. Material summarized from the medical literature and the experience of other newborn screening programs was presented which suggests that, if all the conditions were incorporated in the screening panel, about 15 to 20 affected children would be detected annually in Washington and, at most, about 75 to 100 screening tests would be false positives.

The ten conditions reviewed at the meeting were also found to meet the “treatment available,” and “available technology” criteria applied by the earlier committee of medical and public health experts. The remaining seven conditions were determined to be in somewhat less agreement with the two criteria by the earlier committee. These seven remaining disorders will be reviewed at the next meeting which is scheduled for December 10, 2007 at the Department of Health’s Shoreline facility. The disorders to be reviewed are:

- 3-hydroxy-3-methylglutaric aciduria (HMG)
- Beta-ketothiolase deficiency (BKT)
- Carnitine palmitoyl transferase deficiency type 1-A (CPT)
- Holocarboxylase synthase deficiency (HSCD)
- Carnitine uptake deficiency (CUD)
- 3-methylcrotonyl-CoA carboxylase deficiency (3-MCC)
- Tyrosinemia type 1

**TABLE:** Newborn Screening Advisory Committee  
Scoring of Disorders for 4 of 5 Board's Criteria

DISORDER	ACMG Abbreviation Code	Meets Criteria?	Prevention Potential and Medical Rational	Treatment Available	Public Health Rational	Available Technology
Isovaleric acidemia	IVA	Yes	16	16	16	16
		No	0	0	0	0
		Unsure	0	0	0	0
Glutaric acidemia type1	GA-1	Yes	16	16	16	16
		No	0	0	0	0
		Unsure	0	0	0	0
Methylmalonic acidemia (Cbl A, B)	Cbl A,B	Yes	14	14	15	15
		No	0	0	0	0
		Unsure	2	2	1	1
Methylmalonic acidemia (mutase deficient)	MUT	Yes	14	12	13	15
		No	0	0	0	0
		Unsure	2	4	2	1
Propionic acidemia	PROP	Yes	14	12	14	15
		No	0	0	1	0
		Unsure	2	4	1	1
Long chain acyl CoA dehydrogenase deficiency	LCHAD	Yes	16	15	16	16
		No	0	0	0	0
		Unsure	0	1	0	0
Trifunctional protein deficiency	TFP	Yes	16	13	14	15
		No	0	0	0	0
		Unsure	0	3	2	1
Very long chain acyl CoA dehydrogenase deficiency	VLCAD	Yes	15	14	16	16
		No	0	1	0	0
		Unsure	1	1	0	0
Citrullinemia type 1*	CIT	Yes	11	13	12	14
		No	1	0	0	0
		Unsure	1	1	2	0
Argininosuccinic acidemia*	ASA	Yes	11	13	12	14
		No	1	0	0	0
		Unsure	1	1	2	0

\* Two members were not present when Citrullinemia type 1 and Argininosuccinic acidemia were reviewed

**Members of the Newborn Screening Advisory Committee  
Consideration of Additional Conditions for the Required Panel**

September 24, 2007

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Purpose: To review and make recommendations to the Board regarding which conditions should be considered for addition to our state's screening panel.

**Parents**

1. Diane Nielsen, parent of child with Tri-functional Protein deficiency.
2. Phil Hazel, parent of child with phenylketonuria.

**Childrens Advocacy Groups**

3. Peggy Harris, Save Babies Through Screening Foundation
4. Cherish Hart, March of Dimes, Washington Chapter
5. Sue Elliot, Arc of Washington State

**Professional Associations**

6. \*Susan Blackburn, PhD, FAAN, RNC, Washington State Nursing Association
7. \*Brenda Suiter, Washington State Hospital Association
8. \*Nancy Hanson, MS, CGC Genetic Advisory Committee

**Medical/Clinical Specialties**

9. C. Ronald Scott, MD, Biochemical Genetics, University of Washington
10. Sihoun Hahn, M.D., Ph.D., Children's Hospital and Regional Medical Center
11. Judith Martin, MD, Inland Northwest Genetics Clinic

**Principle Payers**

12. Nancy Anderson, MD, MPH, Department of Social and Health Services, Medical Assistance Administration
13. Nancy Fisher, MD, Washington State Health Care Authority
14. Rana Hilderbrand, Association of Washington Healthcare Plans
15. Donna Dorris, Office of the Insurance Commissioner

**Medical Ethics**

16. Benjamin Wilfond, MD, Trueman Katz Center for Pediatric Bioethics at Children's Hospital and Regional Medical Center

**Public Health**

17. Maxine Hayes, MD, MPH, State Health Officer, Washington State Department of Health
18. Diana Yu, MD, MSPH, Washington State Board of Health
19. Tom Locke, MD, MPH, Washington State Association of Local Public Health Officials

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\* Not in attendance at 9/24/07 meeting