

Title: Use of Technical Experts as Advisors to Broader Newborn Screening Advisory Committee

Presented at the 2007 Newborn Screening and Genetic Testing Symposium

Authors: John D. Thompson and Michael Glass,
Newborn Screening Program
Washington State Department of Health
1610 NE 150th St.
Shoreline, WA 98155

Objectives: Candidate conditions for newborn screening (NBS) often present complex issues of natural history and clinical validity/utility of screening tests. We sought to avoid the potential for these issues to dominate deliberations by key stakeholders who are not medical/laboratory experts. We did this by convening a group of experts to consider the technical merits of candidate disorders. The findings of this technical group will be used to inform a broadly representative group of stakeholders who will consider the range of issues related to adding conditions to the screening panel and make recommendations to our State Board of Health.

Methodology: The technical advisory committee met on September 5, 2006 to consider expansion using tandem mass spectrometry for the 16 disorders recommended by the ACMG but not screened for in Washington. See the table below for membership of the committee. Follow-up staff from Washington's NBS program compiled a set of recent publications regarding the candidate disorders. The objective was for the technical advisory committee to evaluate the conditions for two of the five NBS screening criteria used in Washington:

1. Treatment Available: Appropriate and effective screening, diagnosis, treatment, and systems are available for evaluation and care.
2. Available Technology: Sensitive, specific, and timely tests are available that can be adapted to mass screening.

Results: Discussion among committee members lead to the following informal rankings:

Concordance with Treatment and Screening Technology criteria

Most

- Isovaleric acidemia (IVA)
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Long-chain L-3-OH acyl-CoA dehydrogenase deficiency (LCHAD)
- Trifunctional protein deficiency (TFP deficiency)
- Glutaric acidemia type 1 (GA-1)
- Methylmalonic acidemia – mutase deficient – vitamin B12 non-responsive (MUT)

- Methylmalonic academia – Cbl A,B – vitamin B12 responsive (CblA,B)
- Propionic Acidemia (PROP)
- 3-hydroxy-3-methylglutaric aciduria (HMG)
- Beta-Ketothiolase deficiency (BKD)
- Citrullinemia Type 1 (CIT)
- Argininosuccinic Acidemia (ASA)
- *Carnitine Palmitoyl Transferase 1 Deficiency (CPT-1)**

Intermediate

- Holocarboxylase synthase deficiency (HCSD)/Multiple carboxylase deficiency MCD
- Carnitine uptake deficiency (CUD)

Less

- 3-methylcrotonyl-CoA carboxylase deficiency (3-MCC)
- Tyrosinemia type 1 (TYR-1)

* Not included in the recommended panel, added by Technical Advisory Committee

Committee membership

Co-chairs of the newborn screening advisory committee:

Maxine Hayes, M.D., M.P.H. – Washington State Health Officer

Kim Thorburn, M.D., M.P.H. – (former) Chair, Washington State Board of Health

Medical experts:

C. Ronald Scott, M.D. – biochemical genetics

Sihoun Hahn, M.D., Ph.D. – biochemical genetics

Judith Martin, M.D. – clinical geneticist

Beth Ogata, M.S., R.D., C.D. – metabolic nutritionist

Also in attendance

Newborn screening staff:

Mike Glass, M.S. – program director

Santosh Shaunak, B.S. – laboratory coordinator

Sheila Weiss, M.S. – follow-up coordinator

William Hoffman, B.S. – MS/MS laboratory lead

John D. Thompson, M.P.H., M.P.A. – follow-up consultant

Board of Health staff:

Tara Wolff, MPH – health policy advisor

Conclusions: The use of a sub-committee allowed experts to combine their knowledge and provide guidance about the technical aspects of the NBS conditions being considered by the broader advisory committee.

Update: The State Board of Health co-chair position of the advisory committee was vacant for three months. Due to scheduling conflicts, it has taken some time to bring the new co-chair up to speed. The broader advisory committee will convene in the fall of 2007. Materials and findings from the advisory committee will be provided to members of the broader advisory committee prior to the full committee meetings. We believe this process will be helpful to the remainder of the advisory committee members that do not have expertise in treatment and screening technology for these rare conditions.