



Summary:
Washington State Newborn Screening Advisory Committee
Second Meeting December 10, 2007

The Washington State Newborn Screening Advisory Committee met for the second time at the Department of Health's Public Health Laboratories facility in Shoreline on December 10, 2007. The meeting continued the work of the committee to review seventeen conditions as potential candidates for addition to Washington's required screening panel. Ten of the conditions were reviewed by the committee at its earlier meeting on September 24, 2007.

The meeting began with co-chairs Diana Yu, MD, MSPH, Washington State Board of Health member, and State Health Officer Maxine Hayes, MD, MPH, Department of Health, reviewing the Advisory Committee's role to provide advice to the Board on the possible addition of new disorders to the state's required newborn screening panel. The co-chairs also reviewed the committee's work to date in evaluating ten of the candidate conditions against four of the Board of Health's five criteria for including disorders in the screening panel: prevention potential and medical rationale, treatment available, public health rationale, and available technology.

A process for applying the fifth criteria, cost benefit/cost effectiveness was discussed: Department of Health staff will develop economic models for each of the conditions that the committee determines to meet the other four criteria. The models will include significant outcomes predicted with, and without screening. Then, an economist consultant will be hired to populate the models with cost estimates so that the costs of screening versus not screening can be compared. It was agreed that the completed models would be shared with committee members followed by a conference call to discuss and answer questions before voting on whether each condition meets the cost criteria.

Following discussion of the process for the cost analysis the committee began work to evaluate each of the remaining seven conditions against the four criteria. As at the September meeting, committee members discussed and voted on each of the seven conditions. Members were asked to complete a ballot with check boxes for each of the four criteria. The check boxes were labeled "yes," "no," and "unsure." Members were asked to provide a brief explanation if they voted "unsure" to help others understand the source of uncertainty.

The committee was able to complete reviews of each of the seven additional disorders. A tally of the votes from this meeting and those from the first meeting are included, along with comments received. In summary only three of the seventeen conditions received unanimous "yes" votes for each of the four criteria. These were isovaleric academia (IVA), glutaric academia type 1 (GA-1) which were reviewed in the first meeting and holocarboxylase synthase deficiency/multiple carboxylase deficiency (HCSC/MCD) which was reviewed at this meeting. With a single exception, all of the other conditions received majority "yes" votes for the four criteria. The single condition that did not receive majority votes was 3-methylcrotonyl-CoA carboxylase deficiency (3MCC). The highly variable clinical presentation of this condition was largely judged as too great to meet the prevention potential and medical rational criteria and the public health rationale criteria. There was some discussion and concern about the available technology to screen for tyrosinemia type 1 (TYR-1). However, Department of Health staff reported that the Mayo Clinic in Minnesota recently developed, and is using, a screening test suitable for detecting the condition using the same sample and equipment at the same time the

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other conditions are tested. Based on this information, TYR-1 received 13 'yes' and 1 'no' votes for the available technology criteria.

Following the voting there was discussion of the process and timeline for developing recommendations for the Board of Health. It is estimated that the cost benefit analysis and review by the committee can be completed by the end of March. Recommendations of the committee will then be developed and presented to the Board with a target decision date of early spring so that any expansion of screening can be implemented by the summer of 2008.

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 voting summary from both the September and December meetings, the five criteria and the Advisory Committee membership are attached to this summary.

Conditions considered at September 24 meeting:

- 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 acidemia – Cbl A,B – vitamin B12 responsive (CblA,B)
- Propionic Acidemia (PROP)
- Citrullinemia Type 1 (CIT)
- Argininosuccinic Acidemia (ASA)

Conditions considered at December 10 meeting:

- 3-hydroxy-3-methylglutaric aciduria (HMG)
- Beta-Ketothiolase deficiency (BKD)
- Carnitine Palmitoyl Transferase 1 Deficiency (CPT-1)*
- Holocarboxylase synthase deficiency (HCSH)/Multiple carboxylase deficiency MCD
- Carnitine uptake deficiency (CUD)
- 3-methylcrotonyl-CoA carboxylase deficiency (3-MCC)
- Tyrosinemia type 1 (TYR-1)

Attachments:

Summary of September 24, 2007 Advisory Committee Meeting

Summary of voting from September and December 2007 meetings

Advisory Committee Comments

Five Criteria

Advisory Committee membership

More details can be found at:

http://www.sboh.wa.gov/Goals/HealthyBehaviors/NewbornScreening/NSACMeeting_20070924/index.htm Alternatively, you can contact [Mike Glass](#) (Department of Health) at 206-418-5470 or [Tara Wolff](#) (Washington State Board of Health) at 360 236-4101.