Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

Laboratory Standards & Procedures Subcommittee

Report June 6, 2006

Laboratory Standards & Procedures Subcommittee

- **Duane Alexander**
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Charge of Subcommittee

- **Define and implement a mechanism for the periodic review and assessment of**
  - **the conditions included in the uniform panel**
  - **infrastructure services needed for effective and efficient screening of the conditions included in the uniform panel**
  - **laboratory procedures utilized for effective and efficient testing of the conditions included in the uniform panel**

Uniformity of Newborn Screening

- **17% of babies born in the US receive a routine second screening**
- **Scientific literature indicates that cases of CH and CAH that are missed on the initial screen are detected on the routine second screen**
- **Most newborn screening programs do not support the operation of a routine second screen**
- **To better understand the justification for a routine second screen we are proposing a study to investigate the value of the routine second screen**

Additions to Proposal

- **Include mechanism to capture false positives**
- **Complete second tier testing for CAH positive samples**
- **Include family history on data collection sheets**

Proposed Project Timeline