

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

Laboratory Standards & Procedures Subcommittee

Charter of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

- **“...shall review and report regularly on newborn and childhood screening practices, recommend improvements in the national newborn and childhood screening programs...”**
- **“Provide such recommendations, advice or information as may be necessary to enhance, expand or improve the ability of the Secretary to reduce the mortality or morbidity in newborns and children from heritable disorders.”**
- **“As necessary, standing and ad hoc subcommittees composed of members of the parent committee may be established to perform specific functions within the Committee’s jurisdiction.”**

Laboratory Standards & Procedures Subcommittee

- **Duane Alexander**
- **Amy Brower (chair)**
- **Peter B. Coggins**
- **R. Rodney Howell**
- **Marie Mann (staff)**
- **Piero Rinaldo**

Staff Support – Carrie Diener

Proposed Charge of Subcommittee

- **Define and implement a mechanisms 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**

Periodic Review and Assessment of the Conditions Included in the Uniform Panel

- **Identify the criteria to be used by the committee in recommending inclusion of a new condition in the uniform panel, and define the type of evidence that the subcommittee will utilize in making decisions**
- **Establish ad hoc working groups comprised of people with the necessary expertise to evaluate tests, technologies and benefits to the individual and society**
- **Complete timely reviews**
- **Provide assessment(s) to the Advisory Committee for consideration**

Infrastructure Services Needed for Effective and Efficient Screening of the Conditions Included in the Uniform Panel

- **Examples: Sample collection, sample transport, result reporting to physicians and families, and referral for confirmation of diagnosis and counseling**
- **Establish a working group comprised of people with the necessary expertise to evaluate infrastructure services**
- **Complete timely reviews**
- **Provide assessment(s) to the Advisory Committee for consideration**

Laboratory Procedures Utilized for Effective and Efficient Testing of the Conditions Included in the Uniform Panel

- **Establish a working group comprised of people with the necessary expertise to evaluate laboratory procedures, including nomenclature, testing strategies, cut-off values, reporting requirements, performance metrics, and case definitions.**
- **Complete timely reviews**
- **Provide assessment(s) to the Advisory Committee for consideration.**