

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

Ronald Reagan Building and International Trade Center
Washington, DC
February 13-14, 2006

MONDAY, FEBRUARY 13

- 8:30 a.m. – 9:00 a.m. **CONTINENTAL BREAKFAST** (for Committee and Ex-Officio Members)
- 9:00 a.m. – 9:15 a.m. **OPENING REMARKS AND WELCOME**
- R. Rodney Howell, M.D.
Committee Chair
- Introduction of New Committee Members
James A. Newton, M.D.
Mary Jane Owen, M.S.W.
- Introduction of New Organization Representatives
Association of State and Territorial Health Officials
Christopher A. Kus, MD, MPH
Food and Drug Administration
Ethan Hausman, M.D.
Child Neurology Society
Bennett Lavenstein, M.D.
- 9:15 a.m. – 10:30 a.m. **SUBCOMMITTEE REPORTS**
Update on work. Presentation to include established scope of work and activity elements subcommittee is considering, with projected timelines for completion (20 minutes per subcommittee with 15 minutes of Committee discussion)
- 10:30 a.m. – 10:45 a.m. Break
- 10:45 a.m. – 11:45 a.m. **NATIONAL COORDINATION CENTER FOR THE REGIONAL GENETICS AND NEWBORN SCREENING COLLABORATIVES**
- Michael S. Watson, PhD, FACMG
Executive Director
American College of Medical Genetics
- 11:45 a.m. – 1:00 p.m. **WORKING LUNCH** (for Committee Members)

1:00 p.m. – 2:30 p.m.

**REGIONAL GENETIC SERVICE AND NEWBORN SCREENING
COLLABORATIVES**

- **Epidemiology: Mapping Genetic Needs Relative to Services for Cystic Fibrosis, Hemoglobinopathies and Metabolic Disorders.**

Anne Marie Comeau, PhD
New England Regional Collaborative

- **Performance Metrics and Harmonization of Cut-Off Values for Newborn Screening by Tandem Mass Spectrometry**

Piero Rinaldo, M.D., Ph.D.
Great Lakes Region 4

- **Preparing for Disasters: Genetic/Metabolic Health Care Delivery During and after Hurricanes Katrina and Rita**

Jess Thoene, M.D.
Southeast Regional Genetics Group

2:30 p.m. – 3:00 p.m.

**NIH OFFICE OF RARE DISEASES – RARE DISEASE CENTERS OF
EXCELLENCE**

Stephen Groft, PharmD
Director
Office of Rare Diseases

3:00 p.m. – 3:15 p.m.

BREAK

3:15 p.m. – 5:00 p.m.

SUBCOMMITTEE MEETINGS

5:00 p.m.

ADJOURN

TUESDAY, FEBRUARY 14

- 8:00 a.m. – 8:30 a.m. **CONTINENTAL BREAKFAST** (for Committee and Ex-Officio Members)
- 8:30 a.m. – 10:00 a.m. **NOMINATION PROCESS FOR CANDIDATE CONDITIONS ON THE UNIFORM SCREENING PANEL**
- Framework for the Overall Nomination Process
Criteria Work Group Report Criteria for Nomination Form

Nancy Green, M.D.
Medical Director
March of Dimes
 - A Trial Run of Conditions Using the Proposed Process

Piero Rinaldo, M.D., Ph.D.
ACHDGDNC
- 10:00 a.m. – 10:45 a.m. **SUBCOMMITTEE REPORTS AND DISCUSSION**
- 10:45 a.m. – 11:00 a.m. **LYSOSOMAL STORAGE DISEASES WORKSHOP**
- Robert Vogt, Ph.D.
Centers for Disease Control and Prevention
- 11:00 a.m. – 11:15 a.m. **BREAK**
- 11:15 a.m. – 11:45 a.m. **ORGANIZATIONS REPRESENTING POLICY MAKERS AND LEGISLATORS**
- National Conference of State Legislators
Alissa Johnson, M.A.
 - Association of State and Territorial Health Officials
Christopher A. Kus, M.D., M.P.H.
- 11:45 a.m. – 1:00 p.m. **WORKING LUNCH** (for Committee and Ex-Officio Members)
- 1:00 p.m. – 2:00 p.m. **PUBLIC COMMENTS**
- 2:00 p.m. – 3:00 p.m. **COMMITTEE BUSINESS**
- R. Rodney Howell, M.D.
Committee Chair
- 3:00 p.m. **ADJOURN**