

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

*Ronald Reagan Building and International Trade Center
Washington, DC
May 17-18, 2007*

THURSDAY, MAY 17, 2007

8:30 a.m. – 9:00 a.m. **CONTINENTAL BREAKFAST** (Committee Members and Representatives)

9:00 a.m. – 9:15 a.m.
Rotunda Room **OPENING REMARKS**

- **REVIEW OF COMMITTEE CORRESPONDENCE**
- **APPROVAL OF MINUTES FROM JUNE 2006 MEETING**

R. Rodney Howell, M.D., Committee Chair

9:15 a.m. – 9:45 a.m. **NOMINATION/EVALUATION PROCESS FOR CANDIDATE CONDITIONS ON THE UNIFORM SCREENING PANEL**

James Perrin, M.D.

Professor of Pediatrics, Harvard Medical School Director,
Division of General Pediatrics
Director, Center for Child and Adolescent Health Policy
Harvard Medical School
MassGeneral Hospital for Children

9:45 a.m. – 10:15 a.m. **REPORT FROM THE APRIL 2007 WORKGROUP MEETING:
*THE ROAD MAP TO IMPLEMENT LONG-TERM FOLLOW-UP AND
TREATMENT IN NEWBORN SCREENING***

Coleen Boyle, Ph.D., M.S., Committee Member

Stephen M. Downs, M.D., M.S.

Associate Professor and Director
Children's Health Services Research
Indiana University School of Medicine

Alex Kemper, M.D., M.P.H., M.S.

Associate Professor
Department of Pediatrics
Duke Children's Hospital and Health Center
Duke University

10:15 a.m. – 10:30 a.m. DISCUSSION

10:30 A.M. – 10:45 A.M. BREAK

**10:45 A.M. – 11:30 A.M. REGIONAL GENETIC AND NEWBORN SCREENING
SERVICES COLLABORATIVES:**

LONG-TERM FOLLOW-UP PROJECTS

Stephen M. Downs, M.D., M.S.

Associate Professor and Director
Children's Health Services Research
Indiana University School of Medicine

James Eckman, M.D.

Professor
Department of Hematology and Oncology
Winship Cancer Institute
Emory University School of Medicine, Atlanta, GA

11:30 A.M. – 11:45 A.M. DISCUSSION

11:45 A.M. – 1:00 P.M. WORKING LUNCH

1:00 P.M. – 1:30 P.M.

STATUS OF THE STATES-AN UPDATE

- **GLUCOSE-6-PHOSPHATE DEHYDROGENASE (G6PD) DEFICIENCY**
- **SEVERE COMBINED IMMUNODEFICIENCY**

Bradford Therrell, Ph.D.

Director
National Newborn Screening and Genetics Resource Center

1:30 P.M. – 1:45 P.M.

DEPARTMENT OF DEFENSE NEWBORN SCREENING PROGRAM

Lt. Col. David S. Louder, III, M.D.

Chief Consultant for Maternal-Child Medicine
Air Force Medical Corps
AFMSA/SGOC

1:45 P.M. – 2:00 P.M.

BREAK

2:00 P.M. – 5:00 P.M.

SUBCOMMITTEE MEETINGS

- **SUBCOMMITTEE ON LABORATORY STANDARDS AND PROCEDURES**

INTERNATIONAL GATEWAY ROOM

- **SUBCOMMITTEE ON EDUCATION AND TRAINING**

ROTUNDA COAT ROOM

5:00 P.M.

ADJOURN

FRIDAY, MAY 18

8:00 A.M. – 8:30 A.M. **CONTINENTAL BREAKFAST** (Committee Members and Representatives)

8:30 A.M. – 10:00 A.M. **SUBCOMMITTEE REPORTS AND DISCUSSION**

- **SUBCOMMITTEE ON FOLLOW-UP AND TREATMENT**

Coleen Boyle, Ph.D., M.S.

- **SUBCOMMITTEE ON LABORATORY STANDARDS AND PROCEDURES**

Amy Brower, Ph.D.

- **SUBCOMMITTEE ON EDUCATION AND TRAINING**

Gregory A. Hawkins, Ph.D.

10:00 A.M. – 10:15 A.M. **STANDARD OPERATING PROCEDURES FOR ACHDGDNC**

10:15 A.M. – 10:45 A.M. **BREAK**

10:45 A.M. – 11:30 A.M. **REGIONAL LABORATORY PERFORMANCE**

Piero Rinaldo, M.D., Ph.D.

T. Denny Sanford Professor of Pediatrics
Professor of Laboratory Medicine
Vice-Chair of Academic Affairs and Intramural Practice,
Department of Laboratory Medicine and Pathology
Mayo Clinic College of Medicine, Rochester, MN

Marzia Pasquali, Ph.D.

Associate Professor of Pathology (Clinical)
University of Utah School of Medicine
Medical Director, Biochemical Genetics and
Supplemental Newborn Screening

11:30 A.M. – 11:45 A.M. FEDERAL LEGISLATION: AN UPDATE

Marina Weiss, Ph.D.

Senior Vice President
Public Policy and Government Affairs
March of Dimes Birth Defects Foundation

11:45 A.M. – 1:00 P.M. WORKING LUNCH

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

- **AGENDA ITEMS FOR SEPTEMBER 2007 MEETING**
- **CALENDAR FOR 2007/2008 MEETINGS**

3:00 P.M. ADJOURN