

# **ADVISORY COMMITTEE ON HERITABLE DISORDERS IN NEWBORNS AND CHILDREN**

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**BETHESDA MARRIOTT HOTEL-POOKS HILL  
BETHESDA, MARYLAND  
SEPTEMBER 24-25, 2009**

## **THURSDAY, SEPTEMBER 24, 2009**

**7:30 A.M. – 8:30 A.M. CONTINENTAL BREAKFAST (Committee Members and Presenters)**

**8:30 A.M. – 8:45 A.M. WELCOME**

- Approval of Minutes from May 12, 2009 Meeting
- Committee Correspondence

**R. RODNEY HOWELL, M.D.**  
Committee Chair

**8:45 A.M. – 11:30 A.M. SUBCOMMITTEE MEETINGS**

- **Follow-up and Treatment**
- **Education and Training**
- **Laboratory Standards**

**11:30 A.M. – 11:45 A.M. BREAK**

**11:45 A.M. – 12:20 P.M. SUBCOMMITTEE REPORTS AND DISCUSSION**

- **Subcommittee on Laboratory Standards and Procedures**  
**Gerard Vockley, M.D., PH.D.**  
Committee Member
- **Subcommittee on Education and Training**  
**Jana Monaco and Tracy L. Trotter, M.D.**  
Committee Member
- **Subcommittee on Follow-up and Treatment**  
**Coleen Boyle, PH.D., M.S.**  
Committee Member

**12:20 P.M. – 12:30 P.M. MEDICAL FOODS LEGISLATION**

**Christine Brown, MS**  
Executive Director National PKU Alliance

**12:30 P.M. – 1:15 P.M. LUNCH (Committee Members and Presenters)**

<b>1:15 P.M. – 2:00 P.M.</b>	<b>THE NATIONWIDE HEALTH INFORMATION NETWORK</b>  <b>Charles Friedman, M.D.</b> Deputy Director for the Office of the National Coordinator for Health Information Technology
<b>2:00 P.M. – 2:15 P.M.</b>	<b>STATUS OF THE IMPLEMENTATION OF THE NEWBORN SCREENING USE CASE</b>  <b>Alan Zuckerman, M.D.</b> Georgetown University
<b>2:15 P.M. – 2:45 P.M.</b>	<b>NEWBORN SCREENING CODES &amp; TERMINOLOGY AND AN APPROACH TO A STANDARD REPORT PAYLOAD</b>  <b>Kin Wah Fung, M.D.</b> National Library of Medicine
<b>2:45 P.M. – 3:00 P.M.</b>	<b>NEWBORN SCREENING WEB PORTAL CONCEPT</b>  <b>Gregory Downing D.O., Ph.D. and Constanze Coon, Ph.D.</b> Personalized Health Care Initiative, OS/HHS
<b>3:00 P.M. – 3:15 P.M.</b>	<b>BREAK</b>
<b>3:15 P.M. – 4:00 P.M.</b>	<b>MEASURES FOR QUALITY</b>  <b>Sarah Scholle, DR.PH.</b> National Center for Quality Assurance
<b>4:00 P.M. – 4:30 P.M.</b>	<b>INTERNAL REVIEW WORKGROUP: NOMINATION OF ALPHA THALASSEMIA: HEMOGLOBIN H DISEASE TO THE ACHDNC’S RECOMMENDED UNIFORM SCREENING PANEL</b>  <b>Kwaku Ohene-Frempong, M.D.</b> Committee Member
<b>4:30 P.M. – 5:00 P.M.</b>	<b>COMMITTEE DISCUSSION</b>
<b>5:00 P.M.</b>	Adjourn

FRIDAY, SEPTEMBER 25, 2009

**7:30 A.M. – 8:30 A.M.**            **CONTINENTAL BREAKFAST (COMMITTEE MEMBERS AND PRESENTERS)**

**8:30 A.M. – 9:00 A.M.**            **USE AND STORAGE OF RESIDUAL BLOOD SPOTS WORKGROUP:  
BRIEFING PAPER REPORT**

**Jana Monaco**  
Committee Member

**Brad Therrell, Ph.D.**  
**Harry Hannon, Ph.D.**  
National Newborn Screening and Genetic Resource Center

**9:00 A.M. – 9:30 A.M.**            **COMMITTEE DISCUSSION: BRIEFING PAPER**

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

**9:30 A.M. – 10:00 A.M.**        **PUBLIC COMMENTS**

**10 A.M. – 10:30 A.M.**            **BREAK**

**10:30 A.M. – 11:30 A.M.**        **EVIDENCE REVIEW WORKGROUP: FINAL DRAFT REPORT ON THE  
CANDIDATE NOMINATION OF KRABBE DISEASE**

**James M. Perrin, M.D.**  
Evidence Review Workgroup Chairperson

**11:30 A.M. – 12:00 P.M.**        **PUBLIC COMMENTS**

**12:00 P.M. – 1:00 P.M.**        **LUNCH (Committee Members and Presenters)**

**1:00 P.M. – 2:00 P.M.**            **COMMITTEE DISCUSSION ON THE CANDIDATE NOMINATION OF  
KRABBE DISEASE**

**Piero Rinaldo, M.D., Ph.D.**  
Committee Member

**2:00 P.M. – 2:30 P.M.**            **COMMITTEE REPORT FOR CONGRESS**

**Alaina M. Harris, M.S.W., M.P.H.**  
Health Resources and Services Administration

**POLICY PAPER: NEWBORN SCREENING AND HEALTH CARE REFORM**

**Alissa Johnson, M.A.**  
Johnson Policy Consulting

**2:30 P.M. – 3:00 P.M.**            **COMMITTEE BUSINESS**

- Calendar for 2010 Committee Meetings
- Agenda Items For January 2010 Meeting

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

**3:00 P.M.**

**ADJOURN**