Workshop Summary

Issues in Presymptomatic Diagnoses of Lysosomal Storage Diseases

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Newborn Screening Quality Assurance Program

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Issues in Presymptomatic Diagnoses of Lysosomal Storage Diseases

December 6 – 7, 2005

Marriott Bethesda North Hotel and Conference Center

Hosted by: National Newborn Screening and Genetics Resource Center – Austin, Texas

In collaboration with: Health Resources and Services Administration
Centers for Disease Control and Prevention
National Institutes of Health
American College of Medical Genetics
Genzyme

December 6, 2005

8:00 AM ***Welcome*** — Brad Therrell, Ph.D. (NNSGRC)

8:05 AM ***Meeting Goals*** — Harry Hannon, Ph.D. (CDC)

Background and Issues

Moderator: Celia Kaye, M.D., Ph.D. (NNSGRC)

8:15 AM ***Newborn Screening for Lysosomal Storage Disorders (Tab C)***

- Overview and Status — Bob Vogt, Ph.D. (CDC)
- Current Research into NBS Protocols — Joan Keutzer, Ph.D. (Genzyme)
- State Newborn Screening Program Activities — Ken Pass, Ph.D. (NY DPH)

9:00 AM ***Introduction to Lysosomal Storage Disorders (Tab D)*** — Paul Fernhoff, M.D.
Emory University Medical School

10:00 AM	***The Challenges of Newborn Screening for Mucopolysaccharidosis (Tab E)***	Joseph Muenzer, M.D., Ph.D. UNC Chapel Hill
10:40 AM	***Clinical Overview and Update of Clinical Trials of Myozyme for Pompe Disease. The Role of Newborn Screening? (Tab F)***	Priya Kishnani, M.D. Duke UMC
11:20 AM	***Umbilical Cord Blood Transplantation for the Treatment of Infantile Krabbe Disease (Tab G)***	Maria Luisa Escolar, M.D. UNC Chapel Hill
1:15 PM	***Fabry disease –Is it Time for Newborn Screening? (Tab H)***	Olaf Bodamer, M.D., Ph.D. University Children's Hospital, Vienna
1:55 PM	***Gaucher disease: Impact of Genotype/Phenotype Correlations on Newborn Screening and Treatment (Tab I)***	Gregory A. Grabowski, M.D. Cincinnati CHMC
2:35 PM	***Newborn Screening for Acid Sphingomyelinase Deficiency: Niemann Pick A/B (Tab J)***	Gerald Cox, M.D., Ph.D. Genzyme
3:30 PM	***Neonatal Screening for X-Linked Adreno-leukodystrophy: Rationale and Strategy (Tab K)***	Hugo W. Moser, M.D. Ann B. Moser, B.A. Kennedy Krieger Institute
4:10 PM	***Summary***	Edward Kaye, M.D. (Genzyme)
4:30 PM	***Public Comments***	
5:00 PM	***Final Comments***	Celia Kaye, M.D. (NNSGRC)

Issues in Disease Follow-up

Moderator: Rodney Howell, M.D. (SACHDGDNC)

8:15 AM ***Research and Clinical Brief Reports***

Theoretical Benefits for Perinatal Treatment of MPS1 (Tab L)	Deborah S. Barbouth, M.D. University of Miami School of Medicine
Umbilical Cord Blood Galactocerebroside Levels for Choosing Appropriate Cord Bloods for Transplantation (Tab M)	William Krivit, M.D., Ph.D. University of Minnesota
Correction of a biomarker for Pompe disease by AAV vector-mediated gene therapy (Tab Mc)	Dwight D. Koeberl, M.D., Ph.D. Duke University Medical Center
Transplant Outcomes for Boys with Cerebral X-ALD Diagnosed by Family History vs. by Signs and Symptoms (Tab N)	Charles Peters, M.D. Children's Mercy Hospital Kansas City, Missouri
Intracranial Tumors in Patients with Mucopolysaccharidosis I: New Case and Literature Review Suggest Pathogenic Mechanisms and Need for Surveillance (Tab O)	Virginia Proud, M.D. Children's Hospital of The King's Daughters, Norfolk, Virginia
Pathogenesis of GM1 and GM2 Gangliosidoses and Avenues for Therapy Directed to the CNS (Tab P)	Cynthia Tifft, M.D., Ph.D. Children's National Medical Center Washington, D.C.
The Rationale, Methods, and Challenges of Presymptomatic Diagnosis for Mucopolysaccharidoses and Implications for Other Lysosomal Disorders (Tab Q)	Chester Whitley, M.D., Ph.D. University of Minnesota Medical School

9:30 AM	***Practical Issues Regarding NBS Expansion (Tab R)*** • ***Local issues*** • ***Standardization issues***	Deborah Marsden, M.D. Children's Hospital Boston
10:30 AM	***Confirmatory Testing Following a Positive Newborn Screen (Tab S)***	David Wenger, Ph.D. Thomas Jefferson University
11:00 AM	***Registries and Newborn Screening: What Can We Learn From Them? (Tab T)***	Edward Kaye, M.D. Genzyme
11:30 AM	***Storing Specimens for Translation Research (Tab U)***	Bob Vogt, Ph.D. CDC
11:45 AM	***Role of the Federal Agencies:***	
	Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (Tab V)	Michele Lloyd-Puryear, M.D., Ph.D. HRSA
	NIH Research Initiatives in Newborn Screening for Rare Disorders (Tab W)	James W. Hanson, M.D. NIH/NICHD
12:00 PM	***Lunch/Discussion***	
1:00 PM	***Continued Discussion – Items for Inclusion in MMWR Reports and Recommendations***	Bob Vogt, Ph.D. CDC
2:30 PM	***Final Comments***	Rod Howell, M.D. Celia Kaye, M.D., Ph.D.
3:00 PM	***Adjourn***	

Workshop Follow-Up Activities

Issues in Presymptomatic Diagnoses of Lysosomal Storage Diseases

MMWR Report and Recommendations
(Publication Scheduled Dec 2006 - Jan 2007)

Technical Issues Workshop
(Anticipated late 2006 - early 2007)

NSTRI Laboratory Activities
(Commencing February 2006)

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