

Early Detection and Screening for Neuromuscular Diseases

March 16-17, 2009

Agenda

Monday, March 16, 2009

8:30 -8:50 am	Introductions Duane Alexander, M.D.
8:50-9:20 am	Components of an Early Detection System Piero Rinaldo, M.D.
9:20-9:40 am	Traditional Qualifications for Newborn Screening Bradford Therrell, Ph.D.
9:40-9:55 am	Advisory Committee for Heritable Disorders and Genetics Diseases in Newborns and Children Evidence Review Workgroup: Nomination Process R. Rodney Howell, M.D.
9:55-10:10 am	Pompe Disease: A Journey Through the Nomination Process James Perrin, M.D.
10:10-10:40 am	Break
10:40 am-12:25 pm	Overview and Status of Neuromuscular Disorders and Their Screening Potential
10:40 am-10:55 am	Myotonic Dystrophy Richard (Dick) Moxley III, M.D.
10:55 am-11:10 am	Facioscapulohumeral Muscular Dystrophy Katherine Mathews, M.D.
11:10 am-11:25 am	Duchenne/Becker Muscular Dystrophy Eric Hoffman, Ph.D.
11:25 am-11:40 am	Spinal Muscular Atrophy Thomas Prior, Ph.D.
11:40 am-11:55 am	Mitochondrial Disorders Salvatore DiMauro, M.D.
11:55 am-12:10 pm	Congenital Muscular Dystrophies Anne Rutkowski, M.D.
12:10 pm-12:25 pm	Friedreich's Ataxia

Devin Oglesbee, Ph.D.

12:25pm -1:30 pm

Lunch

1:30 pm-1:50 pm

Universal vs. Voluntary Newborn Screening: The Wales Experience

Evelyn Parsons, Ph.D.

Newborn vs. Early Childhood Screen: Centers for Disease Control and Prevention (CDC)

1:50 pm–2:10 pm

Pilot Screening for DMD

Jerry R. Mendell, M.D.

2:10 pm–2:30 pm

Infant Screening and Diagnosis for DMD

Paul M. Fernhoff, M.D.

2:30 pm-3:00 pm

Panel Discussion

Panel Leader: Alan Fleischman, M.D.

Ben Wilfond, Ph.D.

Lainie Friedman Ross, M.D., Ph.D.

Ben Cumbo

3:00 pm-3:30 pm

Poster Session

3:30 pm-3:50 pm

Current Newborn Screening Environment: Capabilities and Limitations Regarding Piloting and Expansion

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

3:50 pm-4:30 pm

Professional Panel: Changing Perspectives on the Benefits of Early Detection and Newborn Screening

Don Bailey, Ph.D.

Scott Grosse, Ph.D.

Bruce Nedrow (Ned) Calonge, M.D., M.P.H.

4:30 pm-5:10 pm

Parent/Patient Panel: Changing Perspectives on the Benefits of Early Detection and Newborn Screening

Sharon Terry, M.A.

Patricia Furlong

Deborah Heine

Janice Bonner

Tuesday, March 17, 2009: The Future of Screening

8:30 am–8:50 am

Microarray Chip Technology

Madhuri Hegde, Ph.D.

8:50 am–9:10 am

Sequencing

Jeffrey Schloss, Ph.D.

Available Research Networks and Funding Opportunities: Agencies, Institutes/Centers, and

- 9:10 am-9:20 am **Muscular Dystrophy Association**
Valerie Cwik, M.D.
- 9:20 am-9:30 am **TreatNMD**
John Porter, Ph.D.
- 9:30 am-9:40 am **National Institutes of Health**
John Porter, Ph.D.
- 9:40 am-9:50 am **Centers for Disease Control and Prevention**
Edwin Trevathan, M.D., M.P.H.
- 9:50 am-10:00 am **Health Resources and Services Administration**
Michele Puryear, M.D., Ph.D.
- 10:00 am-10:10 am **Agency for Healthcare Research and Quality**
Gurvaneet Randhawa, M.D., M.P.H.
- 10:10 am-10:20 am **Discussion**
- 10:20 am-10:50 am **Poster Session**
- 10:50 am-11:30 am **Recommendations for Research and Public Health Infrastructure Development**
(5 Breakout Groups)
- Priorities for Needed Research: Development of Therapies and Screening/Diagnostic Technologies**
Louis Kunkel, Ph.D. (lead)
Daniel Perez
- Priorities for Needed Epidemiological and Clinical Studies: Natural History, Incidence, Surveillance, etc.**
Paul Romitti, Ph.D. (lead)
- Priorities for Needed Health Services and Health Service Outcome and Utilization Research**
Gurvaneet Randhawa, M.D., M.P.H. (lead)
- Priorities for Developing Provider, Patient, and Public Education and Addressing Ethical, Legal, and Social Issues (ELSI) Topics**
Sharon Terry, M.A. (lead)
Terry Davis, Ph.D.
- Priorities for Creating Systems of Care from Infancy to Adulthood**
Bonnie Strickland, Ph.D. (lead)
Tracey Trotter, M.D.
Valerie Cwik, M.D.
- 11:30 am-12:00 pm **Break**
- 12:00 pm-12:50 pm **Summary Presentations from Breakout Groups & Discussion**

Louis Kunkel, Ph.D.
Paul Romitti, Ph.D.
Gurvaneet Randhawa, M.D., M.P.H.
Sharon Terry, M.A.
Bonnie Strickland, Ph.D.

12:50 pm-1:10 pm

Closing Remarks

R. Rodney Howell, M.D.