**Early Detection and Screening for Neuromuscular Diseases**

**March 16-17, 2009**

**Agenda**

**Monday, March 16, 2009**

<table>
<thead>
<tr>
<th>Time</th>
<th>Activity</th>
<th>Speaker</th>
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<tr>
<td>8:30 - 8:50 am</td>
<td>Introductions</td>
<td>Duane Alexander, M.D.</td>
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<td>8:50 - 9:20 am</td>
<td>Components of an Early Detection System</td>
<td>Piero Rinaldo, M.D.</td>
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<td>9:20 - 9:40 am</td>
<td>Traditional Qualifications for Newborn Screening</td>
<td>Bradford Therrell, Ph.D.</td>
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<td>9:55 - 10:10 am</td>
<td>Pompe Disease: A Journey Through the Nomination Process</td>
<td>James Perrin, M.D.</td>
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<td>10:10 - 10:40 am</td>
<td>Break</td>
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<tr>
<td>10:40 - 12:25 pm</td>
<td>Overview and Status of Neuromuscular Disorders and Their Screening Potential</td>
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<td>10:40 - 10:55 am</td>
<td>Myotonic Dystrophy</td>
<td>Richard (Dick) Moxley III, M.D.</td>
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<td>10:55 - 11:10 am</td>
<td>Facioscapulohumeral Muscular Dystrophy</td>
<td>Katherine Mathews, M.D.</td>
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<td>11:10 - 11:25 am</td>
<td>Duchenne/Becker Muscular Dystrophy</td>
<td>Eric Hoffman, Ph.D.</td>
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<td>11:25 - 11:40 am</td>
<td>Spinal Muscular Atrophy</td>
<td>Thomas Prior, Ph.D.</td>
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<td>11:40 - 11:55 am</td>
<td>Mitochondrial Disorders</td>
<td>Salvatore DiMauro, M.D.</td>
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<td>11:55 - 12:10 pm</td>
<td>Congenital Muscular Dystrophies</td>
<td>Anne Rutkowski, M.D.</td>
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<td>12:10 pm - 12:25 pm</td>
<td>Friedreich's Ataxia</td>
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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
12:50 pm-1:10 pm  
**Closing Remarks**  
R. Rodney Howell, M.D.