



## ADVANCING RARE DISEASE RESEARCH:

### The Intersection of Patient Registries, Biospecimen Repositories, and Clinical Data

January 11–12, 2010 • Doubletree Hotel & Executive Meeting Center • Bethesda, MD

# DAY 1 — MONDAY, JANUARY 11

**Workshop Objectives:** *To discuss the development of an infrastructure for an Internet-based platform with common data elements utilizing a federated rare disease registry able to incorporate:*

1. Existing rare disease registries and any other useful patient registries
2. Patient organizations with no registry looking to establish one
3. Patients with no affiliation with a support group looking to belong to a registry

**Expected Outcomes:** *To gain acceptance of the concept of a federated rare disease patient registry by as many curators of patient registries and other stakeholders as possible and to gain their participation in creating this patient registry. Participating stakeholders will agree on a strategy to harmonize standardized common data elements, vocabulary, and open source software to enable the exchange of data and information to facilitate research collaborations.*

**7:00 A.M. Registration and Continental Breakfast**

**7:50 A.M. Welcome**

*Yaffa Rubinstein, Ph.D.* — Office of Rare Diseases Research (ORDR), National Institutes of Health (NIH) and *Geraldine Pollen, M.A.* — ORDR, NIH

**Order of the Day**

**Moderators:** *Ronald J. Bartek* — Friedreich's Ataxia Research Alliance (FARA) and *Jennifer Farmer, M.S., C.G.C.* — FARA

## PLENARY SESSIONS

### A. Introduction to Objectives for the Collaborative Rare Diseases Registry (CRDR)

**8:00 A.M. Developing the Rare Diseases Registry**

*Stephen C. Groft, Pharm.D.* — ORDR, NIH

**8:15 A.M. Patient and Research Advocate Statement**

*Amy Farber, Ph.D.* — LAM Treatment Alliance

**8:35 A.M. Uniting Rare Diseases**

*Vanessa Rangel Miller, M.S., C.G.C.* — DuchenneConnect



## **B. Alternatives and Future Promises for a National Rare Disease Patient Registry**

- 8:50 A.M. Structure and Function of a Collaborative Rare Disease Patient Registry**  
*Christopher B. Forrest, M.D., Ph.D. — University of Pennsylvania School of Medicine and Children’s Hospital of Philadelphia*
- 9:10 A.M. The National Health Information Network and its Implications for a National Rare Disease Patient Registry/Case study**  
*Daniel C. Russler, M.D. — Oracle Health Sciences Strategy*
- 9:30 A.M. Question and Answer Session**  
**Chair:** *Christopher B. Forrest, M.D., Ph.D.*
- 9:45 A.M. BREAK**

## **PRESENTATIONS**

### **I. Standards, Informatics, and Technology**

- 9:55 A.M. Support for Compatibility and Interoperability**  
*Kyle Brown — Innolyst, Inc.*
- 10:15 A.M. Reuse of Clinical Health Records: Caveat Inquisitor**  
*James J. Cimino, M.D. — Laboratory for Informatics Development, Clinical Center (CC), NIH*
- 10:30 A.M. Pros and Cons of Various Models and Communication Across the Different Models**  
*Clement J. McDonald, M.D. — National Library of Medicine (NLM), NIH*
- 10:50 A.M. Plans for Data Standards in Rare Disease Registries**  
*Rachel Richesson, Ph.D., M.P.H. — University of South Florida (USF) College of Medicine*
- 11:00 A.M. Global Data Aggregation: Case Study/Treat-NMD**  
*Christophe Beroud, Pharm.D., Ph.D. — France, INSERM*
- 11:20 A.M. Question and Answer Session**  
**Chair:** *Kyle Brown*

### **II. Biospecimens/Biorepositories**

- 11:35 A.M. Challenges and Obstacles Obtaining Rare Disease Specimens and the Use of Registries**  
*Christopher A. Moskaluk, M.D., Ph.D. — UVA Biorepository*

- 11:45 A.M. Rare Disease Biospecimens: Quality and Accessibility Challenges**  
*Carolyn C. Compton, M.D., Ph.D. — Office of Biorepositories and Biospecimen Research (OBBR), National Cancer Institute (NCI)*
- 12:05 P.M. Rare Disease Biorepositories and Registries: The Need for Collaborative and Novel Approaches**  
*Benjamin M. Greenberg, M.D., M.H.S. — University of Texas Southwestern*
- 12:20 P.M. The Use of Patient Registries to Increase Procurements of Rare Diseases Biospecimens**  
*Jeffrey A. Thomas — National Disease Research Interchange (NRDI)*
- 12:30 P.M. Investigator Experience: How Research in Rare Diseases Contributes to Understanding the Pathogenesis of Common Diseases**  
*Marsha A. Moses, Ph.D. — Children's Hospital Boston and Harvard Medical School*
- 12:45 P.M. Question and Answer Session**  
**Chairs:** *Jim B. Vaught, Ph.D. and Helen M. Moore, Ph.D.*
- 1:00 P.M. WORKING LUNCH — Keynote Speaker: Advancing Rare Disease Research: Ethical Dimensions**  
*Jonathan D. Moreno, Ph.D. — David and Lyn Silfen University Professor of Ethics and Professor of Medical Ethics and the History and Sociology of Science at the University of Pennsylvania*

### **III. Clinical Research, Patient Care, and Disease Management**

- 2:00 P.M. Role of Rare Disease Registries in Clinical Research**  
*Ronald A. Christensen, M.D. — REGISTRAT-MAPI*
- 2:20 P.M. Regulatory and Other Governmental Influences on Clinical Research**  
*Theresa Toigo, R.Ph., M.B.A. — Office of Special Health Issues, Food and Drug Administration (FDA)*
- 2:35 P.M. Patient Registries and their Role in Understanding Health Outcomes**  
*Jean R. Slutsky, P.A., M.S.P.H. — Center for Outcomes and Evidence, Agency for Healthcare Research and Quality (AHRQ)*
- 2:50 P.M. Data and Test Result Validation: Reporting Research Data and Clinical Test Results to Patients (Researcher and Patient Perspectives)**  
*Andrew Faucett, M.S., C.G.C. — Emory University School of Medicine and Collaboration, Education, and Test Translation (CETT) Program*
- 3:00 P.M. Question and Answer Session**  
**Chair:** *Ronald A. Christensen, M.D.*

#### **IV. Patient Participation and Outreach Activities/Patient Advocacy**

- 3:15 P.M. Patient Advocacy Groups and Patient Registries: An Overview**  
*Sukirti N. Bagal, M.D.* — National Organization for Rare Disorders (NORD)
- 3:25 P.M. The Role of Patient Advocacy Groups in Establishing Common Infrastructure**  
*Sharon F. Terry, M.A.* — Genetic Alliance
- 3:35 P.M. Essential Elements for Translational Research in Rare Diseases: Progeria as a Case Study**  
*Leslie B. Gordon, M.D., Ph.D.* — Progeria Research Foundation
- 3:45 P.M. Common Diseases versus Rare Diseases: Is There Really a Difference?**  
*Susan M. Love, M.D.* — Dr. Susan Love Research Foundation
- 3:55 P.M. Participation of Patients with no Advocacy Group**  
*David S. Goldstein, M.D., Ph.D.* — Clinical Neurocardiology Section, National Institute of Neurological Disorders and Stroke (NINDS), NIH
- 4:05 P.M. Question and Answer Session**  
**Chair:** *Amy Farber, Ph.D.*
- 4:20 P.M. BREAK**

#### **V. Human Subjects: Bioethical and Legal Issues for Clinical Studies**

- 4:35 P.M. Human Subjects: Ethical and Legal Issues/45 CFR 46**  
*Julie Kaneshiro, M.A.* — Office for Human Research Protections (OHRP)
- 4:50 P.M. Ethical and Legal Issues/Government Regulations**  
*P. Pearl O'Rourke, M.D.* — Partners Healthcare
- 5:05 P.M. Legal/Bioethical Issues in Medical Research and Release of Genetic Information**  
*Jack Schwartz, J.D.* — University of Maryland School of Law
- 5:20 P.M. Question and Answer Session**  
**Chairs:** *Sara C. Hull, Ph.D.* and *Jack Schwartz, J. D.*
- 5:35 P.M. Closing Remarks—Day 1**  
*Stephen C. Groft, Pharm.D.* — ORDR, NIH
- 6:00 P.M. Reception for all participants, provided by the generosity of our co-sponsors**



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# DAY 2 — TUESDAY, JANUARY 12

- 7:00 A.M.**    **REGISTRATION AND CONTINENTAL BREAKFAST**
- 8:00 A.M.**    **Keynote Speaker: The Contribution of Large Health Care Systems to Improving Treatment for Patients with Rare Diseases**  
*Joe V. Selby, M.D., M.P.H.* — Division of Research, Kaiser Permanente
- 8:45 A.M.**    **BREAK**

## BREAKOUT SESSIONS

- 9:00 A.M.**    **Morning Parallel Sessions Introduction and Instructions**  
**Moderator:** *Helen M. Moore, Ph.D.* — Office of Biospecimen and Biorepositories Research, NCI
- A. Standardized Vocabulary, Terminology, CDE's, and Diagnosis**  
**Chairs:** *Rachel Richesson, Ph.D., M.P.H.* and *Clement J. McDonald, M.D.*  
**Note taker:** *Kyle Brown*
- Discussion Panel:**  
*Clement J. McDonald, M.D.* — NLM, NIH  
*Rachel Richesson, Ph.D., M.P.H.* — USF College of Medicine  
*Stuart Nelson, M.D., F.A.C.M.I.* — NLM, NIH  
*Michael S. Watson, M.S., Ph.D.* — American College of Medical Genetics  
*George A. Komatsoulis, Ph.D.* — Center for Biomedical Informatics and Information Technology (CBIIT), NCI  
*Kyle Brown* — Innolyst, Inc.
- B.1 Patient Participation/Outreach Activities and Patient Advocacy**  
**Chairs:** *Amy Farber, Ph.D., Kate McCurdy,* and *Paul A. Harris, Ph.D.*  
**Note takers:** *Kate McCurdy* and *Jennifer Farmer, M.S., C.G.C.*
- Discussion Panel:**  
*Ronald J. Bartek* — FARA  
*Jennifer Farmer, M.S., C.G.C.* — FARA  
*Amy Farber, Ph.D.* — LAM Treatment Alliance  
*Leslie B. Gordon, M.D., Ph.D.* — Progeria Research Foundation  
*Lynn Etheredge* — Rapid Learning Project, George Washington University  
*Paul A. Harris, Ph.D.* — Office of Research Informatics Operation, Vanderbilt University



## C.1 Biospecimens/Biorepositories

**Chairs:** *Jim B. Vaught*, Ph.D. and *Christopher A. Moskaluk*, M.D., Ph.D.

**Note takers:** *Josh Sommer*

**Discussion Panel:**

*Jim B. Vaught*, Ph.D. — OBBR, NCI

*Christopher A. Moskaluk*, M.D., Ph.D. — UVA Biorepository

*Simone S. Sommer*, M.D., M.P.H. — Chordoma Foundation

*Sharon F. Terry*, M.A. — Genetic Alliance

*Benjamin M. Greenberg*, M.D., M.H.S. — University of Texas Southwestern

*Ian M. Fore*, D.Phil. — CBIIT, NCI

*Jeffrey A. Thomas* — NDRI

**11:30 A.M. WORKING LUNCH — Chairs, note takers, and discussion panel members prepare breakout session summaries**

**1:00 P.M. Afternoon Parallel Sessions**

## B2. Clinical Trials/Research Studies and Patient Care Management

**Chairs:** *Ronald A. Christensen*, M.D. and *Vanessa Rangel Miller*, M.S., C.G.C.

**Note takers:** *Rachel Richesson*, Ph.D., M.P.H. and *Kate McCurdy*

**Discussion Panel:**

*Ronald A. Christensen*, M.D. — REGISTRAT-MAPI

*Vanessa Rangel Miller*, M.S., C.G.C. — DuchenneConnect

*Christopher B. Forrest*, M.D., Ph.D. — University of Pennsylvania School of Medicine and Children's Hospital of Philadelphia

*Robert H. Shelton*, M.B.A. — Private Access, Inc.

*Santa J. Tumminia*, Ph.D. — National Eye Institute (NEI), NIH

*Dianne M. Finkelstein*, Ph.D. — Harvard University

## C2. Human Subjects: Bioethical and Legal Issues

**Chairs:** *Jack Schwartz*, J.D. and *Sara C. Hull*, Ph.D.

**Note taker:** *Amy Farber*, Ph.D, and *Jennifer Farmer*, M.S., C.G.C. — FARA

**Discussion Panel:**

*Jack Schwartz*, J.D. — University of Maryland School of Law

*Julie Kaneshiro*, M.A. — OHRP

*Sara C. Hull*, Ph.D. — National Human Genome Research Institute (NHGRI), NIH

*P. Pearl O'Rourke*, M.D. — Partners Healthcare

*Barbara I. Karp*, M.D. — NINDS, NIH

*Wendy E. Patterson* — Technology Transfer Center, NCI

## **D. Informatics/Database Technology**

**Chairs:** *Kyle Brown* and *Lisa Forman-Neall*, Ph.D.

**Note taker:** *Rachel Richesson*, Ph.D., M.P.H.

**Discussion Panel:**

*Kyle Brown* — Innolyst, Inc.

*Lisa Forman-Neall*, Ph.D. — National Center for Biotechnology Information (NCBI), NLM

*Christophe Beroud*, Pharm.D., Ph.D. — France, INSERM

*Rachel Richesson*, Ph.D., M.P.H. — USF College of Medicine

*Chalapathy Neti*, Ph.D., B.S. — IBM Research

**3:15 P.M. BREAK FOR PARTICIPANTS — Chairs, note takers, and discussion panel members prepare breakout session summaries**

**3:45 P.M. Workshop Summary**

Day 1: *Ronald A. Christensen*, M.D. and *Christopher B. Forrest*, M.D., Ph.D.

Day 2: Breakout session presentations by the chairs of each session — summary, recommendations, and action items

**5:45 P.M. Summary Statement**

*Stephen C. Graft*, Pharm.D. — ORDR, NIH

**6:00 P.M. ADJOURN**