Please find workshop presentations, speaker abstracts, breakout session background material and summaries, and workshop summaries by clicking on the presentation or breakout session titles below.

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.

PLENARY SESSIONS

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

Developing the Rare Diseases Registry
Stephen C. Groft, Pharm.D. — ORDR, NIH

Patient and Research Advocate Statement
Amy Farber, Ph.D. — LAM Treatment Alliance

Uniting Rare Diseases
Vanessa Rangel Miller, M.S., C.G.C. — DuchenneConnect
B. Alternatives and Future Promises for a National Rare Disease Patient Registry

**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

**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

---

**PRESENTATIONS**

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

**Support for Compatibility and Interoperability**
*Kyle Brown* — Innolyst, Inc.

**Reuse of Clinical Health Records: Caveat Inquisitor**
*James J. Cimino, M.D.* — Laboratory for Informatics Development, Clinical Center (CC), NIH

**Pros and Cons of Various Models and Communication Across the Different Models**
*Clement J. McDonald, M.D.* — National Library of Medicine (NLM), NIH

**Plans for Data Standards in Rare Disease Registries**
*Rachel Richesson, Ph.D., M.P.H.* — University of South Florida (USF) College of Medicine

**Global Data Aggregation: Case Study/Treat-NMD**
*Christophe Beroud, Pharm.D., Ph.D.* — France, INSERM

**II. Biospecimens/Biorepositories**

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

**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)

**Rare Disease Biorepositories and Registries: The Need for Collaborative and Novel Approaches**
*Benjamin M. Greenberg, M.D., M.H.S.* — University of Texas Southwestern

**The Use of Patient Registries to Increase Procurements of Rare Diseases Biospecimens**
*Jeffrey A. Thomas* — National Disease Research Interchange (NRDI)
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

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

Role of Rare Disease Registries in Clinical Research

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

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)

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)

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

IV. Patient Participation and Outreach Activities/Patient Advocacy

Patient Advocacy Groups and Patient Registries: An Overview

Sukirti N. Bagal, M.D. — National Organization for Rare Disorders (NORD)

The Role of Patient Advocacy Groups in Establishing Common Infrastructure

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

Essential Elements for Translational Research in Rare Diseases: Progeria as a Case Study

Leslie B. Gordon, M.D., Ph.D. — Progeria Research Foundation

Common Diseases versus Rare Diseases: Is There Really a Difference?

Susan M. Love, M.D. — Dr. Susan Love Research Foundation

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

**Human Subjects: Ethical and Legal Issues/45 CFR 46**

*Julie Kaneshiro, M.A. — Office for Human Research Protections (OHRP)*

**Ethical and Legal Issues/Government Regulations**

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

**Legal/Bioethical Issues in Medical Research and Release of Genetic Information**

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

Breakout Sessions

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

**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
- 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

**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

**ADDITIONAL MATERIAL**

ORDR/NHGRI Genetic and Rare Diseases (GARD) Information Center

FDA Orphan Drugs Development

ClinicalTrials.gov: Trial Registration and Results Reporting