Tuesday, May 20

Capital Hilton • 1001 16th Street, NW

07:30 **REGISTRATION**

Sessions I-VII in Conjunction with the National Organization for Rare Disorders (NORD) Corporate Council

08:10 **SESSION I. Introductions and Welcome**

Jan-Inge Henter, MD, PhD — Karolinska Institute, Sweden
Domenica Taruscio, MD — National Centre Rare Diseases, Istituto Superiore di Sanità, Italy
Peter L. Saltonstal — National Organization for Rare Disorders, USA, introduction by Frank Sasinowski, JD
Stephen C. Groft, PharmD — Office of Rare Diseases (ORD), National Institutes of Health (NIH), USA

08:30 **SESSION II. Facilitating Cooperative Efforts of the Regulatory Processes: Progress on Collaborative Regulatory Requirements for the Orphan Product Designation Process between Office of Orphan Products Development (OOPD), Food and Drug Administration (FDA), USA and Committee for Orphan Medicinal Products (COMP)/European Medicines Agency (EMEA), Europe**

DISCUSSION LEADERS
Timothy R. Coté, MD, MPH — OOPD, FDA, USA (presentation)
Kerstin M. Westermark, MD — EMEA, Sweden (presentation)

DISCUSSANTS
Alex Kuta, PhD — Genzyme Corporation, USA
Catarina Edfjäll, PhD — Celgene R&D Sàrl, Switzerland
Alyssa J. Wyant — Shire Human Genetic Therapies, Inc., USA
Jordi J. Llinares Garcia, MD — EMEA, England

09:30 **SESSION III. Linking Academic Discoveries and Industry Product Development Strategies**

DISCUSSION LEADERS
Maria Wästfelt, PhD — Karolinska Institutet, Sweden
Barbara H. Wuebbels, RN, MS — BioMarin, USA

SESSION III (continued)

- **Evaluation of Dietary Supplements for the Treatment of Inborn Errors of Metabolism** (presentation)
  Paul M. Coates, PhD — Office of Dietary Supplements, NIH, USA
- **Rapid Access to Interventional Development (The RAID Pilot Program) at NIH** (presentation, abstract)
  David G. Badman, PhD — National Institute of Neurological Disorders and Stroke, NIH, USA
- **The Use of Antidotes in Hospitals and Communities, Supply Issues, and Emerging Research Needs** (presentation)
  Gregory M. Bogdan, PhD — Rocky Mountain Poison & Drug Center - Denver Health, USA
- **E-Rare Project** (presentation)
  Igor Beitia Ortiz de Zarate, PhD — GIS-Institut des Maladies Rares, France
- **Licensing of Rare and Neglected Diseases Discoveries Project** (presentation)
  Bonny Harbinger, PhD, JD — Office of Technology Transfer, NIH, USA
- **Activities at the Academic Research Centers: Identifying Present Activities and Future Opportunities**
  James C. Cloyd, PharmD — University of Minnesota College of Pharmacy, USA (presentation)
  Matt T. Reed, PhD — Keck Graduate Institute, California, USA (presentation)

10:30 **BREAK**

11:45 **Session IV. Linking Patients to Research Programs and Treatment Centers**

DISCUSSION LEADERS
Yann Le Cam, MBA — EURORDIS, France
Jorge L. Braier, MD — De Pediatria Gerrahan, Argentina

- **Experiences in Recruiting Patients for Clinical Trials** (presentation)
  Stuart W. Peltz, PhD — PTC Therapeutics, USA
- **NIH Rare Diseases Clinical Research Network** (presentation)
  Rachel L. Richesson, PhD, MPH — Rare Diseases Clinical Research Network, USA
- **Undiagnosed Diseases**
  William A. Gahl, MD, PhD — National Human Genome Research Institute (NHGRI), NIH, USA
- **Experiences with Langerhans Cell Histiocytosis** (presentation)
  Jorge L. Braier, MD — De Pediatria Gerrahan, Argentina

12:45 **LUNCH ON YOUR OWN** (boxed lunch provided)

13:45 **SESSION V. Research Methodology and Statistical Analyses for Trials of Rare Diseases and Orphan Products — Strength of Evidence: How Much Evidence is Necessary**

DISCUSSION LEADER
Simon Day, PhD — Roche Products, Ltd., England (presentation)

- Reliability of Diagnosis
- Relevance of Historical Controls
- Appropriate Endpoints
- Severity of Disease
- Size of Benefit
- Measurements of Safety

DISCUSSANTS
Jordi J. Llinares Garcia, MD — EMEA, England
Annalisa Trama, PhD — National Centre Rare Diseases, Istituto Superiore di Sanità, Italy (presentation)
Frank Ückert, MD, PhD — University Hospital Muenster, Germany
Timothy R. Coté, MD, MPH — OOPD, FDA, USA

15:00 **BREAK**

15:15 **SESSION VI. Stimulating Awareness and Research on Rare Diseases and Orphan Products through the Media**

DISCUSSION LEADERS
Amy D. Marcus — The Wall Street Journal, USA
Abbey S. Meyers — National Organization for Rare Disorders (NORD), USA (presentation)

PANEL DISCUSSION ON INFORMATION NEEDS
Arnd Brauer, PhD — Alliance for Chronic Rare Diseases, Germany (presentation)
Bo Piela — Genzyme Corporation, USA (presentation)
Virgina A. Llera, MD — Fundación GEISER, Argentina (presentation)

16:30 **SESSION VII. Rare Diseases Research Activities at the NIH**

- **After the Human Genome Project: Applying Genomics to Health**
  Alan E. Guttmacher, MD — NHGRI, NIH, USA (presentation)
- **The NIH Office of Rare Diseases: Current and Future Activities**
  Stephen C. Groft, PharmD — ORD, NIH, USA (presentation)

18:30 **NORD Annual Tribute Banquet and 25th Anniversary Celebration of the Orphan Drug Act (pre-registration with NORD required)**
***Union Station*, Washington, DC**

Wednesday, May 21

Hamilton Crowne Plaza Hotel • 14th and K Streets, NW

08:00 **SESSION VIII. WHO International Classification of Diseases and Rare Diseases Emphasis**

DISCUSSION LEADER
Stephen C. Groft, PharmD — ORD, NIH, USA

- **WHO ICD-X and ICD X-CM Update and Revision Process**
  David Berglund, MD, MPH — Centers for Disease Control and Prevention (CDC), USA (presentation)
- **ICD XI Revision Process and Rare Diseases Topic Advisory Group**
  Ségolène M. Aymé, MD — Orphanet, France (presentation)

08:45 **SESSION IX. The Value and Need for International Collaboration**

DISCUSSION LEADERS
Marlene E. Haffner, MD, MPH — Amgen, USA
Josep Torrent-Farnell, MD — COMP, Spain (presentation)

SPEAKERS
- **Fogarty International Center (FIC), NIH, USA** (presentation)
  Michael P. Johnson, MD — FIC, NIH, USA
- **Report from Latin American Congress (ER2008LA)** (presentation)
  Emilio J. Roldán — Fundación GEISER, Argentina
- **Neglected Diseases** (presentation)
  Luis Alejandro Barrera, PhD — Javeriana University Institute for the Study of Inborn Errors, Colombia
- **Policies for Orphan Drugs in the World** (presentation)
  Alice L. Pomponio, MPP — Genzyme Corporation, USA

10:00 **BREAK**

10:15 **SESSION X. A Global Look at Policy Initiatives for Rare Diseases Research and Orphan Products - Current Activities and Future Needs**
(2 panels, 45 minutes each)

SESSION X (continued)

DISCUSSION LEADERS
Manuel Posada, MD, PhD — Rare Diseases Research Institute, Instituto de Salud Carlos III, Spain
Sonja van Weely, PhD — Dutch Steering Committee on Orphan Drugs, The Netherlands

PANEL A. Global policy needs and what is being done?
Sonja van Weely, PhD — Dutch Steering Committee on Orphan Drugs, The Netherlands (presentation)
Howard H. Yuwen — Shire Human Genetic Therapies, USA
Antonio Bezerra — ANVISA, Brazil (presentation)
Jordi J. Llinares Garcia, MD — EMEA, England

DISCUSSION LEADERS
Rumen Stefanov, MD, PhD — Information Centre for Rare Diseases and Orphan Drugs, Bulgaria
Domenica Taruscio, MD — National Centre Rare Diseases, Istituto Superiore di Sanità, Italy

PANEL B. Europlan and National Plans for Rare Diseases Research and Orphan Products Development
Rumen Stefanov, MD, PhD — Information Centre for Rare Diseases and Orphan Drugs, Bulgaria (presentation)
Ségolène M. Aymé, MD — Orphanet, France (presentation, abstract)
Domenica Taruscio, MD — National Centre Rare Diseases, Istituto Superiore di Sanità, Italy (presentation)
José Marques Robalo, MD — Directorate General of Health, Portugal (presentation)

11:45 **SESSION XI. Genetic Testing and Screening Approaches**

DISCUSSION LEADERS
Joe Boone, PhD — CDC, USA
Domenica Taruscio, MD — National Centre Rare Diseases, Istituto Superiore di Sanità, Italy

- **GeneTests: State of the Art and Current Projects** (presentation)
  Roberta A. Pagon, MD — GeneTests, USA
- **EuroGenTest/Orphanet Database: New Services** (presentation, abstract)
  Ségolène M. Aymé, MD — Orphanet, France
- **Genetic Reference Materials** (presentation)
  Lisa Kalman, PhD — CDC, USA
- **Patients' Interest in Genetic Testing** (presentation)
  Sharon F. Terry, MA — Genetic Alliance, USA
- **Expanding the CETT Genetic Test Development Program** (presentation)
  Giovanna M. Spinella, MD — ORD, NIH, USA
  Andy Faucett, MS, CGC — Emory University School of Medicine, USA

13:00 **LUNCH**

14:00 **SESSION XII. Meeting Patient and Family Needs Across the Lifespan — Access to Health Care, Psychological, and Social Support Programs**

DISCUSSION LEADERS
Diane E. Dorman — NORD, USA
Anders Olauson — Ågrenska Academy, Sweden (presentation)

DISCUSSANTS
Vicky Whittemore, PhD — Tuberous Sclerosis Alliance, USA (presentation)
John Forman — New Zealand Organisation for Rare Disorders, New Zealand (presentation)
Annalisa Trama, PhD — National Centre Rare Diseases, Istituto Superiore di Sanità, Italy (presentation)
Virgina A. Llera, MD — Fundación GEISER, Argentina

15:15 **BREAK**

15:30 **SESSION XIII. Gaining Access to Information on Rare Diseases and to Orphan Products: Policy Issues and Needs**

DISCUSSION LEADERS
Yann Le Cam, MBA — EURORDIS, France
Erik Tambuyzer, PhD — Genzyme Corporation, Belgium

DISCUSSANTS

- **Pharmaceutical Industry Perspective** (presentation)
  Erik Tambuyzer, PhD — Genzyme Corporation, Belgium
- **The Role and Value of Help Lines** (presentation)
  Yann Le Cam, MBA — EURORDIS, France
- **Experiences of the Genetic Alliance** (presentation)
  Natasha Bonhomme — Genetic Alliance, USA
- **Experiences at NORD** (presentation)
  Mary H. Dunkle — NORD, USA
- **Genetic and Rare Diseases Information Center (GARD) sponsored by ORD and NHGRI** (presentation)
  Janine Lewis, MS, CGC — GARD, USA
- **Future Directions** (presentation)
  Annalisa Trama, PhD — National Centre Rare Diseases, Istituto Superiore di Sanità, Italy

16:45 **SESSION XIV. Introduction of Parallel Working Group Sessions for Thursday, May 22**

DISCUSSION LEADERS
Désirée Gavhed, PhD — Karolinska Institute, Sweden
Manuel Posada, MD, PhD — Rare Diseases Research Institute, Instituto de Salud Carlos III, Spain

17:00 **Poster Session**

18:00 **General ICORD Assembly Membership Meeting**
ICORD President, *Jan-Inge Henter*, MD, PhD — Karolinska Institute, Sweden

Thursday, May 22

Hamilton Crowne Plaza Hotel • 14th and K Streets, NW

8:30 **SESSION XV. Parallel Working Group Sessions, Workshops on Planning Future Activities and to Determine Future Needs, Goals, Venues and Implementation Mechanisms**

PANEL 1

GROUP LEADERS
Marlene E. Haffner, MD, MPH — Amgen, USA
Josep Torrent-Farnell, MD — COMP, Spain

FACILITATOR
John Ferguson, MD — ORD, NH, USA

WG I: Gaining Regulatory Approval: Establishing and Meeting Regulatory Requirements

WG III: Access to Rare Diseases Research and Orphan Products Development Assessment Tools: Possibilities Restrictions, and Solutions

PANELISTS
Erik Tambuyzer, PhD — Genzyme Corporation, Belgium
Timothy R. Cote, MD, MPH — OOPD, FDA, USA
Kerstin M. Westermark, MD — EMEA, Sweden
Lawrence Friedman, MD — Consultant, ORD, NIH, USA
Frank Ückert, MD, PhD — University Hospital Muenster, Germany
Simon Day, PhD — Roche Products, Ltd., England
Jordi J. Llinares Garcia, MD — EMEA, England
Antonio Bezerra — ANVISA, Brazil

- **FDA/EMEA Gaining Acceptance of Clinical Trials Results With Small Patient Populations: Guidance and Guidelines**
  Lawrence Friedman, MD — Consultant, ORD, NIH, USA
  Timothy R. Cote, MD, MPH — OOPD, FDA, USA (presentation)
- **Personalized Medicine: Viewing Product Approval Through Mechanism of Action vs. Disease State**
  Marlene E. Haffner, MD — MPH, Amgen, USA (presentation)

SESSION XV (continued)

- **The Precursor Role of Rare Diseases into the Use of Pharmacogenetics Leading to the Concept of Personalized Medicine**
  Open Discussion
- **Gaining Access to Approved Orphan Products – Discuss sustainability of current systems between product approval and physicians gaining access to approved treatment for patients: current business models and current healthcare systems**
  Erik Tambuyzer, PhD — Genzyme Corporation, Belgium (presentation)

PANEL 2

GROUP LEADERS
Barbara H. Wuebbels, RN, MS — BioMarin, USA
Tricia Brooks — Biotechnology Industry Organization, USA
James C. Cloyd, PharmD — University of Minnesota College of Pharmacy, USA

FACILITATORS
Rashmi Gopal-Srivastava, PhD — ORD, NIH, USA
David J. Eckstein, PhD — ORD, NIH, USA

WG II: Product Discovery and Development: Linking the Academic Research Community to the Pharmaceutical and Biotechnology Industries

- **Activities at the Academic Research Centers: Identifying Present Activities and Future Opportunities**
  Open Discussion
- **Venture Capitalist Support for Orphan Products Development**
  Open Discussion and Planning for Future Meetings

PANEL 3

GROUP LEADERS
Diane E. Dorman — NORD, USA
Anders Olauson — Ågrenska Academy, Sweden

FACILITATOR
Henrietta D. Hyatt-Knorr, MA — ORD, NIH

WG IV: Recruiting Patients for Clinical Research Studies and the Value of International Collaboration

WG VI: Patient and Family Needs Across the Lifespan: the Value of International Collaboration

WG VII: Rare Diseases Research and Orphan Products Development Activities: Expanding the Informational and Geographical Boundaries

PANEL 3 (continued)

PANELISTS
Anders Olauson — Ågrenska Academy, Sweden
John Forman — New Zealand Organisation for Rare Disorders, New Zealand
Diane E. Dorman — NORD, USA
Sharon F. Terry, MA — Genetic Alliance, USA
Arnd Brauer, PhD — Alliance for Chronic Rare Diseases, Germany
Rumen Stefanov, MD, PhD — Information Centre for Rare Diseases and Orphan Drugs, Bulgaria

- **The Role of Patient Organizations as an Advisory Council at the National Level**
  Domenica Taruscio, MD — National Centre Rare Diseases, Istituto Superiore di Sanità, Italy
- **Newborn Screening**
  John Adams — Canadian Organization for Rare Disorders, Canada
  Joan M. Keutzer, PhD — Genzyme Corporation, USA
- **The Future Role of Information Centers and Help Lines**
- **Standards of Care for Treatment of Rare Diseases**
- **The Need for Standardization of Patient Registries — Goals and Requirements**
- **Do Patients and Families Understand the Information They Obtain from Sources of Rare Diseases and Orphan Products? Are They Able to Make Informed Decisions Based on this Information?**

PANEL 4 (presentation)

GROUP LEADERS
Joe Boone, PhD — CDC, USA
Lisa Kalman, PhD — CDC, USA

FACILITATOR
Giovanna Spinella, MD — ORD, NIH

WG V: Genetic Testing for Rare Diseases in International Settings — Genetic Reference Materials, Clinical Validity and Utility of Genetic Tests and Genetic Test Standards
Joe Boone, PhD — CDC, USA
Lisa Kalman, PhD — CDC, USA

- **Expanding CETT Programs**
  Andy Faucett, MS, CGC — Emory University School of Medicine, USA
  Giovanna M. Spinella, MD — ORD, NIH, USA
  Stuart J. Hogarth, PhD — Loughborough University, England

PANEL 4 (continued)

- **Use of Standardized Mutation Nomenclature in Genetic Test Results Reporting and Databases**
  Joe Boone, PhD — CDC, USA

10:45 **SESSION XVI. Responses from Panels and Working Groups** (10 minutes each and 5 minutes of questions from audience)

DISCUSSION LEADERS
Désirée Gavhed, PhD — Karolinska Institute, Sweden
Manuel Posada, MD, PhD — Rare Diseases Research Institute, Instituto de Salud Carlos III, Spain

11:45 **SESSION XVII. Open Discussions/New Issues Forum**

DISCUSSION LEADERS
Stephen C. Groft, PharmD — ORD, NIH, USA
Yann Le Cam, MBA — EURORDIS, France
Jan-Inge Henter, MD, PhD — Karolinska Institute, Sweden

12:15 **SESSION XVIII. Summary of Meeting, Plans for ICORD 2009, and Closing**
Stephen C. Groft, PharmD — ORD, NIH, USA
Jan-Inge Henter, MD, PhD — Karolinska Institute, Sweden
Domenica Taruscio, MD — National Centre Rare Diseases, Istituto Superiore di Sanità, Italy

12:30 **ADJOURN**