

**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 (EMA), Europe**

**DISCUSSION LEADERS**

*Timothy R. Coté, MD, MPH — OOPD, FDA, USA ([presentation](#))*

*Kerstin M. Westermarck, MD — EMA, 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 — EMA, 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 Gerraahan, 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 Gerraahan, 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](#))

*Virginia 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 25<sup>th</sup> 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 Whittlemore*, 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](#))

*Virginia 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. Westermarck, 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**