



*5th International Conference on
Rare Diseases and Orphan Drugs*

**Global Approaches for Rare Diseases
and Orphan Products**

February 23-25, 2009

Programme

**Istituto Superiore di Sanità
Viale Regina Elena, 299
00161 – Roma**

Monday, February 23

**Istituto Superiore di Sanità
Viale Regina Elena, 299 • Rome**

08:00 REGISTRATION

08:30 SESSION I. Introductions and Welcome

Enrico Garaci, — President, Istituto Superiore di Sanità, Italy
Stephen Groft, — Office of Rare Diseases Research, NIH, USA
Domenica Taruscio — Centro Nazionale Malattie Rare (CNMR), Istituto
Superiore di Sanita, Italy

09:00 SESSION II. Recent and Future EU Actions on Rare Diseases

Nick Fahy — DG Sanco, European Commission

09:30 SESSION III-A. Rare Diseases: an International Public Health Priority

Yann Le Cam — EURORDIS
John Forman — NZORD

- Why a Position Paper
 - Promotion in World Health Bodies of the WHO and UN
- Development of the Concept and the Position Paper
- Outline and Methodology for Review Between 2009 – 2011
- Discussion
 - Receiving Input
 - Consultation Partners
 - Ownership
 - Dissemination and Use

**SESSION III-B. Spreading the Word of Rare Diseases Internationally - Rare
Disease Day 2008 & 2009: Experiences and Plans**

PANEL DISCUSSION

- **The new strategy of NORD for the USA**
Peter Saltonstall

SESSION III-B (continued)

• **The new paradigms of EURORDIS in EU**

Yann Le Cam

• **Promoting the cause of rare diseases over Latin America**

Virginia Llera

• **CORD is Back With an Agenda for Canada**

Durhane Wong-Rieger

• **A Lighthouse in the Sub-Saharan Africa**

Hawa Fitima

• **NZORD - Providing Direction in the South Pacific Region**

John Forman

11:00 BREAK

11:15 SESSION IV-A. Support of Networks and Patient Organizations in Rare Diseases - Consideration of Need for Working Group

DISCUSSION LEADERS

Giuliano d'Agnolo – CNMR, Istituto Superiore di Sanità, Italy

Fiorentino Capozzoli – CNMR, Istituto Superiore di Sanità, Italy

Sharon Terry – Genetic Alliance, USA

- Collect Possibilities and Ideas
- Identify Common Needs
- Search for Already Existent Solutions
- Providing Consultation to Networks
- Develop Ideas and Proposals for Different Funding Partners and Future Projects

SESSION IV-B. Facilitating Cooperative Efforts of the Regulatory Processes: Progress on Collaborative Regulatory Activities OOPD/FDA, USA and COMP/EMA, Europe

DISCUSSION LEADERS

Timothy Coté – Office of Orphan Products Development, FDA, USA

Kerstin Westermarck – European Union, Committee on Orphan Medicinal Products, Sweden

DISCUSSANT

Catarina Edfjäll – Celgene

Jordi Llinares-Garcia – EMA, UK

○ **Review of Orphan Product Designations and Approvals**

SESSION IV-B (continued)

- **European Union**

Kerstin Westermark – COMP, EU

- **United States**

Miles Braun – OOPD, FDA, USA

- **Japan**

Yukiko Nishimura – Tokyo University, Japan

- **Canada**

Maurica Maher – Associate Director of the Office of Legislative and Regulatory Modernization, Health Products and Food Branch of Health, Canada

12:30

LUNCH

13:30

SESSION V. WHO International Classification of Diseases and Rare Diseases Emphasis

Ségolène Aymé – INSERM / Orphanet, France

Antoni Montserrat – DG Sanco, European Commission

- **Orphanet Classification of Rare Diseases**

Ana Rath

- **ICD XI Revision Process and Rare Diseases Topic Advisory Group and WHO ICD-X and ICD X-CM Update and Revision Process**

Ségolène Aymé – INSERM / Orphanet, France

- **Office of Rare Diseases Research Terms in the MeSH System of the National Library of Medicine USA**

Stephen Groft, – ORDR, NIH, USA

14:15

SESSION VI. A Global Look at Policy Initiatives for Rare Diseases Research and Orphan Products - Current Activities and Future Needs

- **Global policy needs and what is being done?**

Manuel Posada – ISCIII, Spain

Sonja van Weely – the Netherlands

- **The National Program on Rare and Intractable Diseases**

Yukiko Nishimura – University of Tokyo, Japan

- **Current Activities in South Korea**

Soo Kyung Koo – South Korea National Institute of Health

SESSION VI (continued)

- **Review of Rare Diseases Research and Orphan Products Development Activities by the USA National Academy of Sciences and Institute of Medicine**

Stephen Groft – ORDR, NIH, USA

Timothy Coté – Office of Orphan Products Development, FDA, USA

- **Review of Rare Diseases Research and Orphan Products Development Activities by the European Commission**

Kerstin Westermark – COMP

Josep Torrent Y Farnell – COMP

Antoni Montserrat – DG Sanco, European Commission

15:30 **BREAK**

15:45 **SESSION VII. European and National Plans for Rare Diseases Research and Orphan Products Development**

DISCUSSION LEADERS

Domenica Taruscio – CNMR, Istituto Superiore di Sanità, Italy

Rumen Stefanov, – ICROD, Bulgaria

Nick Fahy – DG Sanco, European Commission

- **France**

Alexandra Fourcade – INSERM, France

- **Italy**

Domenica Taruscio – CNMR, Istituto Superiore di Sanità, Italy

- **Portugal**

Jose Robalo – Director General of Health, Portugal

- **Bulgaria**

Rumen Stefanov – Director, ICROD, Bulgaria

- **Germany**

Mirjam Mann – ACHSE (Alliance for Rare Diseases), Germany

17:00 – 18:00 **SESSION VIII. ICORD Board of Directors Meeting**

Tuesday, February 24

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08:00 POSTER SET-UP TIME

**08:30 SESSION IX. Linking Academic Discoveries and Industry Product
Development Strategies**

DISCUSSION LEADERS

Carlo Tomino – National Drug Agency, Italy

Barbara Wuebbels – BioMarin, USA

Tricia Books – BIO, USA

- **Innovative Medicines Initiative**

*European Federation of Pharmaceutical Industries and Associations
(EFPIA) and European Commission (to be confirmed)*

- **E-Rare Project**

Sophie Koutouzov – INSERM Paris, France

- **TEDDY –Task Force in Europe for Drug Development in the Young**

Adriana Ceci – Consortium for Biological and Pharmacological Evaluations

- **Activities at the Academic Research Centers: Identifying Present
Activities and Future Opportunities**

Jan-Inge Henter – Karolinska Institute, Stockholm, Sweden

Jim Cloyd – School of Pharmacy, University of Minnesota, USA

Ian Phillips – Keck Graduate Institute, California, USA

**09:45 SESSION X. Linkings Patients to Research Programs and Treatment
Centers – The Value of Patient Registries and Experiences in
Recruiting Patients for Clinical Trials – Report of Working Group**

OVERVIEW

Ronald A. Christensen – Arizona, USA

SESSION X (continued)

DISCUSSION LEADERS

Rachel Richesson – Rare Diseases Clinical Research Network, Tampa FL, USA

Stefano Vella – Drug Department, Istituto Superiore di Sanità, Italy

- **Utilization and Expansion of a Patient Contact Registry to Recruit Patients to the NIH Rare Diseases Clinical Research Network**

Rachel Richesson – Rare Diseases Clinical Research Network, Tampa FL, USA

(EFPIA) and European Commission (to be confirmed)

- **ECRIN**

Arrigo Schieppati – Mario Negri Institute, Italy

- **EUROCAT – Epidemiological Studies**

Fabrizio Bianchi – Italy Council of Research and Tuscany Registry of Rare Diseases, Italy

- **Italian Interregional Experiences - Linking Diagnoses with Epidemiological Data and Registries:**

- **Veneto Region Registry: the experience in the Tri-veneto**

Paola Facchin – Veneto Region Administration, Italy

- **Piedmont and Valle d'Aosta Registry of Rare Diseases:**

Dario Roccatello – University of Turin, Italy

10:45 **BREAK AND POSTER VIEWING**

11:00 **SESSION XI. The Value and Need for International Collaboration**

DISCUSSION LEADERS

Josep Torrent Y Farnell – COMP, Spain

Luciano Vittozzi – CNMR, Istituto Superiore di Sanità, Italy

- **Report from Latin American Congress (ER2008LA)**

Emilio Rolda – GEISER Foundation

Virginia Llera – Ministry of Health, Argentina

- A Latin American campaign: uniting people, organizations... and nations toward rare diseases -
- Organizations view
- Academia view
- Governments view
- Including neglected diseases: Regional problems demanding international solutions.
- Accessibility to orphan products in low income regions: including the price dilemma within international R&D programs, or working in global strategies

SESSION XI (continued)

- **Necobelac network to promote information product and diffusion in public health. A possible application in the field of Rare Diseases.**
Paola De Castro – Istituto Superiore di Sanità, Italy
- **The Need for Collaborative Partners**
Kante Sitou Amede Kangni – Togo, West Africa
Koudjo Sam Devotsou – Togo, West Africa

12:00 LUNCH AND POSTER VIEWING WITH POSTER AUTHORS AT THE POSTERS

13:15 SESSION XII-A. Meeting Patient and Family Needs Across the Lifespan – Access to Information and Health Care, Psychological, and Social Support Programs

DISCUSSION LEADERS

Anders Olauson – Ågrenska Academy, Sweden
Peter Saltonstall – NORD, USA

- **Survey of Available Programs for Patients and Families**
Anders Olauson – Ågrenska Academy, Sweden
- *John Forman* - New Zealand Organization for Rare Disorders (NZORD)
- *Corrado Teofili* - National Consulta Patients' Group, Italy
- *Simona Bellagambi* – UNIAMO, Italy
- *Sharon Terry* - Genetic Alliance, USA
- *Peter Saltonstall* - NORD, USA
- **The experience of the Italian Helpline for Rare Diseases**
Agata Polizzi – CNMR, Istituto Superiore di Sanità, Italy

SESSION XII-B. Genetic Testing Collaborative Projects and Screening Approaches

DISCUSSION LEADERS

Andy Faucett – CDC, Atlanta, USA
Domenica Taruscio – CNMR, Istituto Superiore di Sanità, Italy

- **Genetic Tests: Current Status of EuroGenTest and Orphanet Database**
Ségolène Aymé – INSERM / Orphanet, France

SESSION XII-B (continued)

- **Genetic Reference Materials**

Lisa Kalman – CDC, Atlanta, USA

- **Clinical Utility of Genetics Tests**

Bruno Dallapiccola – Mendel Institute, Italy

- **Establishing a Rare Genetic Disease Testing Portal**

Giovanna Spinella – ORDR, USA

Janine Lewis – Genetic and Rare Diseases Information Center, ORDR, USA

14:45 **BREAK**

15:00 **SESSION XIII. Discussion of Working Group Procedures and Presentation of Results and Recommendations**

Annalisa Trama – CNMR, Istituto Superiore di Sanità, Italy

Manuel Posada – ISCIII, Spain

PARALLEL WORKING GROUP SESSIONS:

Working Group A – Regulatory Needs

Kerstin Westermark – COMP, EU

Timothy Coté – OOPD USA

Jordi Llinares-Garcia – EMEA, EU

- Facilitating Cooperative Efforts of the Regulatory Processes: Progress on Collaborative Regulatory Activities OOPD/FDA, USA and COMP/EMA, Europe
- Research Methodology and Statistical Analyses for Trials of Rare Diseases and Orphan Products
- Institutional Review Board Approval
- Informed Consent Documents
- Managing Potential Conflicts of Interest

Working Group B – Research Collaborations

Giuseppe Traversa – National Drug Agency, Italy

Barbara Wuebbels – Bio Marin, USA

Tricia Brooks – BIO, USA

Ian Philips – Keck Graduate Institute, USA

- Linking Academic Discoveries and Industry Product Development Strategies
- Linking Patients to Research Programs and Treatment Centers – The Value of Patient Registries and Experiences in Recruiting Patients for Clinical Trials – Report of Working Group
- The Value and Need for International Collaboration

Working Group C - Patient/Family Needs and Informational Needs
Continue Panel Discussion From General Session

Anders Olauson – Ågrenska Academy, Sweden

Peter Saltonstall – NORD, USA

Working Group D - Patient and Research Registries and Epidemiological Studies

Rachel Richesson – Rare Diseases Clinical Research Network, Tampa FL, USA

Manuel Posada – ISCIII, Spain

Working Group E – Obtaining the Diagnosis of Rare Diseases

Domenica Taruscio – CNMR, Istituto Superiore di Sanità, Italy

Sharon Terry – Genetic Alliance, USA

- Undiagnosed Diseases
- Genetic Testing
- Newborn Screening (Note: This Subject May Need a Separate Working Group in the Future)

17:15 – 18:15 SESSION XIV. General Assembly Membership Meeting

CHIAR

Stephen Groft – ORDR, NIH, USA

Wednesday, February 25

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08:30 **SESSION XV. Research Methodology and Statistical Analyses for Trials of Rare Diseases and Orphan Products**

DISCUSSANT

Jordi Llinares-García, – EMEA, UK

Timothy Coté, – OOPD, USA

- **The Science of Small Clinical Trials - Report of Training Course and Value to Other Regulatory and Research Agencies**

Timothy Coté – OOPD, FDA, USA

Simon Day – Roche Products, UK

- **Bayesian Methods to ‘Strengthen’ Limited Trial or Study Data**

Simon Day – Roche Products, UK

- **Methodology Issues for Trials in Rare Diseases**

Paolo Bruzzi – Istituto dei Tumori, Genua, Italy

09:45 **SESSION XVI. Conclusions from Working Groups**

10:45 **BREAK**

11:00 **SESSION XVII. Open Discussions/New Issues Forum/Future Emphasis of ICORD**

DISCUSSION LEADERS

Stephen Groft – ORDR, NIH, USA

Jan-Inge Henter – Karolinska Institute, Stockholm, Sweden

11:45 **SESSION XVIII. Closing Session – Summary of Meeting**

Stephen Groft – ORDR, NIH, USA

Domenica Taruscio – CNMR, Istituto Superiore di Sanità, Italy

Yann Le Cam – EURORDIS, France

SESSION XVIII (continued)

Future Meeting

- **2010 – Buenos Aires, Argentina**
- **2011 – To be determined**

12:00

SESSION XIX. Adjourn