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, Europian 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/EMEA, Europe

DISCUSSION LEADERS
Timothy Coté – Office of Orphan Products Development, FDA, USA
Kerstin Westermark – European Union, Committee on Orphan Medicinal Products, Sweden

DISCUSSANT
Catarina Edfjäll – Celgene
Jordi Llinares-Garcia – EMEA, UK

○Review of Orphan Producty 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, Europian 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 ResearchTerms in the MeSH System of the National Library of Medicine USA**
  S*tephen 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**
  S*tephen 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, Europian Commission

15:30 **BREAK**

15:45 **SESSION VII. Europlan 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, Europian 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

Istituto Superiore di Sanità
Viale Regina Elena, 299 ● Rome

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 Diseses:**
    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

- **Estabilishing 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/EMEA, 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 Collaboratins
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

Istituto Superiore di Sanità
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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 Sessione – 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**