5th International Conference on
Rare Diseases and Orphan Drugs

Global Approaches for Rare Diseases and Orphan Products

February 23-25, 2009

Programme and Abstract Book

Istituto Superire di Sanità
Viale Regina Elena, 299
00161 – Roma

Monday, February 23, 2009

08:00 Registration

08:30-09:00 I. Introductions and Welcome:

Enrico Garaci President, Istituto Superiore di Sanita, Italy
Stephen Groft, Office of Rare Diseases Research, NIH, USA
Domenica Taruscio, CNMR, Istituto Superiore di Sanita, Italy

09:00-09:30 II. Recent and Future EU Actions on Rare Diseases –

- *Nick Fahy - DG Sanco*

09:30-11:00 III. A. Rare Diseases: An International Public Health Priority - Yann Le Cam EURORDIS and 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

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

Panel Discussion

- *Peter Saltonstall*: "The new strategy of NORD for the USA"
- *Yann LeCam* "The new paradigms of EURORDIS in EU"
- *Virginia Llera*: "Promoting the cause of rare diseases over Latin America"
- *Durhane Wong-Rieger* "CORD is Back With an Agenda for Canada"
- *Hawa Fitima*: a Lighthouse in the Sub-Saharan Africa"
- *John Forman* NZORD – Providing Direction in the South Pacific Region

11:00-11:15 Break

11:15-12:30 Concurrent Sessions

IV. (A) IT - Support of Networks and Patient Organizations in Rare Diseases - Consideration of Need for Working Group - *Giuliano D'Agnolo, Fiorentino Capozzoli, CNMR, Istituto Superiore di Sanità, Italy and 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

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 and Kerstin Westermark, European Union , Committee on Orphan Medicinal Products, Sweden

- Review of Orphan Product Designations and Approvals
  - European Union - *Kerstin Westermark, COMP, EU*
  - United States - *Miles Braun* , OOPD, FDA. USA
  - Japan - *Yukiko Nishimura, Tokyo University*,
  - Canada - *Maurica Maher, Associate Director of the Office of Legislative and Regulatory Modernization, Health Products and Food Branch of Health Canada*

Discussants: Catarina Edfjäll, Celgene and Jordi Llinares-Garcia, EMEA, United Kingdom

12:30-13:30 Lunch

13:30-14:15 V. WHO International Classification of Diseases and Rare Diseases Emphasis Segolene Ayme and Ana Rath Orphanet and INSERM, Paris France and Antoni Montserrat, DG Sanco

- 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 *(Segolene Ayme INSERM and Orphanet)*
- Office of Rare Diseases ResearchTerms in the MeSH System of the National Library of Medicine USA – *Stephen Groft, ORDR, NIH, USA*

14:15 – 15:30 VI. A Global Look at Policy Initiatives for Rare Diseases Research and Orphan Products - Current Activities and Future Needs

1. Global policy needs and what is being done? Discussion Leaders: *Manuel Posada, ISCIII, Spain, and Sonja van Weely, the Netherlands*
2. The National Program on Rare and Intractable Diseases - *Yukiko Nishimura, University of Tokyo, Japan*
3. Current Activities in South Korea – *Soo Kyung Koo - South Korea National Institute of Health*
4. Review of Rare Diseases Research and Orphan Products Development Activities by the USA National Academy of Sciences and Institute of Medicine – *Steve Groft, ORDR, NIH, USA, and Timothy Coté, Office of Orphan Products Development, FDA, USA*
5. Review of Rare Diseases Research and Orphan Products Development Activities by the European Commission – *Kerstin Westermark, COMP, Josep Torrent, COMP, Antoni Montserrat (DG Sanco)*

15:30 – 15:45 BREAK

15:45- 17:00 VII. Europlan and National Plans for Rare Diseases Research and Orphan Products Development - Discussion Leaders: Domenica Taruscio, ISS, Italy, Rumen Stefanov, ICROD, Bulgaria and 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*
- Bulgaria – *Rumen Stefanov, Director, ICROD*
- Germany– *Mirjam Mann, ACHSE (Alliance for Rare Diseases)*

17:00- 18:00 VIII. ICORD Board of Directors Meeting

Tuesday, February 24, 2009

08:00 - 08:30 Poster Set-up Time

08:30 – 09:45 IX. Linking Academic Discoveries and Industry Product Development Strategies

Discussion Leaders: Dr. Carlo Tomino, National Drug Agency, Italy, Barbara Wuebbels, BioMarin, USA and Tricia Brooks BIO USA

A. Innovative Medicines Initiative – *European Federation of Pharmaceutical Industries and Associations (EFPIA) and European Commission (to be confirmed)*

B. E-Rare Project - *Sophie Koutouzov, INSERM Paris, France*

C. TEDDY –Task Force in Europe for Drug Development in the Young – *Dr. Adriana Ceci, Consortium for Biological and Pharmacological Evaluations;*

D. 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 and Ian Phillips, Keck Graduate Institute, California.*

9:45 - 10:45 X. 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 – Overview: *Ronald A. Christensen, Arizona, USA*
Discussion Leader(s): 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*
- ECRIN – *Arrigo Schieppati, Mario Negri Institute, Italy*
- EUROCAT – Epidemiological Studies - *Fabrizio Bianchi, Italy Council of Research and Tuscany Registry of Rare Diseases*
- 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*

10:45-11:00 Break and Poster Viewing

11:00-12:00 XI. The Value and Need for International Collaboration

Discussion leaders: Josep Torrent y Farnell, COMP, Spain and Luciano Vittozzi, ISS, Italy

- Report from Latin American Congress (ER2008LA) - *Emilio Roldan GEISER Foundation and 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

- "Necobelac, a network of collaboration between Europe and Latin American Carribean countries to promote scientific writing and open access information for the safeguard of public health"– *Paola De Castro, ISS, Italy*
- The Need for Collaborative Partners - *Kante Sitou Amede Kangni and Koudjo Sam Devotsou - Togo (West Africa)*

12:00 – 13:15 Lunch and Poster Viewing with Poster Presenters at the Posters

13:15 - 14:45 Concurrent Sessions

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 and Peter Saltonstall NORD, USA

- *Anders Olauson* - Survey of Available Programs for Patients and Families
- *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
- *Agata Polizzi* – The experience of the Italian Helpline for Rare Diseases

XII.(B) Genetic Testing Collaborative Projects and Screening Approaches

Discussion Leaders: Andy Faucett CDC, Atlanta and Domenica Taruscio, ISS, Italy

- Genetic Tests: Current Status of EuroGenTest and Orphanet Database - *Segolene Ayme - INSERM and Orphanet, France*
- Genetic Reference Materials - *Lisa Kalman, CDC, Atlanta, USA*
- Clinic Utility of Genetic Tests – *Bruno Dallapiccola, Mendel Institute, Italy*
- Establishing a Rare Genetic Disease Testing Portal – *Giovanna Spinella , ORDR, USA and Janine Lewis, Genetic and Rare Diseases Information Center, ORDR, USA*

14:45 – 15:00 Break

15:00-17:15 XIII. Discussion of Working Group Procedures and Presentation of Results and Recommendations - Annalisa Trama, ISS Italy, and Manuel Posada, 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 Collaborations – *Giuseppe Traversa, National Drug Agency, Italy, Barbara Wuebbels, Bio Marin, US, Tricia Brooks, BIO, USA and 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 and Peter Saltonstall*

Working Group D - Patient and Research Registries and Epidemiological Studies *– Rachel Richesson and Manuel Posada*

Working Group E – Obtaining the Diagnosis of Rare Diseases – *Domenica Taruscio, ISS, Italy and 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 XIV. General ICORD Assembly Membership Meeting

Chair: Stephen Groft, ORDR, NIH, USA

Wednesday, February 25, 2009

8:30-9:45 XV. Research Methodology and Statistical Analyses for Trials of Rare Diseases and Orphan Products

- The Science of Small Clinical Trials - Report of Training Course and Value to Other Regulatory and Research Agencies – *Timothy Coté, OOPD, FDA, and Simon Day, Roche Products, UK*
- Bayesian Methods to 'Strengthen' Limited Trial or Study Data - *Simon Day, Roche* Products, United Kingdom
- Methodology Issues for Trials in Rare Diseases – *Paolo Bruzzi, Istituto dei Tumori, Genua, Italy*

Discussants: Jordi Llinares-García, EMEA. United Kingdom, Timothy Coté, OOPD, FDA

9:45-10:45 XVI. Conclusions from Working Groups

10:45-11:00 Break

11:00-11:45 XVII. Open Discussions/New Issues Forum/Future Emphasis of ICORD Meetings
Discussion leaders: Stephen Groft, ORDR, NIH, USA, Jan-Inge Henter, Karolinska Institute, Stockholm, Sweden

11:45-12:00 XVIII. Closing Session - Summary of Meeting
Stephen Groft, ORDR, NIH, USA
Domenica Taruscio, CNMR, ISS, Italy
Yann Le Cam, EURORDIS, France

Future Meeting

- **2010 - Buenos Aires, Argentina**
- **2011 – To Be Determined**

12:00 XIX. Adjourn