



# Rare Diseases Registries Workshop

Hospital Universitario La Paz, Madrid, Spain

21-22 March 2017



## PROGRAMME

### Day 1

**12:30-13:30** **Welcome lunch**

**13:30-13:40** **Welcome and introduction**

*European Commission - Consumers, Health, Agriculture and Food Executive Agency; Spanish Ministry of Health, Social Services and Equality; Instituto de Salud Carlos III and H. La Paz-CIBERER*

**13:40-14:00** **Overview of EU Health Programme projects funded between 2008-2015 related to Rare Diseases registries**

**Speaker**

*Hristina Mileva - European Commission - Consumers, Health, Agriculture and Food Executive Agency*

### **Session 1: Towards an European platform on Rare Diseases Registration**

*Chair: Agnieszka Kinsner-Ovaskainen - DG Joint Research Centre, Directorate F-Health, Consumer and Reference Materials, Health in Society Unit (JRC F1), Italy*

**14:00-14:20** **European Platform on Rare Diseases Registration**

**Speaker**

*Agnieszka Kinsner-Ovaskainen - DG Joint Research Centre*

**14:20-14:40** **EUROCAT - European Surveillance of Congenital Anomalies**

**Speaker**

*Amanda Julie Neville, University of Ferrara  
President of EUROCAT Association and leader of EUROCAT Registry, Italy*

**14:40-15:00 SCPE - Surveillance of Cerebral Palsy in Europe**

**Speaker**

*Catherine Arnaud, Toulouse III University  
Chair SCPE, France*

**15:00-15:20 EUCERD Core Recommendations on RD patient registration and data collection**

**Speaker**

*Victoria Hedley, Newcastle University  
RD-ACTION Thematic Coordinator, UK*

**15:20-16:00 Plenary Discussion: Interoperability of RD Registries**

**16:00-16:30 Coffee break**

**Session 2: Main results of the European Union's Health Programme – Rare Diseases Registries related projects**

Chair: *Eva Bermejo, Institute of Rare Diseases Research (IIER) and Research Centre on Congenital Anomalies – Institute of Health Carlos III.*

**16:30-16:50 EPIRARE - European Platform for Rare Disease Registries**

**Speaker**

*Luciano Vittozzi, National Centre for Rare Diseases, National Institute for Health.  
Coordinator of EPIRARE, Italy*

**16:50-17:10 PARENT Joint Action - Cross Border Patient Registries iNiTiative**

**Speaker**

*Marija Magajne, Nacionalni Inštitut za Javno Zdr  
Coordinator of the PARENT JA, Slovenia*

**17:10-17:30 RD-ACTION (Rare Diseases Joint Action)**

**Speaker**

*Victoria Hedley, Newcastle University  
RD-ACTION Thematic Coordinator, UK*

**17:30-17:50 Sharing experience with the European Union's Research and Innovation Programme Projects – RD-CONNECT (An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for RD research)**

**Speaker**

*Elizabeth Wood, Newcastle University  
Project manager of RD-Connect, UK*

**17:50-18:30 Plenary Discussion: Best practices in the field of RD Registries**

**19: 30 Reception cocktail / dinner**

## Day 2

### Session 3: Example of successful EU funded registries

Chair: *Susan Webb, Scientific Assistant Director of CIBERER and coordinator of ERCUSYN, Spain*

**9:30-9:45 E-HOD - European registry and network for homocystinurias and methylation defects**

**Speaker**

*M<sup>a</sup> Luz Couce Pico, Complejo Hospitalario Universitario de Santiago-CIBERER E-HOD, Spain*

**9:45-10:00 E-IMD - European registry and network for Intoxication type Metabolic Diseases**

**Speaker**

*Angels García-Cazorla, Hospital Sant Joan de Déu-CIBERER E-IMD, Spain*

**10:00-10:10 Q & A**

### Session 4: Challenges encountered by RD patient registries

Chair: *Pablo Lapunzina, Scientific Director of CIBERER, Spain*

**10:10-10:30 Challenge 1: Integration and interoperability of EU and national RD registries**

**Speaker**

*Manuel Posada de la Paz, Instituto de Investigación en Enfermedades Raras – Instituto de Salud Carlos III Spanish Rare Diseases Registry Coordinator, Spain*

**10:30-10:50 Challenge 2: Public-Private Partnership for Rare Diseases Registries**

**Speaker**

*Vinciane Pirard, Co-chair of EFPIA-Europabio Rare Diseases and OMP Task Force and Sanofi Genzyme Head of Special Care EU Public Affairs, Belgium*

**10:50-11:20 Coffee break**

**11:20-11:40 Challenge 3: Registries and patient involvement: Patient-Powered Patient Registries**

**Speaker**

*Virginie Bros-Facer, Research Infrastructure Project Manager Eurordis, France*

**11:40-12:00 Challenge 4: Overview of Health Policies on Rare Diseases. State of play of the European Reference Networks**

**Speaker**

*Enrique Terol - DG-SANTE*

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**12:00-12:45 Plenary Discussion: Current and future situation of patient registries**

**12:45-13:00 Closing remarks**

*Hristina Mileva – Chafea*  
*Susan Webb - CIBERER*

**13:00 Lunch**

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