OCTOBER 8 2012

8.00 Registration

9.00 Welcome addresses
ENRICO GARACI

9.30 EPIRARE and aim of the Workshop
DOMENICA TARUSCIO

SESSION I
REGISTRIES: EPIDEMIOLOGY AND HEALTHCARE SERVICES
Chairs: STEPHEN GROFT, DOMENICA TARUSCIO

10.00 Global Rare Diseases Patient Registry and Data Repository-GRDR Creating a global standardized resource for medical research, patient follow-up and patient outcome
YAFFA RUBINSTEIN

10.15 Current status of National Intractable Disease (Nambyo) registry in Japan – History, current issues, new trials, and future directions
HIROSHI MIZUSHIMA

10.30 RAREDIS - The Nordic Database for Rare diseases
KETIL HEIMDAL

10.45 The Italian National Registry for Rare Diseases
YLLKA KODRA

11.00 Congenital anomaly registries improve the knowledge on genetic syndromes in Europe
INGEBORG BARISIC

11.15 *Coffee break*

SESSION II
REGISTRIES: EVALUATION OF TREATMENTS AND OUTCOMES
Chairs: CARLA HOLLAK, YAFFA RUBINSTEIN

11.45 EBE Europabio Joint Task Force on Rare Diseases and Orphan Medicinal Products strategy towards a consistent framework for registries
SAMANTHA PARKER

12.00 Italian managed entry agreements applied to orphan drugs
ENTELA XOXI

12.15 Hosting a postmarketing study commitment within an existing independent registry: The plerixafor – EBMT (European Bone Marrow Transplantation) collaboration - CALM study
VINCIANE PIRARD

12.30 Approaches and challenges in measuring treatment risk and benefit in rare disease registries
ISABELLE MORIN

12.45 *Lunch and Poster Session*

SESSION III
REGISTRIES: GENETIC AND CLINICAL RESEARCH
Chairs: CRISTOPHE BEROUD, ANIL MEHTA

13.45 European Registry and network for intoxication type metabolic diseases (EIMD): developing a unique source of data
CARLO DIONISI-VICI

14.00 The Severe Chronic Neutropenia International Registry (SCNIR): An example for a multipurpose rare disease registry
CORNELIA ZEIDLER

14.15 EuroWilson: a European Network to improve the management of Wilson disease
JEAN-MARC TROCELLO

14.30 The TREAT-NMD Registries for neuromuscular diseases. International and Italian experience
ANNA AMBROSINI

14.45 The European Skeletal Dysplasia Network; 10 years of expert diagnosis of genetic skeletal diseases provided through telemedicine (The European Skeletal Dysplasia Network and Certus Technology)
MICHAEL BRIGGS

15.00 Lessons learned in the management of rare disease registries: The Euro- WABB Registry. Recruitment and Data Collection
AMY FARMER

15.15 The International Registry of Recurrent and Familial HUS/TTP as a tool for investigating two rare diseases
ARRIGO SCHIEPPATI

15.30 *Coffee break*

16.00 ROUND TABLE
APPROACHES TO THE DEFINITION OF COMMON DATA ELEMENTS
Chairs: PAUL LANDAIS, LUCIANO VITTOZZI
Participants: FABRIZIO BIANCHI, PAUL LANDAIS, EMANUELA MOLLO, MANUEL POSADA, CHRISTIANE STEINMUELLER, RUMEN STEFANOV, ELFRIEDE SWINNEN, DOMENICA TARUSCIO

17.00 DISCUSSION SESSION

17.30 PLENARY POSTER SESSION AND AWARD TO BEST POSTER
Chairs: SABINA GAINOTTI, EMANUELA MOLLO

18.30 End of the first day

OCTOBER 9 2012

SESSION IV
REGISTRIES AND PATIENTS' INVOLVEMENT
Chairs: MONICA ENSINI, RUMEN STEFANOV

8.30 Preliminary Results of the EURORDIS Patient Survey on Rare Disease registries
MONICA ENSINI

8.45 UK Strategy for Rare Kidney Disease: linking patients with experts through a sustainable registry
MARC TAYLOR

9.00 EU collaborative registry on Gaucher Disease (EuroGo)
CARLA HOLLAK

9.15 Establishment of research oriented portal web site for patients and advocacy group
YOKO SATO

9.30 The European Cystic Fibrosis Society Patient Registry: information to patients
JACQUI VAN RENS

9.45 The development of a regional Ataxia database and patients'perceptions and motivations for inclusion
CHRISTINE BLUNT

10.00 Problems and possibilities addressing rare disease in underdeveloped nations
NEPAL BISHNU PRASSAD

10.15 *Coffee Break*

10.30 PETITION TO THE EUROPEAN PARLIAMENT
ANTONI MONTSERRAT, FLAMINIA MACCHIA, DAVID TOWNEND

SESSION V
CASE STUDIES
Chairs: TZONKA MITEVA, ELENA NICOD

11.30 The DICE-APER protocol: a novel Rare Diseases best practice for improving the patient health care by general practitioners
MANUEL POSADA

11.45 Beyond drug registries and disease registries: a population-based registry globally monitoring treatments for RD patients
MONICA MAZZUCATO

12.00 Data collection methods to improve quality control: CNDR Innovation at Work
MEGAN JOHNSTON

12.15 An electronic cystic fibrosis service: a model with potential for wider use
DANIEL PECKHAM

12.30 From data collection to clinical quality management - insights into a growing web-based patient registry platform
MARTIN VERDINO

12.45 Conducting health economic evaluations for rare diseases: the use of patients registries
MARJE VAN WEELDEN

13.00 *Lunch and Poster Session*

DISCUSSION SESSION
TOWARDS A EU MULTIPURPOSE PLATFORM FOR RARE DISEASES AND ORPHAN MEDICINAL PRODUCTS
Chairs: ANTONI MONTSERRAT, LUCIANO VITTOZZI

14.00 Facilities and outputs
LUCIANO VITTOZZI

14.30 Quality assurance guidelines
MANUEL POSADA

14.50 Governance models
MONICA ENSINI

15.10 Integration with Centres of Expertise and EU Reference Networks
ANTONI MONTSERRAT

15.30 Coordination with other initiatives
MATIC MEGLIC, STEPHEN LYNN, STEPHEN GROFT

16.30 PLENARY POSTER SESSION AND AWARD TO BEST POSTER
Chairs: FABRIZIO BIANCHI, SABINA GAINOTTI, MARTINA GRECO

17.00 Conclusions
DOMENICA TARUSCIO

17.30 End of the meeting

Speakers and Chairs

Anna Ambrosini	*Fondazione Telethon, Italy*
Ingeborg Barisic	*Medical School University of Zagreb, Croatia*
Christophe Beroud	*INSERM, Paris, France*
Fabrizio Bianchi	*National Council of Reseach (CNR), Italy*
Nepal Bishnu Prasad	*Rasuwa Langtang Liring Orphan Society Bhaktapur Nepal*
Christine Blunt	*London South Bank University, UK*
Michael Briggs	*Newcastle University, UK*
Carlo Dionisi-Vici	*Bambino Gesù Children Hospital of Rome, Italy*
Monica Ensini	*EURORDIS, France*
Amy Farmer	*Birmingham Children's Hospital, UK*
Sabina Gainotti	*National Centre for Rare Diseases, National Institute of Health, Italy*
Enrico Garaci	*President of Italian National Institute of Health, Italy*
Martina Greco	*National Centre for Rare Diseases, National Institute of Health, Italy*
Stephen Groft	*Office of Rare Diseases Research, National Institutse of Health, USA*
Ketil Heimdal	*Oslo University Hospital, Norway*
Carla Hollak	*Academic Medical Center of Amsterdam, The Netherlands*
Megan Johnston	*University of Calgary, Canada*
Yllka Kodra	*National Centre for Rare Diseases, National Institute of Health, Italy*
Stefan Kölker	*University Children Hospital of Heidelberg, Germany*
Paul Landais	*Université Paris Descartes, France*
Stephen Lynn	*Newcastle University, UK*
Flaminia Macchia	*EURORDIS, France*
Monica Mazzucato	*Coordinating Centre for Rare Diseases -Veneto Region, Italy*
Matic Meglic	*Cross-border Patient Registries Initiative (PARENT), Slovenia*
Anil Mehta	*University of Dundee, UK*
Tsonka Miteva	*Bulgarian Association for Promotion of Education and Science (BAPES), Bulgaria*
Hiroshi Mizushima	*Center for Public Health Informatics, National Institute of Public Health, Japan*
Emanuela Mollo	*National Centre for Rare Diseases, National Institute of Health, Italy*
Antoni Montserrat	*European Commission, DG Health and Consumers*
Isabelle Morin	*Shire HGT Outcomes Research*
Elena Nicod	*London School of Economics, UK*
Samantha Parker	*Orphan Europe Recordati, France*
Daniel Peckham	*St James's University Hospital of Leeds, UK*
Vinciane Pirard	*Genzyme*
Manuel Posada	*Istitudo de Salud Carlos III of Madrid, Spain*
Yaffa Rubinstein	*Office of Rare Diseases Research, National Institutes of Health, USA*
Yoko Sato	*Tokyo Medical and Dental University Akira Yamamoto, Japan*
Arrigo Schieppati	*Mario Negri Institute for Pharmacological Research, Italy*
Rumen Stefanov	*Bulgarian association for Promotion of Education and Science (BAPES), Bulgaria*
Christiane Steinmuller	*Health Research, German Aerospace Center, Germany*
Elfriede Swinnen	*Scientific Institute of Public Health, Belgium*
Domenica Taruscio	*National Centre for Rare Diseases, National Institute of Health, Italy*
Marc Taylor	*University of Birmingham, UK*
David Townend	*University of Maastricht, the Netherlands*
Jean-Marc Trocello	*EuroWilson, Centre national de référence pour la maladie de Wilson, Hôpital Lariboisière, France*
Herman Van Oyen	*Scientific Institute of Public Health, Brussels, Belgium*
Jacqui Van Rens	*European Cystic Fibrosis Society Patient Registry*
Marje Van Weelden	*Shire HGT Outcomes Research*
Martin Verdino	*Asoluto public, Interactive relations, Wien, Austria*
Luciano Vittozzi	*National Centre for Rare Diseases, National Institute of Health, Italy*
Entela Xoxi	*Italian Medicines Agency (AIFA), Italy*
Cornelia Zeidler	*European Branch of the Severe Chronic Neutropenia International Registry (SCNIR), Germany*

General Information

EPIRARE (European Platform for Rare Disease Registries) is a three-year project co-founded by the European Commission within the EU Program of Community Action in the field of Public Health.

For more information: www.epirare.eu

Venue

Aula Pocchiari
Italian National Institute of Health (ISS)
Viale Regina Elena, 299 – Rome – Italy

Scientific Director

Domenica Taruscio
Italian National Centre for Rare Diseases
Italian National Institute of Health

Viale Regina Elena, 299 - 00161 Rome
Tel.06.4990.4017
Fax. 06.4990.4370

Web site:
www.iss.it/cnmr
www.epirare.eu

E-mail:
epirare@iss.it

EPIRARE
European Platform for Rare Disease Registries

International Workshop

Rare disease and orphan drug registries

October 8 - 9, 2012

Aula Pocchiari
Italian National Institute of Health (ISS)
Viale Regina Elena, 299 - Rome (Italy)