SESSION I
REGISTRIES: EPIDEMIOLOGY AND HEALTHCARE SERVICES
Chairs: Stephen Groft, Domenica Tarusco
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
Ylka Koudal
10.45 The Italian National Registry for Rare Diseases
Ketil Heimdal
11.00 Congenital anomaly registries improve the knowledge on genetic syndromes in Europe
Ingeborg Bargsic
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 Xoki
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: Christophe Beroud, Ahmed 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 Trocillo
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 expertise 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/ITP as a tool for investigating two rare diseases
Arrigo Schieppati
15.30 Coffee break
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, Elfreide Swinnen, Domenica Tarusco
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 Satoh
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 Prasad
10.15 Coffee Break
10.30 PETITION TO THE EUROPEAN PARLIAMENT
Antoni Montserrat, Flaminia Macchia, David Townend
SESSION V
CASE STUDIES
Chairs: Tzofka Mitseva, 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 Verdiso
12.45 Conducting health economic evaluations for rare diseases: the use of patients registries
Marie Van Weelden
13.00 Lunch and Poster Session
DISCUSSION SESSION
TOWARDS A EU MULTIPURPOSE PLATFOR M FOR RARE DISEASES AND ORPHAN MEDICINAL PRODUCTS
Chairs: Antoni Montserrat, Luciano Vittozzi
14.00 Facilities and outputs
Luciano Vitozzi
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 Miglic, Stephen Lynn, Stephen Groft
16.30 PETITION POSTER SESSION AND AWARD TO BEST POSTER
Chairs: Fabrizio Bianchi, Sabina Gainotti, Martina Greco
17.00 Conclusions
Domenica Tarusco
17.30 End of the meeting

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8.00 Registration
9.00 Welcome addresses
Enrico Garatt
9.30 EPI RARE and aim of the Workshop
Domenica Tarusco
SPEAKERS AND CHAIRS

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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.

EPIRARE is coordinated by the Italian National Centre for Rare Diseases (Istituto Superiore di Sanità) and involves 22 Partners in 13 Countries.

For more information: www.epirare.eu

VENUE

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International Workshop

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