

8.00 Registration

SESSION I
THE INTERNATIONAL SCENE

CHAIRS: D. TARUSCIO, M. POSADA

9.00 – European initiatives and implementation of the European Reference Networks

J. WALIGÓRA

9.30 – The National Institutional Rare Diseases Registries in Europe

9.50 D. TARUSCIO, L. VITTOZZI

10.00 – eHealth Implementation status in EU MS: perspectives on using EHR and other services

10.20 Z. KOLITSI

10.30 Break

11.00 – Ethical and Legal Aspects of Data Linkage and Sharing

11.20 M. HANSSON

11.30 – Patients willingness to participate in registries

11.50 A. KOLE

12.00 – Patients' empowerment and registries

12.20 R. BARBON GALLUPPI

12.30 GENERAL DISCUSSION

13.00 Lunch break

14.00 Poster session

SESSION II
RESULTS, EXPERIENCES AND USE OF REGISTRIES

CHAIRS: MA. STAZI, M. SALVATORE

15.00 – The Eurofever registry

15.20 M. GATTORNO

15.30 – Characterization of high-quality Rare Disease Registries by using a data mining approach (*)

15.45 A. COI

15.45 – Activities of the National Registry of Hemolytic Uremic Syndrome (HUS) in Italy, 1988-2014 (*)

16.00 G. SCAVIA

16.00 – Eurocat surveillance: Making Congenital Anomalies Preventable Rare Diseases (*)

16.15 A.J. NEVILLE

16.15 – Guidelines for optimal use of registries in trial design for small populations (*)

16.30 M.C. JANSEN-VAN DER WEIDE

16.30 – Medicines for paediatric rare diseases in EU and US (*)

16.45 V. GIANNUZZI

16.45 Discussion and Adjourn

SESSION III
THE INTEGRATION OF REGISTRIES WITH OTHER RESEARCH TOOLS

CHAIRS: P. TASCHNER, L. VITTOZZI

9.00 – The Human Phenome Ontology Project

9.20 S. KÖHLER

9.30 – BBMRI-ERIC and Rare Diseases – a platform for sustainability

9.50 M. PASTERK

10.00 – Discovering Value in RD and Registry Data

10.20 A. BROOKES

10.30 – Genetic variation databases and the HGVS nomenclature

10.50 P. TASCHNER

11.00 – Bring Your Own Data parties and beyond: make your data linkable to speed up rare disease research

11.20 M. ROOS

11.30 Break

12.00 – The European Society for Immunodeficiencies (ESID) Registry: recent advancements in the epidemiology of Primary Immunodeficiencies and how does that translate in clinical care

12.20 N. MAHLAOUI

12.30 GENERAL DISCUSSION

13.00 Adjourn

SPEAKERS AND CHAIRS

R. BARBON GALLUPPI – UNIAMO FIMR onlus

A.J. BROOKES – University of Leicester, UK

A. COI – Istituto di Fisiologia Clinica, Consiglio Nazionale delle Ricerche, Pisa, Italy

M. GATTORNO – IRCCS G. Gaslini, Italy

V. GIANNUZZI – Fondazione per la Ricerca Farmacologica Gianni Benzi onlus

M. HANSSON – Centre for Research Ethics & Bioethics, Sweden

M.C. JANSEN-VAN DER WEIDE – Academic Medical Center, The Netherlands

S. KÖHLER – Charité - Universitätsmedizin Berlin, Germany

A. KOLE – Eurordis Rare Diseases Europe, France

Z. KOLITSI – Aristotelean University of Thessaloniki, Greece

N. MAHLAOUI – Hôpital Universitaire Necker-Enfants Malades, France

A.J. NEVILLE – University of Ferrara, Italy

M. PASTERK – BBMRI-ERIC, Austria

M. POSADA – Instituto de Salud Carlos III, Spain

M. ROOS – Leiden University Medical Center, The Netherlands

G. SCAVIA – Istituto Superiore di Sanità, Rome, Italy

M. SALVATORE – National Centre for Rare Diseases, Istituto Superiore Sanità, Italy

MA. STAZI – National Center for Epidemiology, Surveillance and Health Care Promotion, Istituto Superiore di Sanità, Italy

D. TARUSCIO – National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy

P. TASCHNER – Leiden University Medical Center, The Netherlands

L. VITTOZZI – National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy

J. WALIGÓRA – Directorate General Health and Consumers European Commission

DOMENICA TARUSCIO
NATIONAL CENTRE FOR RARE DISEASES
ISTITUTO SUPERIORE DI SANITÀ

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

International Scientific Committee

Andrea Martinuzzi – IRCCS "E. Medea", Italy

Anna Kole – Eurordis Rare Diseases Europe, France

Fabrizio Bianchi – CNR, Pisa, Italy

Hanns Lochmueller – Newcastle University, UK

Manuel Posada – Instituto de Salud Carlos III, Spain

Marco Gattorno – IRCCS G. Gaslini, Italy

Markus Pasterk – BBMRI-ERIC, Austria

Paul Landais – University of Montpellier, France

Peter Robinson – Charité - Universitätsmedizin Berlin, Germany

Peter Taschner – Leiden University Medical Center, The Netherlands

Rumen Stefanov – University of Plovdiv, Bulgaria

Scientific Secretariat

Sabina Gainotti, Luciano Vittozzi, Marco Salvatore

NATIONAL CENTRE FOR RARE DISEASES

Istituto Superiore di Sanità

Via Giano della Bella 34, Rome

Tel. 06/49904395,

Fax 06/49904370

e-mail: epirare@iss.it

Technical Secretariat

Linda Agresta, Norina Serpa, Patrizia Crialeso,

Giorgio Vincenti, Stefano Diemoz

NATIONAL CENTRE FOR RARE DISEASES

Istituto Superiore di Sanità

Via Giano della Bella 34, Rome

Tel. 06/49904418

Fax 06/49904370

e-mail: epirare@iss.it

General Information

The Workshop is available free of charge.

The official language will be English.

For more information visit:

www.iss.it/cnmr

www.epirare.eu

The Final Program of the Workshop includes oral presentations of selected abstracts (*).

This workshop intends to increase the visibility of RD patient registries and databases within and beyond the scientific community and to promote the debate and the interaction among the many stakeholders of patient registration activities.

The Workshop is opened to researchers, clinicians, patients' and parents' associations, policy makers and enterprises.

The workshop consists of invited presentations as well as open oral and poster sessions based on a call for abstracts and selection by the International Scientific Committee.

Abstracts are welcome on any subject and experience useful to inform and support the debate regarding the main aims of the workshop.

It is expected that special interest will be dedicated to:

- the contribution of registration activities to recent advancements in the natural history, epidemiology and pathogenesis of rare diseases as well as patient care and quality of life;
- practical and innovative applications of registries, such as recruitment of patients in clinical trials, social and health service planning, patients' support networks and integration with other initiatives, such as biobanks and databases for genomic and phenomic analysis;
- lessons learned in the management of RD registries, e.g. regarding financial sustainability, quality assurance, ethical issues and patients' confidence, data protection, ownership and accessibility, as well as patients' contribution, involvement and advocacy initiatives;
- the impact of e-health initiatives, new communication technologies and social networks.



3rd International Workshop
RARE DISEASE AND
ORPHAN DRUG REGISTRIES

National Center Rare Diseases
Istituto Superiore di Sanità

c/o Casa dell'Aviatore
Viale dell'Università, 20

November 24th -25th, 2014

Rome (Italy)