**Monday November 24th, 2014**

**SESSION I**

**THE INTERNATIONAL SCENE**

**CHAIRS:** D. TARUSCIO, M. POSADA

- **9.30 – 9.50** EU action on rare diseases
  - A. MONTSERRAT

- **10.00 – 10.20** The National Institutional Rare Diseases Registries in Europe
  - D. TARUSCIO, L. VITTOZZI

- **10.30 – 10.50** eHealth Implementation status in EU MS: perspectives on using EHR and other services
  - (SPEAKER TBC)

- **11.00** Break

- **11.30 – 11.50** Ethical and Legal Aspects of Data Linkage and Sharing
  - M. HANSSON

- **12.00 – 12.20** Patients willingness to participate in registries
  - A. KOLE

- **12.30** Discussion

- **13.00** Lunch break

- **14.00** Poster session

**SESSION II**

**RESULTS, EXPERIENCES AND USE OF REGISTRIES**

**CHAIRS:** M. STADI, M. SALVATORE

- **15.00 – 15.20** The Eurofever registry
  - M. GATTORNO

- **15.30 – 15.45** Characterization of high-quality Rare Disease Registries by using a data mining approach (*)
  - A. COI

- **15.45 – 16.00** Activities of the National Registry of Hemolytic Uremic Syndrome (HUS) in Italy, 1988-2014 (*)
  - G. SCAVIA

- **16.00 – 16.15** Eurocat surveillance: Making Congenital Anomalies Preventable Rare Diseases (*)
  - A.J. NIEVLE

- **16.15 – 16.30** Guidelines for optimal use of registries in trial design for small populations (*)
  - M.C. JANSSEN-VAN DER WEIDE

- **16.30 – 16.45** Medicines for paediatric rare diseases in EU and US (*)
  - V. GIANNUZZI

- **16.45** Discussion and Adjourn

**Tuesday November 25th, 2014**

**SESSION III**

**THE INTEGRATION OF REGISTRIES WITH OTHER RESEARCH TOOLS**

**CHAIRS:** P. TASCHNER, L. VITTOZZI

- **9.00 – 9.20** The Human Phenome Ontology Project
  - S. KÖHLER

- **9.30 – 9.50** BBMRI-ERIC and Rare Diseases – a platform for sustainability
  - M. PASTERK

- **10.00 – 10.20** Discovering Value in RD and Registry Data
  - A. BROOKES

- **10.30 – 10.50** Genetic variation databases and the HGVS nomenclature
  - P. TASCHNER

- **11.00 – 11.20** Bring Your Own Data parties and beyond: make your data linkable to speed up rare disease research
  - M. ROOS

- **11.30** Break

- **12.00 – 12.20** The European Society for Immunodeficiencies (ESID) Registry: recent advancements in the epidemiology of Primary Immunodeficiencies and how does that translate in clinical care
  - N. MAHLADOU

- **12.30** Discussion

- **13.00** Adjourn

**SPEAKERS AND CHAIRS**

- 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. JANSSEN-VAN DER WEIDE – Academic Medical Center, The Netherlands
- S. KÖHLER – Charité – Universitätsmedizin Berlin, Germany
- A. KOLE – Eurordis Rare Diseases Europe, France
- N. MAHLADOU – Hôpital Universitaire Necker-Enfants Malades, France
- A. MONTSERRAT – Directorate of Public Health in the European Commission
- A.J. NIEVLE – University of Ferrara, Italy
- M. PASTERK – BBMRI-ERIC, Austria
- M. POSADA – Instituto de Salud Carlos III, Spain
- M. ROOS – Leiden University Medical Centre, The Netherlands
- G. SCAVIA – Istituto Superiore di Sanità, Rome, Italy
- M. SALVATORE – National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
- M.A. STADI – 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 Centre, The Netherlands
- L. VITTOZZI – National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy

**EpiRARE**

**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

Rome (Italy)

**November 24th - 25th, 2014**
**General Information**

The Workshop is available free of charge. The official language will be English.

For more information visit:
[www.iss.it/cnmr](http://www.iss.it/cnmr)
[www.epirare.eu](http://www.epirare.eu)

**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 open 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.

Registration is carried out by filling the on-line form which can be found at this link:
[https://www.surveymonkey.com/s/III_EPIRARE_Workshop](https://www.surveymonkey.com/s/III_EPIRARE_Workshop)

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