Monday November 24th, 2014

8.00  Registration

SESSION I
THE INTERNATIONAL SCENE
CHAIRS: D. TARUSCIO, M. POSSADA

9.00  –  European initiatives and implementation of the European Reference Networks
   J. WALISGORA

9.30  –  The National Institutional Rare Diseases Registries in Europe
   D. TARUSCIO, L. VITTOZZI

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

10.30  Break

11.00  –  Ethical and Legal Aspects of Data Linkage and Sharing
   M. HANSSON

11.50  –  Patients willingness to participate in registries
   A. KOLE

12.00  –  Patients’ empowerment and registries
   R. BARBIN GALLUPPI

12.30  General Discussion

13.00  Lunch break

14.00  –  Poster session

Tuesday November 25th, 2014

SESSION II
RESULTS, EXPERIENCES AND USE OF REGISTRIES
CHAIRS: MA. STAZI, M. SALVATORE

15.00  –  The Eurofever registry
   M. GATTORNIO

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

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

16.00  –  Eurocat surveillance: Making Congenital Anomalies Preventable
   A.J. NEVILLE

16.15  –  Rare Diseases (*)
   A.J. NEVILLE

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

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

9.20  –  BBMRI-ERIC and Rare Diseases – a platform for sustainability
   M. PASTERK

9.30  –  Discovering Value in RD and Registry Data
   A. BROOKES

9.50  –  Genetic variation databases and the HGVS nomenclature
   P. TASCHNER

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

10.30  Break

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

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   A.J. NEVILLE

14.20  –  Patients’ empowerment and registries
   N. MEVILLE – National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy

14.30  –  Patients’ willingness to participate in registries
   M. POSADA – Instituto de Salud Carlos III, Spain

14.45  –  The Eurofever registry
   M. PASTERK – BBMRI-ERIC, Austria

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16.45  –  Break

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**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)

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.