

MONDAY OCTOBER 21, 2013

- 8.00 Registration
9.00 Welcome addresses
FABRIZIO OLEARI
MINISTER OF HEALTH (TO BE CONFIRMED)
9.15 Aim of the Workshop
DOMENICA TARUSCIO

SESSION I THE MOVING CONTEXT OF RARE DISEASE REGISTRIES

Chairs: ANTONI MONTSERRAT, DOMENICA TARUSCIO

- 9.30 Future scenarios in the field of rare diseases
ANTONI MONTSERRAT
9.50 Toward a European Platform for rare disease Registries
SIMONA MARTIN
10.10 The EPIRARE proposal for a European Platform
LUCIANO VITTOZZI
10.30 *Coffee break*

SESSION II THE INTERNATIONAL AND EUROPEAN CONTEXT FOR RARE DISEASE RESEARCH

Chairs: ANTONI MONTSERRAT, DOMENICA TARUSCIO

- 11.00 International Consortium for rare diseases research - IRDiRC
STEPHEN GROFT
11.20 E-RARE
DARIA JULKOWSKA
11.40 EU research infrastructures
FILIPPO BERARDELLI
12.00 Gaps and synergies to develop new treatments for rare disease patients
GIOVANNI MIGLIACCIO
12.20 General Discussion
13.00 *Lunch break*
14.00 **POSTER SESSION**
(*Discussants*: SABINA GAINOTTI, EMANUELA MOLLO)

SESSION III INTEROPERABILITY OF NATIONAL AND INTERNATIONAL RARE DISEASE REGISTRY PLATFORMS

Chairs: FABRIZIO BIANCHI, RUMEN STEFANOV

- 15.00 Global Rare Diseases Patient Registry
STEPHEN GROFT, YAFFA RUBINSTEIN

- 15.20 The Spanish National RD Registry
MANUEL POSADA
15.40 The system of French National RD Registries BaMaRa-BNDMR-RaDiCo
PAUL LANDAIS
16.00 The Italian National RD Registry
YLLKA KODRA
16.20 The Paroxysmal Nocturnal Hemoglobinuria (PNH) registry
LUCIO LUZZATTO
16.40 General Discussion
17.15 End of the first day
17.15 – 19.00 **EPIRARE PARTNERS MEETING (EPIRARE PARTNERS)**

TUESDAY OCTOBER 22, 2013

SESSION IV CASE STUDIES

Chairs: SILVIA MANEA, MANUEL POSADA

- 9.00 The Coordination of Rare Diseases at Sanford (CoRDS) patient registry for all rare diseases
LIZ DONOHUE
9.15 The Eurofever registry: update on enrolment after 3 years
MARCO GATTORNO
9.30 eurlPFreg: A European Registry and Biobank for Patients with Idiopathic Pulmonary Fibrosis
FOTIOS DRAKOPANAGIOTAKIS
9.45 The I-DSD Registry – A forum for international research & professional networking
FAISAL AHMED
10.00 TOSCA, TuberOus Sclerosis Registry to IncreAse Disease Awareness: Registry Desi
CARLA FRAWDROSKI
10.15 International Registry of Steroid-Resistant Nephrotic Syndrome: updated epidemiological and clinical data
MANUELA CURRERI

- 10.45 EUROMAC- European Registry of Patients Affected by McArdle Disease and other Rare Glycogenosis presenting with exercise intolerance
ANDREA MARTINUZZI
11.00 EPIRARE Video “Half of a score”
11.15 *Coffee break*
11.40 The Law’s Role in rare disease and orphan drug registries: privacy and data
DAVID TOWNEND

SESSION V CASE STUDIES

Chairs: ANGELA BRAND, OLIVIA SCHREIBER

- 12.00 The Lombardy Regional Registry for Rare Diseases: an example of record linkage across different data sources
GEDEONE BARARLDO, LAURA BOTTANELLI
12.15 The disease registries as instruments of knowledge and research in rare skeletal dysplasias
MARINA MORDENTI
12.30 Pharmacovigilance using patient registries
JACQUI VAN RENS
12.45 Why developing an orphan drugs registry?
VIVIANA GIANNUZZI
13.00 *Lunch*

SESSION VI CASE STUDIES

Chairs: MONICA ENSINI, LUCIANO VITTOZZI

- 14.00 Issues highlighted by the EPIRARE surveys of rare disease registries
EMANUELA MOLLO, LUCIANO VITTOZZI
14.20 Advanced neuromuscular registries
KAREN RAFFERTY
14.40 Alpha-1-Antitrypsin International Registry
MAURIZIO LUISETTI
15.00 Analysis of the phenotypes in the Rett Syndrome Networked Database
ALESSANDRA RENIERI
15.20 Rare diseases and transplantations
ALESSANDRO NANNI COSTA
15.40 The future of RD registries in the EU
GEORGIOS MARGETIDIS
16.00 General Discussion
16.30 Conclusions

SPEAKERS AND CHAIRS

FAISAL AHMED	University of Glasgow, UK
GEDEONE BARALDO	General Direction for Healthcare – Lombardy Region, Italy
FILIPPO BELARDELLI	Department of Haematology, Oncology and Molecular medicine, Istituto Superiore di Sanità, Italy
FABRIZIO BIANCHI	National Council of Research (CNR), Italy
LAURA BOTTANELLI	Institute for pharmacological research "Mario Negri", Milan, Italy
ANGELA BRAND	University of Maastricht, the Netherlands
NATALIA CIRILLI	Centre for Cystic Fibrosis, Marche Region, Italy
MANUELA CURRERI	Institute for pharmacological research "Mario Negri", Milan, Italy
LIZ DONOHUE	Liz Donohue - Coordination of Rare Diseases at Sanford (CoRDS) South Dakota, USA
FOTIOS	Coordination of Rare Diseases at Sanford (CoRDS) South Dakota, USA
DRAKOPANAGIOTAKIS	the European IPF Registry Universities of Giessen and Marburg Lung Center (UGMLC), Germany
CARLA FLADROWSKI	Associazione Sclerosi Tuberosa Onlus, Milan, Italy
SABINA GAINOTTI	National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
MARCO GATTORNO	Ospedale Gaslini, Genova, Italy
VIVIANA GIANNUZZI	Foundation for pharmacological research Gianni Benzi Onlus, Bari, Italy
STEPHEN GROFT	Office of Rare Diseases Research, National Institute of Health, USA
DARIA JULKOWSKA	INSERM, Paris, France
YLKA KODRA	National Centre for Rare Diseases, National Institute of Health, Italy
PAUL LANDAIS	Université Paris Descartes, France
MAURIZIO LUISETTI	Institute for scientific research and cure - Policlinico San Matteo, Pavia, Italy
LUCIO LUZZATTO	Istituto Toscano Tumori, Firenze, Italy
SILVIA MANEA	Department of Paediatrics, University of Padua, Italy
GEORGIOS MARGETIDIS	Executive Agency of Health and Consumers, Luxembourg
SIMONA MARTIN	European Commission's Joint Research Centre, Ispra, Varese, Italy
ANDREA MARTINUZZI	Eugenio Medea-Institute for scientific research and cure, Association "La Nostra Famiglia", Treviso, Italy
GIOVANNI MIGLIACCIO	Department of Cell Biology & Neuroscience, Istituto Superiore di Sanità, Italy
EMANUELA MOLLO	National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
ANTONI MONTERRAT	European Commission, DG Health and Consumers, Brussels, Belgium
MARINA MORDENTI	Rizzoli Orthopaedic Institute, Bologna, Italy
ALESSANDRO NANNI COSTA	Director of National Centre for Transplantations, Istituto Superiore di Sanità, Rome, Italy
FABRIZIO OLEARI	President of the Istituto Superiore di Sanità, Italy
MANUEL POSADA	Istituto de Salud Carlos III of Madrid, Spain
KAREN RAFFERTY	Institute of Genetic Medicine, Newcastle University, Newcastle, UK
ALESSANDRA RENIERI	Santa Maria delle Scotte Teaching Hospital, Siena, Italy
YAFFA RUBINSTEIN	Office of Rare Diseases Research, National Institutes of Health, USA
OLIVIA SCHREIBER	Friedrich-Baur-Institut, München, Germany
RUMEN STEFANOV	Bulgarian association for Promotion of Education and Science (BAPES), Bulgaria
DOMENICA TARUSCIO	Director of National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
DAVID TOWNEND	University of Maastricht, the Netherlands
ANN UUSTALU	European Commission, DG RTD, Brussels, Belgium
JACQUI VAN RENS	European Cystic Fibrosis Society Patient Registry, Aarhus University Hospital, Denmark
LUCIANO VITTOZZI	National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy

GENERAL INFORMATION

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

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
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

Scientific Secretariat:
Attilio Anastasi, Sabina Gainotti, Emanuela Mollo,
Luciano Vittozzi
NATIONAL CENTRE FOR RARE DISEASES
Istituto Superiore di Sanità
Via Giano della bella 34 CAP 00161 Roma
Tel. 06/49904395, 06/49904371
Fax 06/49904370
e-mail: epirare@iss.it

Technical Secretariat:
Linda Agresta, Giuseppe Bernardo,
Stefano Diemoz, Giorgio Vincenti
NATIONAL CENTRE FOR RARE DISEASES
Istituto Superiore di Sanità
Via Giano della bella 34 CAP 00161 Roma
Tel. 06/49904418
Fax 06/49904417
e-mail: giorgio.vincenti@iss.it



2nd International Workshop

RARE DISEASE AND ORPHAN DRUG REGISTRIES

October 21-22, 2013

Aula Pocchiari
Istituto Superiore di Sanità (ISS)
Viale Regina Elena, 299 - Rome (Italy)