

Course Director

Domenica TARUSCIO

National Centre for Rare Diseases, Istituto Superiore di Sanità

Didactic Coordinators

Giovanni DE VIRGILIO

Office for External Relations and Centre for International Affairs, Istituto Superiore di Sanità

Amalia Egle GENTILE

National Centre for Rare Diseases, Istituto Superiore di Sanità

Scientific Secretariat

Claudio CARTA (coordinator), **Yllka KODRA**, **Paola TORRERI**

National Centre for Rare Diseases, Istituto Superiore di Sanità

Technical Secretariat

Linda AGRESTA, **Giuseppe BERNARDO**, **Luana BERNARDO**, **Patrizia CRIALESE**,
Stefano DIEMOZ, **Andrea VITTOZZI**

National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome

CONTACTS

National Centre for Rare Diseases, Istituto Superiore di Sanità

www.iss.it/cnmr rareregistries-school@iss.it +39 06 4990 4418



5th International Summer School Rare Disease & Orphan Drug Registries

GENERAL INFORMATION

Registries represent key resources for Rare Disease to increase timely and accurate diagnosis, improve patients management, tailor treatments, facilitate clinical trials, support healthcare planning and speed up research.

The "5th International Summer School on Rare Disease and Orphan Drug Registries" aims to promote the establishment of Findable, Accessible, Interoperable, Reusable (FAIR) Rare Disease (RD) registries in compliance with IRDiRC and EU Recommendations. The Summer School is organized by the National Centre for Rare Diseases (ISS) in collaboration with *Office for External Relations and Centre for International Affairs* (ISS), RD-Connect, ELIXIR, RD-Action, EURORDIS, EpiRare, European Reference Networks, ICORD

The Summer School will consist of plenary presentations and interactive small-group exercises, according to the Problem-Based Learning methodology.

The course will provide participants with useful tools and methodologies for establish a registry, the quality of the data collected and how a registry is turned into a FAIR resource.

PARTICIPANTS AND REGISTRATION

The Summer School is open to health professionals, researchers, medical specialists, registry curators, database managers and representatives of patients associations who are involved in or intend to establish a rare disease registry, mainly inside a European Reference Network [priority will be given to Registry Curators and IT support staff, ePAG members (or a patients/patient representatives linked with ePAGs)].

The Summer School will accept max 27 participants. A selection process will be applied based on the participant's background, role with reference to registry activities, and involvement in ERNs.

Important dates and further information at www.iss.it/cnmr.

FEES AND COSTS Registration is free of charge. ISS does not cover any travel, subsistence and other costs incurred by the participants to attend the event.

FINAL TEST The participants' performance will be mainly evaluated on the basis of the group work written solutions.

ATTENDANCE CERTIFICATES At the end of the Course a certificate of attendance will be handed to the participants who attended 100% of the program. No credits of Continuing Education in Medicine will be issued.

OFFICIAL LANGUAGE English.

VENUE Aula Marotta, Istituto Superiore di Sanità, Viale Regina Elena, 299 – 00161 Rome, Italy.

September 18-20, 2017

Rome, Italy

organised by



National Centre for Rare Diseases, Istituto Superiore di Sanità

in collaboration with

Office for External Relations and Centre for International Affairs, Istituto Superiore di Sanità



Day 1 **Monday, Sept. 18**

- 08:30** *Participants registration*
- 09:15** **Welcome and presentation of the course in the new European scenario** (*D. Taruscio*)
- 09:30** **European Reference Networks** (*E. Terol*)
- 09:45** **The European Platform on Rare Diseases Registration** (*S. Martin*)
- 10:00** **National registries for rare diseases** (*L. Vittozzi*)
- 10:15** **RD-Action and policy activities** (*V. Hedley*)
- 10:30** **Role of Patients (ePAGs)** (*V. Bros-Facer*)
- 10:45** **Introduction to Problem-Based Learning** (*G. De Virgilio*)
- 11:00** *Coffee-break*
- 11:15** **Session "Governance and sustainability of registries"**
PROBLEM ANALYSIS_Working in small groups with facilitators
- 12:30** *Lunch*
- 13:30** **Governance and Sustainability** (*J. Giuliano, P. Torreri*)
- 15:00** **PROBLEM SOLUTION**_Working in small groups with facilitators
- 16:00** **PRESENTATION GROUP SOLUTIONS AND FEEDBACK FROM PEERS AND EXPERTS**
- 17:30** *End of the day*

Day 2 **Tuesday, Sept. 19**

- 09:00** **Session "Quality and Legal issues"**
PROBLEM ANALYSIS_Working in small groups with facilitators
- 10:15** *Coffee-break*
- 10:30** **Quality issues** (*M. Posada, Y. Kodra*)
- 12:00** **Practical implications for data sharing under the new EU "General Data Protection Regulation"s** (*M. Tomasi*)
- 13:00** *Lunch*
- 14:00** **PROBLEM SOLUTION**_Working in small groups with facilitators
- 15:30** **PRESENTATION OF GROUP SOLUTIONS AND FEEDBACK FROM PEERS AND EXPERTS**
- 17:30** *End of the day*

Day 3 **Wednesday, Sept. 20**

- 09:00** **Session "FAIR data"**
PROBLEM ANALYSIS_Working in small groups with facilitators
- 10:15** *Coffee-break*
- 10:30** **Tools to make data FAIR**
- **What is FAIR?** (*C. Carta, M. Roos*)
 - **Common data set JRC** (*S. Martin*)
 - **Privacy preserving record linkage** (*D. van Enkevort*)
 - **Orphanet Nomenclature & Knowledge management** (*M. Hanauer*)
 - **Clinical data models and terminologies** (*R. Cornet*)
- 13:00** *Lunch*
- 14:00** **PROBLEM SOLUTION**_Working in small groups with facilitators
- 15:30** **PRESENTATION of GROUP SOLUTIONS AND FEEDBACK FROM PEERS AND EXPERTS**
- 17:30** *End of the course*

OBJECTIVES

The Course will enable the participants to establish a registry on rare diseases. In particular, at the end of the course the participants will be able to:

- Describe the resources needed for the establishment of a rare disease registry;
- Describe the features of successful strategies to ensure long time sustainability of the registry, including data quality and FAIR Data.

LEARNING METHOD

The training method will be Problem-Based Learning (PBL), a highly interactive and learner-centered approach where learning occurs by working in a small group assisted by a facilitator to develop a solution of a problem.

Scientific articles, expert lectures, consultations and feed-back are the learning resources that will support the students in the problem solution.

SPEAKERS/TRAINERS

Virginie Bros-Facer, Eurordis, France

Claudio Carta, National Centre For Rare Diseases, Istituto Superiore di Sanità, Italy

Ronald Cornet, Academic Medical Center, Universiteit van Amsterdam, The Netherlands

Giovanni De Virgilio, Office for External Relations and Centre for International Affairs, Istituto Superiore di Sanità, Italy

Joseph Giuliano, Global Medical Operations & Patient Registries Amicus Therapeutics, USA

Marc Hanauer, Directeur technique Orphanet, Inserm, France

Victoria Hedley, Newcastle University, United Kingdom

Yllka Kodra, National Centre For Rare Diseases, Istituto Superiore di Sanità, Italy

Simona Martin, Joint Research Centre, European Commission, Italy

Manuel Posada, Institute of Health Carlos III, Spain

Marco Roos, BioSemantics group, Leiden University Medical Centre, The Netherlands

Domenica Taruscio, National Centre For Rare Diseases, Istituto Superiore di Sanità, Italy

Enrique Terol, DG SANTE, European Commission, Belgium

Marta Tomasi, University of Bolzano, Italy

Paola Torreri, National Centre For Rare Diseases, Istituto Superiore di Sanità, Italy

David van Enkevort, University Medical Centre Groningen, The Netherlands

Luciano Vittozzi, former staff, National Centre For Rare Diseases, Istituto Superiore di Sanità, Italy

FACILITATORS

Maria Cristina Barbaro, Scientific Knowledge and Communication Service, Istituto Superiore di Sanità, Italy

Viviana Digiangdomenico, former staff, Istituto Superiore di Sanità, Italy

Roberto Esposito, former staff, Istituto Superiore di Sanità, Italy

Lorenza Scotti, Office for External Relations and Centre for International Affairs, Istituto Superiore di Sanità, Italy