



# 6<sup>th</sup> International Summer School Rare Disease & Orphan Drug Registries

**September 10-14, 2018**

*Istituto Superiore di Sanità - Rome, Italy*

*organised by*



National Centre for Rare Diseases, Istituto Superiore di Sanità  
(Rome, Italy)

*in collaboration with*



Preliminary program



**6<sup>th</sup> International Summer School  
Rare Disease & Orphan Drug Registries  
September 10-14, 2018 - Rome, Italy**



# GENERAL INFORMATION

## BACKGROUND and OBJECTIVES

The "6<sup>th</sup> International Summer School on Rare Disease and Orphan Drug Registries" is organized by the National Centre for Rare Diseases – Istituto Superiore di Sanità in collaboration with RD-Connect, ELIXIR-IT, ELIXIR-NL, EURORDIS, Orphanet, EuRRECa, European Reference Networks, EPIRARE and ICORD.

Registries are key resources in order to increase timely and accurate diagnosis, improve patients management, tailor treatments, facilitate clinical trials, support healthcare planning and speed up research.

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.

It intends *i)* to promote the establishment of Findable, Accessible, Interoperable, Reusable (FAIR) registries in compliance with IRDiRC and EU Recommendations and *ii)* to support cooperation among different registry stakeholders and coordination with registries that are developed within European Reference Networks and National Plans in the EU.

The **course** is organized in **two training modules** with the following objectives:

- the first one ("**Rare Disease Registries**", **September 10-12, 2018**) will provide participants with useful tools and methodologies to plan, establish and manage the registry activities.
- the second one ("**Bring Your Own Data - BYOD**", **September 13-14, 2018**) will be a hands-on experience (Bring Your Own Data), where the attendees work with FAIR data experts to make their data FAIR and linked to other data that has been made FAIR before.

## LEARNING METHOD

The first training module ("**Rare Disease Registries**", September 10-12, 2018) will consist of plenary presentations and interactive small-group exercises, according to the **Problem-Based Learning** (PBL) method. PBL is 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.

The second training module ("**Bring Your Own Data - BYOD**", September 13-14, 2018) will consist of brief plenary introductions and **practical working groups** where participants see how to make selected sample data FAIR and see how easy it becomes to answer difficult queries when a registry is FAIR.

## PARTICIPANTS AND REGISTRATION

The "6<sup>th</sup> International Summer School on Rare Disease and Orphan Drug Registries" is open to health professionals, researchers, medical specialists, registry curators, database managers and representatives of patients' associations involved in or intend to establish a rare disease registry, including ERN registries.

To ensure active participation and exchange with teaching staff and fellow participants, a maximum of 27 participants will be admitted to each training module. A selection process will be applied based on the participant's background, role with reference to registry activities, and involvement in ERNs.

Registration is possible for:

- the first training module ("**Rare Disease Registries**", September 10-12, 2018)
- the second training module ("**Bring Your Own Data - BYOD**", September 13-14, 2018)
- the entire course ("**Rare Disease Registries**" and "**Bring Your Own Data - BYOD**", September 10-14, 2018).

Important dates and further information at [www.iss.it/cnmr](http://www.iss.it/cnmr)

## FEES AND COSTS

Registration is free of charge. National Centre for Rare Diseases - Istituto Superiore di Sanità does not cover travel, subsistence and other costs incurred by the participants to attend the event.

## FINAL TEST

The attendants performance will be 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 single module or the entire course program.

No credits of Continuing Education in Medicine will be issued.

## OFFICIAL LANGUAGE

English.

## VENUE

Aula Rossi, Istituto Superiore di Sanità, Via Gianio della Bella, 34 - Rome, Italy.

6<sup>th</sup> International Summer School on Rare Disease and Orphan Drug Registries  
1<sup>st</sup> Training module "Rare Disease Registries" (September 10-12, 2018)  
PROGRAM

---

**Day 1**

**Monday, Sept. 10**

---

- 08:30 *Participants registration*
- 08:45 **Welcome and presentation of the course** (*D. Taruscio*)
- 08:50 **The European Platform on Rare Diseases Registration** (*A. Papadopoulou*)
- 09:10 **National registries for rare diseases** (*D. Taruscio*)
- 09:30 **Role of Patients (ePAGs)** (*V. Bros-Facer*)
- 09:50 **Introduction to Problem-Based Learning** (*G. De Virgilio*)
- 10:05 *Coffee-break*
- 10:20 **Session "Governance and sustainability of registries"**  
**PROBLEM ANALYSIS** *Working in small groups with facilitators*
- 11:20 **Governance and Sustainability** (*J. Giuliano, M. Salvatore, P. Torreri*)
- 13:00 *Lunch*
- 14:00 **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. 11**

---

- 09:00 **Session "Data Quality"**  
**PROBLEM ANALYSIS** *Working in small groups with facilitators*
- 10:15 *Coffee-break*
- 10:30 **Quality issues** (*S.F. Ahmed, Y. Kodra*)
- 12:30 *Lunch*
- 13:30 **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. 12**

---

- 09:00 **Session "Legal Issues and FAIR data"**  
**PROBLEM ANALYSIS** *Working in small groups with facilitators*
- 10:00 *Coffee-break*
- 10:15 **Tools to make data FAIR**
- **What is FAIR?** (*C. Carta, M. Roos, D. van Enckevort*)
  - **Orphanet Nomenclature & Knowledge management** (*M. Hanauer*)
  - **Clinical data models and terminologies** (*R. Cornet*)
  - **Privacy preserving record linkage** (*D. van Enckevort*)
- 12:00 **Practical implications for data sharing under the new EU "General Data Protection Regulation"** (*M. Tomasi*)
- 12:45 **RD-Connect Platform** (*D. Piscia*)
- 13:15 *Lunch*
- 14:00 **PROBLEM SOLUTION** *Working in small groups with facilitators*
- 16:00 **PRESENTATION of GROUP SOLUTIONS AND FEEDBACK FROM PEERS AND EXPERTS**
- 17:30 *End of the day*
-

6<sup>th</sup> International Summer School on Rare Disease and Orphan Drug Registries  
2<sup>nd</sup> Training module "Bring Your Own Data - BYOD" (September 13-14, 2018)  
PROGRAM

---

Day 4

Thursday, Sept. 13

---

- 08:30 *Participants registration*
- 09:00 **Welcome address** (*D. Taruscio*)
- 09:15 **Introduction of the BYOD** (*M. Roos, C. Carta*)
- 09:30 Session "FAIR data and related tools"  
**Introduction to FAIRification workflow and tools**
- 09:50 **Five minute presentations for FAIR data trainers to introduce their special expertise**
- 10:10 **Document ontologies and clinical ontologies. What they are and where to look** (*R. Cornet*)
- 10:30 **Use Case Introduction and work group division** (*M. Roos, C. Carta*)
- 11:00 Coffee break
- 11:30 **Working in small groups with FAIR data -Trainers**
- 13:00 Lunch
- 14:00 **Working in small groups with FAIR data Trainers**
- 16:30 **Self-sketching the FAIRification workflow**
- 17:00 **First impressions / progress report** (*M. Roos, C. Carta and D. Taruscio*)
- 17:30 *End of the day*

---

Day 5

Friday, Sept. 14

---

- 09:00 **Day 1 Recap**
- 09:30 **Working in small groups with FAIR data Trainers**
- 10:45 Coffee break
- 11:00 **Use case presentations & discussion (Attendees and FAIR data Trainers)**  
**FAIRification workflow sketch and Implications for registry managers**
- 13:00 Lunch
- 14:00 **FAIR project planning** (*M. Roos, D. van Enckevort and C. Carta*)
- 15:00 **Future challenges**
- 15:30 **Evaluation of the BYOD**
- 16:00 **Remarks and Conclusion** (*M. Roos, D. Taruscio*)
- 16:30 **End of the Course**
- 

## **SPEAKERS/TRAINERS**

*Syed Faisal Ahmed, University of Glasgow, UK*

*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, Member of the Scientific Committee for Continuing Medical Education, Istituto Superiore di Sanità, Italy*

*Friederike Ehrhart, Department of Bioinformatics-BiGCaT Maastricht University, GKC-Rett Expertise Centre, Maastricht University Medical Centre, Netherlands*

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

*Marc Hanauer, Directeur technique Orphanet, Inserm, France*

*Annika Jacobsen, Leiden University Medical Centre, The Netherlands*

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

*Luana Licata, Bioinformatics and Computational Biology Unit, Dept. of Molecular Biology, University of Rome Tor Vergata, Italy*

*Andri Papadopoulou, European Commission's Joint Research Centre, Ispra, Italy*

*Davide Piscia, Centre nacional d'anàlisi genòmica, Centre for Genomic Regulation, Barcelona, Spain*

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

*Marco Salvatore, National Centre For Rare Diseases, Istituto Superiore di Sanità, Italy*

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

*Marta Tomasi, University of Bolzano, Italy*

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

*David van Enckevort, University Medical Centre Groningen, The Netherlands*

*Allegra Via, Institute of Molecular Biology and Pathology, National Research Council, Italy*

## **PBL FACILITATORS**

*Andrea Barbara, Istituto di Sanità Pubblica, Università Cattolica del Sacro Cuore, Italy*

*Alice Borghini, Istituto di Sanità Pubblica, Università Cattolica del Sacro Cuore, Italy*

*Alice Corsaro, Istituto di Sanità Pubblica, Università Cattolica del Sacro Cuore, Italy*

*Tiziana Grassi, National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy*

### **Course Director**

**Domenica TARUSCIO**

*National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy*

### **Didactic Coordinators**

#### **1<sup>st</sup> Training module**

**Giovanni DE VIRGILIO**

*Member of the Scientific Committee for Continuing Medical Education, Istituto Superiore di Sanità, Italy*

**Amalia Egle GENTILE**

*National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy*

#### **2<sup>nd</sup> Training module**

**Marco ROOS**

*Leiden University Medical Center, The Netherlands*

## **National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy**

### **Scientific Secretariat**

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

### **Technical Secretariat**

**Giuseppe BERNARDO, Luana BERNARDO, Patrizia CRIALESE, Andrea VITTOZZI**

### **CONTACTS**

[www.iss.it/cnmr](http://www.iss.it/cnmr)

[rareregistries-school@iss.it](mailto:rareregistries-school@iss.it)