



# **International Summer School on Rare Disease Registries and FAIRification of Data**

**25 – 29 September 2023  
Istituto Superiore di Sanità, Rome, Italy**

**Endorsed by**



**We inspire global collaboration  
among all rare disease stakeholders**



This Training has received funding from the European Union's Horizon 2020 Research and Innovation Programme under Grant Agreement No 825575 - European Joint Programme on Rare Diseases

## GENERAL INFORMATION

### INTRODUCTION AND OBJECTIVES

The **International Summer School on Rare Disease Registries and FAIRification of Data** is a part of a series of training activities proposed by the European Joint Programme on Rare Diseases (EJP-RD). EJP-RD is a European Commission funded project (grant agreement No 825575, 2019 – 2023) with the goal “to create a comprehensive, sustainable ecosystem allowing a virtuous circle between research, care and medical innovation”. For more information about the EJP-RD, see <https://www.ejprarediseases.org/>

In particular this Course is a part of WP14, which aims to organize residential training courses in different Countries on Data Management and Quality. Task 14.5: Training on rare disease registries and FAIRification of data at the source”. WP and Task Leader: Dr. Claudio Carta, ISS.

Course Director: Dr. Claudio Carta, ISS.

The Course is made up of 5 days of residential training organized by Istituto Superiore di Sanità (ISS) in close collaboration with, mainly, EJP-RD task partners [LUMC & UoG (Endo-ERN), IOR (Bond-ERN), ASUIUD (Metab-ERN), EURORDIS; ISCIII, LUMC, INSERM (RaDiCo), UMCG, DTL-Projects (EIXIR-NL), CNR (ELIXIR -IT), AMC, ELIXIR/EMBL-EBI (UNIMAN)]

The Course is endorsed by ICORD.

ISS has gained vast experience by organizing numerous courses focused on rare disease registries with the support of key partners. In particular since 2013 ISS has organized and hosted the “International Summer School on Rare Disease and Orphan Drug Registries” and since 2014 the “Bring Your Own Data To Link Rare Disease Registries”.

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

This course is composed of two training modules:

- in the first module (25-27 September) “Rare disease registries” participants will learn (a) what resources are needed for the establishment/maintenance of a high-quality registry (b) the features of successful strategies to ensure (i) long-time sustainability of the registry, (ii) quality, (iii) legal and ethical issues in compliance with the EU General Data Protection Regulation and (iv) FAIR principles



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- in the second module “FAIRification of data”, (28-29 September) participants, working with FAIR/Data stewards will deepen the FAIRification process. The potential of a FAIR registry, as the basis for cross resource questions, will be demonstrated by executing a query across use cases. In this part a time slot will be allocated to discuss FAIR data management and FAIR project planning.

## LEARNING METHOD

In the first module there will be plenary presentations with interactive question & answer sessions between speakers and participants, hands on and work in small groups.

In the second module, the final two days of the course, there will be interactive plenary session and hands-on experience (Bring Your Own Data, BYOD).

## PARTICIPANTS AND REGISTRATION

The training course is open to the international research community, clinicians, medical specialists, registry curators, database managers, healthcare professionals and rare disease patient representatives.

To ensure active participation and exchange with teaching staff and participants, a maximum of 30 attendees will be admitted to each training module. A selection process will be applied based on the participants' background, role with reference to registry activities, and involvement in ERNs.

This course foresees:

- a) three fellowships for participants living in a EU-13 Country or in Turkey. For more information about eligibility and criteria for selection, contact Claudio Carta at: [claudio.carta@iss.it](mailto:claudio.carta@iss.it) (in Cc [laura.cellai@iss.it](mailto:laura.cellai@iss.it))
- b) three fellowships for selected rare disease patient representatives. For more information about eligibility and criteria for selection, contact Roseline Favresse at: [roseline.favresse@eurordis.org](mailto:roseline.favresse@eurordis.org)

For each fellowship a maximum of 420,00 euro for the round travel and 150,00 euro/night for hotel accommodation for a maximum of 5 nights are available.

Participants are asked to bring their laptops in order to participate to the hands on and the practical demonstrations.

Please note: inside ISS the wearing of FFP2 masks is recommended. Social distancing will be required during the training course



## REGISTRATION

REGISTRATION IS NOW OPEN [HERE](#) until 5 July, 2023

### Registration is possible for:

- > the first training module: "Rare Disease Registries", 25-27 September, 2023
- > the second training module: "FAIRification of Data", 28-29 September, 2023
- > the entire course: "Rare Disease Registries" and "FAIRification of data", 25-29 September, 2023.

**The participants selected to attend with and without fellowship will receive an email by 30 of July 2023.**

The non-selected participants should consider themselves on a reserve list until 5 September 2023.

Applicants non contacted by e-mail by 5 September 2023 should consider themselves as not selected and are invited to apply for the next edition of the training course.

## FEES AND COSTS

The course and registration are free of charge.

The course organizers will not cover expenses incurred by the participants in any case

## LEARNING ASSESSMENT

At the end of each training module a learning assessment, based on an online multiple-choice questionnaire will take place and a satisfaction survey will be submitted to the participants.

## ATTENDANCE CERTIFICATES

At the end of the course a certificate of attendance will be handed to the participants who attended 100% of the single training module or the entire course program. No credits of Continuing Education in Medicine will be issued.



## OFFICIAL LANGUAGE

English

## VENUE

Aula Bovet, Istituto Superiore di Sanità, Viale Regina Elena, 299- Rome, Italy

**For important dates, deadlines, registration form, and further information, please visit the website at the following [LINK](#)**

## CONTACT

If you have questions, please write to Dr. Claudio Carta: [claudio.cart@iss.it](mailto:claudio.cart@iss.it) (in Cc [laura.cellai@iss.it](mailto:laura.cellai@iss.it))



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## Programme of the Course

### DAY 1

1<sup>st</sup> Training Module, 25 September, 2023

- 09:30 Participants registration
- 09:45 Welcome address & Faculty & Presentation of the course  
Marco Silano, Domenica Taruscio, Claudio Carta
- 10:00 Ice-Breaking session
- 10:10 Overview of the European Joint Programme on Rare Diseases  
Claudio Carta
- 10:20 The European Platform on Rare Disease Registration (EU RD Platform)  
Andri Papadopoulou (ONLINE)
- 10:40 Coffee-break
- 11:00 Role of Patients (ePAGs)  
Gulcin Gumus, Simone Louisse
- 11:40 Introduction to the work in small groups  
Claudio Carta
- 11:50 Working in small groups
- 12:45 Lunch
- 13:45 Aims, Governance and Sustainability of Registries  
Joseph Giuliano
- 14:45 Finalization of the work in small groups
- 15:15 Plenary presentation of group work
- 16:00 Feedback from peers and experts
- 16:45 End of the day

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## DAY 2

## 1<sup>st</sup> Training Module, 26 September, 2023

09:30 Welcome Participants

09:45 Working in small groups

10:30 Coffee-break

10:45 UnFAIR GAME (Accessibility)

Marco Roos, Claudio Carta

11:15 Lesson Learned from the UnFAIR game (by the participants)

11:30 GDPR/Ethics/Informed Consent (part 1)

Marta Tomasi, Sara Casati

12:15 Lunch

13:30 GDPR/Ethics/Informed Consent (part 2)

Marta Tomasi, Sara Casati

14:15 Finalization of the work in small groups

15:00 Plenary presentation of group work

15:40 Feedback from peers and experts

16:30 End of the day

## DAY 3

## 1<sup>st</sup> Training Module, 27 September, 2023

09:30 Welcome Participants

10:00 The Research Data Management in the life sciences:

RDM KIT and introduction to the hand on Exercise

Munazah Andrabi

10:45 Coffee-break



11:00 RDM KIT: hand on Exercise

Munazah Andrabi

11:30 Working in small groups

12:30 A framework for quality management of registries

Domenica Taruscio

12:50 Lunch

13:30 Quality of the registry

Syed Faisal Ahmed

14:30 Finalization of the work in small groups

15:00 Plenary presentation of group work

15:40 Feedback from peers and experts

16:20 Evaluation of the 1<sup>st</sup> Training Module: Satisfaction Questionnaire

16:30 Closing and remarks

Marco Silano, Domenica Taruscio, Claudio Carta

16:40 Free Networking Attendees/Speakers/Data/FAIR/stewards

17:00 End of module 1

## DAY 4

## 2<sup>nd</sup> Training Module, 28 September, 2023

09:30 Welcome participants & Presentation of the 2<sup>nd</sup> training module

Marco Roos, Claudio Carta

10:10 (Un)FAIR Game (Interoperability)

Bruna Dos Santos Vieira, Martijn Kersloot, César Bernabé, Joeri van der Velde,  
Marco Roos, Claudio Carta

11:00 Coffee-break (with Avatars)





11:20 Lesson Learned from the UnFAIR game (by the participants)

11:45 Introduction to FAIR principles and the main steps of FAIRification

Marco Roos, Claudio Carta

12:15 Introduction to drawing a conceptual model

César Bernabé

13:00 *Lunch*

14:00 Ontologies and Schemas - what they are and where to look

Ronald Cornet, Marc Hanauer

14:45 Finding and accessing registries with Metadata & Hands On

Martijn Kersloot, Joeri van der Velde

- Presentaton of Castor
- Presentation of Molgenis
- Practical demonstration

16:45 Wrap Up – Container Level -

17:00 End of the Day

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## DAY 5

## 2<sup>nd</sup> Training Module, 29 September, 2023

09:30 Welcome participants

09:45 Wrap-Up

Marco Roos

10:00 Interoperability and Reusability of data in registries

Alberto Cámara, Pablo Alarcon, Martijn Kersloot

10:45 Break



11:00 DEMO Querying across registries with ontologies

Alberto Cámara, Pablo Alarcon, Martijn Kersloot, Joeri van der Velde

11:45 Wrap up – Record Level -

12:00 Data FAIRification: Implications for “registry managers” and project planning

Marco Roos, Bruna Dos Santos Vieira, Claudio Carta

- FAIRification for Managers
- Hands on: “create your own FAIRification team and project”

13:00 Lunch

14:00 Parking Lot

14:20 Evaluation of the 2nd Training Module: Satisfaction Questionnaire

14:30 Closing Remarks

Marco Roos, Claudio Carta

14:45 Free Networking for Going FAIR Attendees/Speakers/Data/FAIR/stewards

16:00 End of the Course



## SPEAKERS/IT-TRAINERS

**Syed Faisal Ahmed**, University of Glasgow, UK (EndoERN)

**Pablo Alarcon**, Centro de Biotecnología y Genómica de Plantas (CBGP, UPM-INIA), Spain

**Munazah Andrabi**, The University of Manchester, UK

**César Bernabé**, Leiden University Medical Centre, The Netherlands

**Alberto Cámara**, Centro de Biotecnología y Genómica de Plantas (CBGP, UPM-INIA), Spain

**Sara Casati** BMMRI-ERIC ELSI Service & Research Unit, Italy

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

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

**Bruna dos Santos Vieira**, Center for Molecular and Biomolecular Informatics, Radboud University Medical Center, The Netherlands

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

**Gulcin Gumus**, EURORDIS, France

**Marc Hanauer**, Orphanet, INSERM, France

**Martijn Kersloot**, Academic Medical Center, Universiteit van Amsterdam, The Netherlands

**Simone Louise**, ePAG ERN Guard Heart, The Netherlands

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

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

**Marco Silano**, 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 Trento, Italy

**Joeri van der Velde**, Groeningen University Medical Center, The Netherlands



### **COURSE DIRECTOR**

Claudio Carta, National Centre for Rare Diseases, ISS, Italy

### **SCIENTIFIC SECRETARIAT**

Domenica Taruscio, Marta De Santis, National Centre for Rare Diseases, ISS, Italy

### **ORGANISING SECRETARIAT**

Linda Agresta, Laura Lee Cellai, Patrizia Crialesse, Stefano Diemoz, Girolamo Donata  
Sandro Ghirardi,  
National Centre for Rare Diseases, ISS, Italy

