



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

**September 28 – October 2, 2020  
Istituto Superiore di Sanità, Rome, Italy**

## GENERAL INFORMATION

Due to the force majeure situation, the international travel policies associated with COVID-19 and extraordinary measures to limit the spread of the virus, the Italian health authorities (including the Istituto Superiore di Sanità, ISS), recommend adopting a precaution approach and cancel the planned trips and events. In order to ensure the correct progress of the planned tasks of EJP RD, ISS ensures that the international course “ International Summer School on Rare Disease Registries and FAIRification of Data ”, September 28 October 2,2020, ISS, Rome **will be held ONLINE**.

### RELEVANCE, 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 the WP14 on “Data Management & Quality Training”, which aims to organize residential training courses in different Countries. WP and Task Leader: Dr. Claudio Carta, ISS.

Course Director: Dr. Domenica Taruscio, ISS.

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

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”.

### SPECIFIC LEARNING OBJECTIVES

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

This course is composed of two training modules:

- During the first three days module (28-30 September, 2020), 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

- During the second two days module “FAIRification of data”, (1-2 October, 2020) participants will deepen their knowledge on the single steps of the FAIRification of data and will discover the potential of FAIR registries. 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, interactive and question & answers sessions between speakers and participants.

In the second module, on the last two days of the course, for each step of the FAIRification process there will be plenary sessions followed by hands-on exercises and a wrap up session.

## 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 patients 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.

## REGISTRATION

### Registration is possible for:

- > the first training module: “Rare Disease Registries”, September 28-30, 2020
- > the second training module: “FAIRification of Data”, October 1-2, 2020
- > the entire course: “Rare Disease Registries” and “FAIRification of data”, September 28 – October 2, 2020.

Online registration form is available at <https://sondage.inserm.fr/index.php/257338/lang-en> until July 19, 2020.

An e-mail will be sent, by July 31, 2020, to the selected participants for the course

Respondents who are not contacted by email should consider themselves not selected but will be kept on a waiting list until September 7, 2020.

## FEES AND COSTS

The course and registration is free of charge.

The course organisers 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 and an oral test, will take place

**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**

ONLINE

**CONTACT**

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

## Program of the Course

### DAY 1

1<sup>st</sup> Training Module, September 28, 2020

14:00 Welcome address & Faculty & Presentation of the course

**Domenica Taruscio**

14:15 Presentation of the European Joint Programme on Rare Diseases

**Domenica Taruscio, Claudio. Carta**

14:30 The European Platform on Rare Disease Registration (EU RD Platform)

**Andri Papadopoulou**

15:00 Questions & Answers

15:30 Aims, Governance & Sustainability

**Joseph Giuliano**

16:30 Questions & Answers

17:00 End of the Day 1

### DAY 2

September 29, 2020

14:00 Quality of RD Registries part 1

**Manuel Posada**

14:45 Questions & Answers

15:15 Quality of RD Registries part 2

**Yilka Kodra**

16:00 Questions & Answers

16:30 End of the Day 2

### DAY 3

September 30, 2020

14:00 Roles of RD patients in registries & research - ePAGs in ERNs

**Gulcin Gumus**

14:45 Questions & Answers

15:15 Ethics, GDPR and Informed Consent

**Marta Tomasi**

16:00 Questions & Answers

16:30 Experiences from ERNs: EuRR-Bone

**Syed Faisal Ahmed**

17:00 Questions & Answers

17:30 Evaluation of the 1<sup>st</sup> Training Module: Satisfaction Questionnaire

18:00 End of the Day 3

## **DAY 4**

## **2<sup>nd</sup> Training Module, October 1, 2020**

09:50 Welcome address & Presentation of the course

**Domenica Taruscio**

10:00 Prepare for FAIRification:

- Boosting the use of data in registries: overview of FAIRification

**Marco Roos, Claudio Carta**

- Describing your data for machines: the role of Ontologies

**Ronald Cornet**

10:20 Hands on

10:40 Wrap-up; questions & answers

11:00 Describe your registry for machines:

- Describing your registry at source

**Rajaram Kaliyaperumal**

- Describing registry access protocols at source

**Esther van Enckevort**

11:20 Hands on

11:40 Wrap-up; questions & answers

12:00 Let humans and machines know what you have

- Let the EU RD platform know what you are

**Holger Storf**

- How Orphanet helps human and machine know what you have

**Marc Hanauer**

12:20 Hands on

12:40 Wrap-up; questions & answers

13:00 End of the Morning Session

### **Afternoon Session**

15:00 The magic of preparing metadata for machines: Make metadata linkable

- The EJPRD Linked data platform

**Mark Wilkinson**

15:20 Hands on

15:40 Wrap-up; questions & answers

16:00 Describe data elements and how they are connected for machines:

- Conceptual modelling

**Marco Roos**

- Conceptual modelling with Draw.io

**Rajarm Kaliyaperumal**

16:20 Hands on

16:40 Wrap-up; questions & answers

17:00 End of the Day 4

### **DAY 5**

**October 2, 2020**

10:00 Describe your data for humans and machines around the world with ontologies

- Ontologising tools

**Esther van Enckevort, Marco Roos**

10:20 Hands on

10:40 Wrap-up on step

11:00 The magic of preparing data for machines Make Data Linkable & Host FAIR data

- The Vascern Registry Example of a registry becoming FAIR

**Martijn Kersloot**

11:20 Hands on

11:40 Wrap-up on step

12:00 The magic of preparing metadata and data for machines:

- Assess FAIR Data counting patients with certain conditions

**Mark Wilkinson, Rajarm Kaliyaperumal**

- SPARQL for humans: introduction to break-out

**Marco Roos, Claudio Carta**

12:20 Hands on

12:40 Wrap-up on

13:00 End of the Morning Session

### **Afternoon Session**

15:00 Data FAIRification: Implications for “registry managers” and project planning

**Esther van Enckevort, Marco Roos, Claudio Carta**

15:20 Hands on

15:40 Wrap-up on step

16:00 How global open FAIR data are changing the world

**Barend Mons, Erik Schultes**

16:30 Parking lot & Q&A next steps for your own FAIR registry

16:50 Evaluation of the 2<sup>nd</sup> Training Module: Satisfaction Questionnaire

17:10 Concluding remarks

**Domenica Taruscio, Marco Roos, Claudio Carta**

17:30 End of the Course



## **SPEAKERS**

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

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

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

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

**Gulcin Gumus**, Eurordis, France

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

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

**Rajaram Kaliyaperumal**, Leiden University Medical Centre, The Netherlands

**Martijn Kersloot**, Academic Medical Center, The Netherlands

**Barend Mons**, Leiden University Medical Centre, GO FAIR, The Netherlands

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

**Manuel Posada**, Institute of Health Carlos III, Madrid, Spain

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

**Erik Schultes**, GO FAIR, The Netherlands

**Holger Storf**, University Hospital Frankfur, Germany

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

**Marta Tomasi**, University of Bolzano, Italy

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

**Mark Wilkinson**, Centro de Biotecnología y Genómica de Plantas UPM-INIA (CBGP),  
Spain

## **IT-TRAINERS**

**Nirupama Benis**, Academic Medical Center, Universiteit van Amsterdam,  
The Netherlands

**Cesar Bernabe**, Leiden University Medical Centre, The Netherlands

**Bruna dos Santos Vieira** Radboudumc university medical center, The Netherlands

**Núria Queralt Rosinach** Leiden University Medical Centre, The Netherlands

**Lieze Thielemans**, Imperial College London, UK

### **COURSE DIRECTOR**

**Domenica Taruscio**, National Centre for Rare Diseases, ISS, Rome, Italy

### **SCIENTIFIC SECRETARIAT**

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Lorenza Scotti, Research Coordination and Support, ISS, Rome, Italy

### **ORGANIZING SECRETARIAT**

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