



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

**23 – 27 September 2019
Istituto Superiore di Sanità, Rome, Italy**

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.

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), HSK (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”.

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:

- The first module starts on September 23 till September 25, 2019, during these three days 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

- The second module “FAIRification of data”, starts on September 26 till September 27, 2019 during these two days participants, working with IT-trainers, will make use case data FAIR. The potential of a FAIR registry, as the basis for cross resource questions, will be demonstrated by executing a query across the use cases that become FAIR. 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 and problem-based learning methodology (PBL). PBL is a highly interactive and learner-centred approach, in which participants working in small groups assisted by a facilitator find the solution to a problem that will be discussed at the end with the experts.

In the second module, the final two days of the course, there will be an hands-on experience (Bring Your Own Data, BYOD) with plenary sessions alternated with breakout sessions. Attendees will work in breakout groups with IT trainers.

During the first stage attendees will follow a tutorial that takes them step by step through the process of FAIRification, using a fake dataset and a set of lightweight tools. At the end of each step participants will present the results of their group to the other participants and the experts. In the second stage they can try to FAIRify their own anonymised sample data.

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

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.

This course foresees:

- a) three fellowships for participants living in an EU13 Country. For more information about eligibility and criteria for selection, contact Claudio Carta at: claudio.carta@iss.it
- b) three fellowships for selected rare disease patient representatives. For more information about eligibility and criteria for selection, contact Virginie Bros-Facer at: virginie.bros-facer@eurordis.org

For each Fellowship a maximum of 350 euros for travel and 120 euros/night for hotel accommodation and a maximum of 5 nights is available.

REGISTRATION

Registration is possible for:

- > the first training module: "Rare Disease Registries", September 23-25, 2019
- > the second training module: "FAIRification of Data", September 26-27, 2019
- > the entire course: "Rare Disease Registries" and "FAIRification of data", September 23-27, 2019.

Online registration form is available at

<https://sondage.inserm.fr/index.php/184837/lang-en> until **June 20, 2019**.

An e-mail will be sent, by July 1, 2019, to the selected participants for the course and the selected attendees for the travel fellowships.

Respondents who are not contacted by email should consider themselves not selected but will be kept on a waiting list until July 30.

FEES AND COSTS

The course and registration is free of charge. Coffee refreshments and lunches will be offered during the course. Participants must arrange their own travel, accommodation and other costs incurred to attend the course.

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

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

CONTACT

If you have questions please write to the course organiser Claudio Carta, PhD:
claudio.cart@iss.it

Preliminary Program of the Course

DAY 1

1st Training Module, September 23, 2019

- 08:45 Participants registration
- 09:10 Welcome address & Faculty & Presentation of the course_D. Taruscio
- 09:30 The European Platform on Rare Disease Registration (EU RD Platform)
A. Papadopoulou
- 10:00 Introduction to Problem Based Learning and small groups_C. Carta
- 10:15 Coffee-break
- 10:30 PROBLEM ANALYSIS (Session #1)_Working in small groups with facilitators
[Reading and analysis of the problem in groups with facilitators]
- 12:30 *Lunch*
- 13:30 PROBLEM ANALYSIS (Session #2)_Working in small groups with facilitators
[Reading and analysis of the problem in groups with facilitators]
- 14:00 Introduction to Survey: checklist for quality_C. Carta, Y. Kodra, M. Roos
- 14:30 Individual work Session with Facilitators; Fill in Surveys
- 15:00 RD registries in the Eastern EU: Situation, bottlenecks and opportunities
R. Stefanov
- 15:30 Aims, Governance & Sustainability_J. Giuliano, P. Torreri
- 17:00 End of the day

DAY 2

1st Training Module, September 24, 2019

- 08:45 Welcome Participants
- 09:15 Quality of Registries part 1_Y. Kodra, M. Posada, E. Xoxi
- 10:00 Coffee-break
- 10:15 Quality of Registries part 2_Y. Kodra, M. Posada, E. Xoxi
- 11:00 Ethics, GDPR and Informed Consent_A. Landi, M. Tomasi
- 12:30 *Lunch*
- 13:15 The FAIR guiding Principles C. Carta, M. Roos
- 13:30 A FAIR ecosystem to enable analysis, and reuse of sensitive rare disease data
M. Roos, D. van Enckevort
- 14:00 Structuring data: Ontologies_R. Cornet

- 14:30 Roles of RD patients in registries & research - ePAGs in ERNs_V. Bros Facer
- 15:10 Experiences with RD registries: EuRECa_S.F. Ahmed
- 15:40 Experiences with RD registries: ERN PaedCan and the registry for very rare tumors_G. Bisogno
- 16:10 Experiences with RD registries: Unified European Registry for Inherited Metabolic Disorders registry - U-IMD_Florian Gleich
- 16:40 Introduction to Surveys: FAIR Metrics and Digital Environment for Trainings C. Carta, A. Jacobsen, A. Via
- 16:55 Individual work Session with Facilitators; Fill in Surveys
- 17:30 End of the day

DAY 3

1st Training Module, September 25, 2019

- 08:45 Welcome Participants
- 09:00 PROBLEM SOLUTION_Working in small groups with facilitators
- 10:15 Coffee-break
- 10:30 PRESENTATION of GROUP SOLUTIONS AND FEEDBACK FROM PEERS AND EXPERTS
- 13:00 Lunch
- 14:00 Surveys results and "QA"
- 14:45 Evaluation of the 1st Training Module: Satisfaction Questionnaire
- 15:00 Concluding remarks
- 16:00 Free Networking Attendees/Speakers/Facilitators
- 17:00 End of module 1

DAY 4

2nd Training Module, September 26, 2019

- 08:30 Participants registration
- 09:00 Welcome address_Domenica Taruscio
- 09:10 Introduction of the BYOD FAIRification workflow and Round of introductions C. Carta, M. Roos
- 09:40 Introduction to FAIR metrics assessment 1 and Individual hands-on A. Jacobsen
- 10:10 Coffee break
- 10:30 Introduction to drawing a conceptual model_A. Jacobsen

- 10:45 Group hands-on 1 - Conceptual modelling_ IT-Trainers
- 11:15 Group report in plenary on hands-on 1_Experts and IT-Trainers
- 11:45 Ontologies what they are and where to look_M. Roos
- 12:15 Clinical ontologies. What they are and where to look_R. Cornet
- 12:45 Lunch
- 13:30 Group hands-on 2 - Finding ontologies_ IT-Trainers
- 14:00 Group report in plenary on hands-on 2_Experts and IT-Trainers
- 14:30 Describing rare diseases using HPO and the Orphanet Rare Disease Ontology
M. Hanauer
- 15:00 Introduction to FAIRifier tutorial_A. Jacobsen
- 15:15 Group hands-on 3 FAIRifier tutorial_IT-Trainers
- 16:45 Group report in plenary on hands-on 3_Experts and IT-Trainers
- 17:15 First impressions and Recap of the Day_All
- 17:30 End of the day

DAY 5

2nd Training Module, September 27, 2019

- 09:00 Machine readable and querying linkable data
R. Cornet, M. Roos, M. Wilkinson
- 09:45 Group hands-on 4: querying linkable data_IT-Trainers
- 10:15 Coffee break
- 10:30 Group report on hands-on 4 in plenary_Experts and IT-Trainers)
- 11:00 Group hands-on 5: "Your own data group", FAIRification workflow_IT-Trainers
- 12:00 Group report and sketch "your own data"_Experts and IT-Trainers
- 12:45 Individual hands-on survey on: FAIR metrics assessment 2
- 13:00 Individual hands-on survey on: Digital Environment for Trainings
- 13:15 Lunch
- 14:00 Data FAIRification: Implications for "registry managers" and project planning
C. Carta, M. Roos, D. van Enckevort
- 14:45 Reflections on FAIR metrics 1&2_A. Jacobsen
- 15:15 Evaluation of the 2nd Training Module: Satisfaction Questionnaire
- 15:30 Remarks and Conclusion_M. Roos, D. Taruscio

16:00 Free Networking Attendees/Speakers/IT-trainers

17:00 End of the Course

SPEAKERS/IT-TRAINERS

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

Gianni Bisogno, Università degli Studi di Padova, Italy (ERN PaedCan)

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

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

Florian Gleich, University Hospital Heidelberg (MetabERN)

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

Annalisa Landi, Fondazione per la Ricerca Farmacologica Gianni Benzi

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

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

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

Rumen Stefanov, Medical University of Plovdiv, Bulgaria

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

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

Entela Xoxi, Catholic University "Sacro Cuore" Rome, Former Coordinator AIFA
registries