EUROPEAN JOINT PROGRAMME
RARE DISEASES

International Summer School on Rare Disease Registries and FAIRification of Data

September 26 – 30, 2022
Istituto Superiore di Sanità, Rome, Italy

Endorsed by

ICORD
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

26th-30th September 2022.

The training course will be held in an ONLINE FORMAT, given the uncertainties linked to the evolution of the Covid-19 pandemic and to the international situation.

REGISTRATION IS NOW OPEN HERE until the 29th of May 2022.

Respondents who are not selected and contacted by email by the 17th of June should consider themselves on a reserve list until the 9th of September 2022.

After this date the selection process will close, and we hope that the non-selected respondents will apply for the next edition of the training course.

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/
This Course is a part of the WP14 on "Data Management & Quality Training", which aims to organise residential training courses in different Countries. WP and Task Leader: Dr. Claudio Carta, ISS.

Course Director: Dr. Claudio Carta, ISS.

The Course is made up of 5 days of training organised 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 (ELIXIR-NL), CNR (ELIXIR -IT), AMC, ELIXIR/EMBL-EBI (UNIMAN).

The Course is endorsed by the International Conference On Rare Diseases and Orphan Drugs (ICORD).

ISS has gained vast experience by organising numerous courses focused on rare disease registries with the support of key partners. Since 2013 ISS has organised 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".

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

SPECIFIC LEARNING OBJECTIVES

This course is composed of two training modules:
– During the first three days module (26th -28th September 2022), 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 (European Union) General Data Protection Regulation.

– During the second two days module "FAIRification of data", (29th -30th September 2022) 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.

TRAINING METHOD

In the first and second module there will be interactive plenary presentations and question & answer sessions between speakers and participants. Moreover, in the second module there will be 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 NOW OPEN HERE until the 29th of May 2022.

Registration is possible for:
– the first training module: "Rare Disease Registries", 26th -28th September 2022
– the second training module: "FAIRification of Data", 29th -30th September 2022
– the entire course: "Rare Disease Registries" and "FAIRification of data", 26th -30th September 2022.

Respondents who are not selected and contacted by email by the 17th of June should consider themselves on a reserve list until the 9th of September 2022.

After this date the selection process will close, and we hope that the non-selected respondents will apply for the next edition of the training course.

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

For important updates, deadlines and for the online registration please visit the website at the following LINK

FEES AND COSTS

The course and registration are 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 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

ONLINE, on the Microsoft Teams Platform of the EJP RD.

The connection details will be sent by email to the selected participants some days before the start of the training course.

CONTACTS

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

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

Programme of the Course

DAY 1 (CEST Time) 1st Training Module, September 26, 2022

13:30 Registration of participants

14:00 Welcome address & Presentation of the course

Domenica Taruscio

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

Claudio Carta, Domenica Taruscio

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

Andri Papadopoulou

15:00 Questions & Answers

15:15 Break

15:45 Aims, Governance & Sustainability

Joseph Giuliano

16:45 Questions & Answers

17:00 End of Day 1

DAY 2 (CEST Time) 1st Training Module, September 27, 2022

10:00 Quality of RD Registries

Yllka Kodra

10:45 Questions & Answers

11:00 Break

11:20 Ethics, GDPR and Informed Consent

Marta Tomasi

12:20 Questions & Answers

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

12:30 Lunch

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

Gulcin Gumus

14:30 Questions & Answers

14:45 Implementing the active partnership with patients' representatives in a specific ERN Registry: the management of TogethERN ReCONNET

Diana Marinello

15:15 Questions & Answers

15:30 ERN GUARD-Heart Patient Registries

Ahmad S. Amin

16:15 Questions & Answers

16:30 End of Day 2

DAY 3 (CEST Time) 1st Training Module, September 28, 2022

10:00 Rare disease registries and how they work in real life – the EuRRECa experience

Syed Faisal Ahmed

10:45 Questions & Answers

11:00 Break

11:30 Experiences with building and managing a registry: The Italian Cystic Fibrosis Patients Registry

Marco Salvatore

12:15 Questions & Answers

12:30 Lunch

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

14:00 The Research Data Management in the life sciences:

RDM KIT and hand on Exercise

Munazah Andrabi, Carole Goble

15:30 Learning assessment of the 1st Training Module and satisfaction survey

16:00 End of Day 3

16:00 – 17:00 Social Event

DAY 4 (CEST Time) **2nd Training Module, September 29, 2022**

09:50 Welcome address & Presentation of the 2nd training module

Domenica Taruscio

10:00 Introduction of the FAIRification module

Claudio Carta, Marco Roos

10:10 (Un)FAIR Game

Bruna Dos Santos Vieira Marco Roos, Rajaram Kaliyaperumal, Martijn Kersloot, César Bernabé, Clémence Le Cornec, Shuxin Zhang, Alberto Cámara, Joeri van der Velde

11:00 Break with the Avatars

11:15 (Your) Cross-Resource questions and the Benefits of making data FAIR

Marco Roos, Claudio Carta

11:45 The main steps of FAIRification

Marco Roos, Claudio Carta

12:00 Introduction to drawing a conceptual model and hands on

César Bernabé

12:30 Break

13:30 Ontologies and Schemas - what they are and where to look

Ronald Cornet, Marc Hanauer

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

14:15 Hands on: Finding and accessing registries with Metadata

Martijn Kersloot, Rajaram Kaliyaperumal

15:00 Break

15:30 Hands on: Finding and accessing registries with Metadata

Martijn Kersloot, Rajaram Kaliyaperumal

16:30 Wrap-up; Questions & Answers

16:45 End of Day 4

DAY 5 (CEST Time) **2nd Training Module, September 30, 2022**

09:30 Wrap-Up

10:00 Hands on: Querying across registries with ontologies

Mark Wilkinson, Martijn Kersloot, Rajaram Kaliyaperumal

10:30 Break

11:00 Hands on: Querying across registries with ontologies

Mark Wilkinson, Martijn Kersloot, Marco Roos

11:45 FAIR Game Data & Metadata

Bruna Dos Santos Vieira, Marco Roos, Rajaram Kaliyaperumal, Martijn Kersloot, César Bernabé, Clémence Le Cornec, Shuxin Zhang, Alberto Cámara, Joeri van der Velde

12:15 Wrap Up

12:30 Lunch

14:00 Data FAIRification: Implications for "registry managers" and project planning

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

Marco Roos, Nawel Lalout, Claudio Carta, Bruna Dos Santos Vieira

15:30 Break

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

15:50 How global open FAIR data are changing the world in practice

Erik Schultes, Barend Mons

16:20 Outstanding issues, Q&A and next steps for your own FAIR registry

16:50 Learning assessment of the 2nd Training Module and satisfaction survey

17:00 Concluding remarks

Domenica Taruscio, Marco Roos, Claudio Carta

17:10 End of the Course

TRAINERS

Syed Faisal Ahmed, University of Glasgow, UK

Ahmad S. Amin, Academic Medical Centre Amsterdam, Netherlands

Munazah Andrabi, The University of Manchester, 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

Carole Goble, The University of Manchester, UK

Gulcin Gumus, EURORDIS, France

Marc Hanauer, Orphanet, INSERM, France

Rajaram Kaliyaperumal, Leiden University Medical Centre, Universiteit van Amsterdam, The Netherlands

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

Yllka Kodra, Ministry of Health Directorate-General for Health Planning Government Organisation, Italy

Nawel Lalout, Radboud University Medical Center, The Netherlands

Diana Marinello, Azienda Ospedaliera Universitaria Pisana, Italy

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

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

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

Marco Roos, Leiden University Medical Centre, The Netherlands

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

Erik Schultes, GO FAIR, The Netherlands

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

Marta Tomasi, University of Trento, Italy

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

FAIRification Stewards

César Bernabé, Leiden University Medical Centre, The Netherlands

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

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

Clemence le Cornec, Heidelberg University Hospital, Germany

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

Shuxin Zhang, Academic Medical Center, Universiteit van Amsterdam, The Netherlands

COURSE DIRECTOR

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

SCIENTIFIC SECRETARIAT

Marco Salvatore, Domenica Taruscio, Paola Torreri, National Centre for Rare Diseases, ISS, Italy

ORGANISING SECRETARIAT

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

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