International Summer School on FAIRification of Rare Disease Registries and Data

18-20 September 2024

organised by

ISTITUTO SUPERIORE DI SANITÀ
National Centre for Rare Diseases

N° ID: 148C24_R

Relevance

Registries are key resources for increasing timely and accurate diagnosis, improving patient management, tailoring treatments, facilitating clinical trials, supporting healthcare planning and speeding up research for the benefit of rare disease patients. In rare disease registries, the data need to be collected following the 15 FAIR principles (FAIR-Findable, Accessible, Interoperable, Reusable for Humans and Computers) that have been recognized and approved in 2017 by the IRDiRC-International Rare Disease Research Consortium as a fundamental resource.

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”.

Since 2019, in the framework of the European Joint Programme on Rare Diseases (EJP RD), a European Commission funded project (Grant Agreement No 825575, 2019 – 2023) “The International Summer School on Rare Disease Registries and FAIRification of Data”.

Learning Objectives

The course focuses on the importance of high-quality FAIR rare disease registries and data as key resources to increase timely and accurate diagnosis, improve patient management, tailor treatments, facilitate clinical trials, support healthcare professionals and speed up research.

Specific aims

Upon successful completion of this course, participants will be able to:

- explain the importance of having Findable, Accessible, Interoperable, and Reusable (FAIR) data and metadata
- outline the process of making data FAIR
- propose a FAIRification process for their registry and data
- describe the steps of conceptual modelling and conversion of data to machine-readable
- identify the stakeholders of a multi-disciplinary FAIRification team, and their required skills.

Training method

There will be plenary presentations with interactive question & answer sessions between speakers and participants, work in small groups and hands-on experience sharing (Bring Your Own Data, BYOD).

PROGRAMME

Day 1
Wednesday, September 18
(CEST time)

13:30 Welcome participants & Presentation of the training
Claudio Carta
13:45 Ice-breaking session
Claudio Carta
14:15 (Un)FAIR Game
Marco Roos
14:45 (Un)FAIR Game: Lesson learned
Marco Roos
15:00 Introduction to the FAIR principles and the benefits of FAIR data
Marco Roos, Claudio Carta
15:20 Finding and accessing registries with metadata: Introduction
Martijn Kersloot, Daphne Wijnbergen
15:50 *Virtual Coffee*
16:10 Finding and accessing registries with metadata: Hands on
Martijn Kersloot, Daphne Wijnbergen
16:50 Finding and accessing registries with Metadata: Wrap up
Martijn Kersloot, Daphne Wijnbergen, Marco Roos
17:00 End of Day 1

Day 2
Thursday, September 19
(CEST Time)

13:30 Recap Day 1
Marco Roos, Claudio Carta
13:45 Drawing a conceptual model to describe your (meta)data: Introduction
César Bernabé
14:15 Drawing a conceptual model to describe your (meta)data: Hands on
César Bernabé
14:45 *Virtual Coffee*
15:15 Ontologies and Schemas: What they are and where to find them
Ronald Cornet
15:45 Interoperability and Reusability of data in registries
DEMO Querying across registries with ontologies
Mark Wilkinson, Pablo Alarcon, Alberto Cámara
16:45 FAIRification at Record Level: wrap up
Mark Wilkinson, Pablo Alarcon, Alberto Cámara
17:00 End of Day 2

Day 3
Friday, September 20
(CEST Time)

13:30	Bring Your Own (Mock) Data: your first steps in data FAIRification (Breakout Rooms, work in small groups) - Breakout Rooms, with IT Trainers Room A: **Pablo Alarcon** Room B: **Alberto Cámara** Room C: **Martijn Kersloot, Daphne Wijnbergen** Room D: **César Bernabé** - Bring Your Own (Mock) Data: Sharing experiences (open discussion with participants) **Marco Roos, Mark Wilkinson, Ronald Cornet, Martijn Kersloot, A Pablo Alarcon, Alberto Cámara, César Bernabé, Daphne Wijnbergen, Claudio Carta**
15:00	*Virtual Coffee*
15:20	Data FAIRification: Implications for “registry managers” and project planning - FAIRification for Managers - Hands on: “create your own FAIRification team and project” **Marco Roos**
16:00	Parking Lot (risposte alle domande emerse)
16:15	Evaluation of the Summer School: Satisfaction Questionnaire
16:30	Closing Remarks **Claudio Carta, Marco Roos**
16:40	Free Networking for Going FAIR **Attendees/Speakers/Trainers**
17:00	End of the Course

SPEAKERS, IT-TRAINERS

Pablo Alarcon, Centro de Biotecnología y Genómica de Plantas (CBGP, UPM-INIA), Spain
César Bernabé, Leiden University Medical Center, the Netherlands
Alberto Cámara, Centro de Biotecnología y Genómica de Plantas (CBGP, UPM INIA), Spain
Claudio Carta, National Centre for Rare Diseases, Istituto Superiore di Sanità, Italy
Ronald Cornet, Academic Medical Center, Universiteit van Amsterdam, The Netherlands
Martijn Kersloot, Academic Medical Center, Universiteit van Amsterdam, The Netherlands
Marco Roos, Leiden University Medical Center, the Netherlands
Mark Wilkinson, Centro de Biotecnología y Genómica de Plantas (CBGP, UPM INIA), Spain
Daphne Wijnbergen, Leiden University Medical Center, the Netherlands

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GENERAL INFORMATION

Venue

Online, Ms Teams Platform

Participants

The training course is open to the international research community, to clinicians, medical specialists, registry curators, database managers, healthcare professionals and rare disease patients' representatives. To ensure active participation and exchange between teaching staff and participants, a maximum of 24 attendees will be admitted.

Registration

The online registration form is available at the following link: **ONLINE REGISTRATION** until August 26, 2024. Participation in the event is free of charge.

Selection process

Selection will be made based on the participant's background, role with reference to registry activities, and involvement in ERNs and in RD projects that require the FAIRification of data.

Participants selected to attend the course will receive the confirmation email by September 2, 2024.

Applicants not contacted by email by September 9, 2024 should consider themselves as not selected and are invited to apply for the next edition of the training course.

Learning assessment

Participants will be required to complete a multiple-choice learning assessment questionnaire at the end of the training course. Moreover, a satisfaction survey will be submitted to the participants.

Attendance Certificates

At the end of the course, certificates of attendance will be issued to participants who attended 100% of the course and responded correctly to at least 75% of the learning assessment test.
No CME credits will be issued.

For any further information, please write to the course director, Dr. Claudio Carta, claudio.carta@iss.it (in Cc: linda.agresta@iss.it).

[Firma elettronica del Legale Rappresentante]

Firmato con FEQ da:
Bellantone Rocco
Domenico Alfonso
Data: 04/09/2024

Per la verifica di conformità del Contrassegno Elettronico apposto è possibile scaricare il documento, conforme al DPCM 13/11/2014, tramite l'App Honos.
Visita https://honos.land.it/ per le istruzioni.