INTERNATIONAL SUMMER SCHOOL ON RARE DISEASE REGISTRIES AND FAIRIFICATION OF DATA

27 September-1 October 2021

organised by

ISTITUTO SUPERIORE DI SANITÀ

National Centre for Rare Diseases

In collaboration with the Partners of the European Joint Programme on Rare Diseases (EJP RD), Grant Agreement No 825575

with the endorsement of the International Conference for Rare Diseases and Orphan Drugs (ICORD)

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.

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), 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/.

Learning objectives

This training course is a part of Work Package 14 (WP Leader Claudio Carta, ISS) of the EJP RD, aiming at organising residential training courses in different Countries on "Data Management and Quality Training".

In particular this training course focuses on the importance of high quality rare disease registries as key resources to increase timely and accurate diagnosis, improve patients management, tailor treatments, facilitate clinical trails, support healthcare professionals and speed up research.

Specific aims

The course consists of two training modules, each one with specific aims. During the first three days module (27-29 September 2021), 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", (30 September-1 October, 2021) 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.

At the end of the training course the participants will be able to:

- Identify the methodologies and appropriate characteristics to realize and run a rare disease registry
- Describe how to collect quality data
- List the main steps of the FAIRification process
- Describe the main features of a FAIR registry

Training method

In the first and second module there will be interactive plenary presentations and question & answers sessions between speakers and participants. Moreover, in the second training module there will be hands-on exercises and a wrap-up session.

PROGRAMME

1st Training Module "Rare Disease Registries"

DAY 1

27 September

14:00 Welcome address & Faculty & 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

17:30 End of Day 1

DAY 2

28 September

14:00 Quality of RD Registries

Yllka Kodra

14:45 Questions & Answers

15:00 Break

15:15 Ethics, GDPR and Informed Consent

Marta Tomasi

16:15 Questions & Answers

17:00 End of Day 2

DAY 3

29 September

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

15:00 The perspective of patient representatives in an ERN Registry

Johan de Graaf

15:15 Questions & Answers

15:30 Break

15:45 RegistRare: a platform for patient registries

Tiziana Grassi, Paola Torreri

16:15 Questions & Answers

16:30 Rare disease registries and how they work in real life-the EuRRECa experience

Syed Faisal Ahmed

17:15 Questions & Answers

17:45 Evaluation of the 1st training module and Satisfaction Survey

18:00 End of Day 3

19:00 Social Event

2nd Training Module "FAIRification of Data"

DAY 4

30 September

09:50 Welcome address & Presentation of the course

Domenica Taruscio

10:00 BYOD introduction

Claudio Carta, Marco Roos

10:10 FAIR Game

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

11:00 Break

11:15 Lessons Learned and the Benefits of making data FAIR

Marco Roos, Rajaram Kaliyaperumal, Bruna Dos Santos Vieira, Martijn Kersloot

12:00 The main steps of FAIRification

Marco Roos, Claudio Carta

12:10 Introduction to making data linkable and machine-readable

Mark Wilkinson

12:40 Break

13:30 Describing your registry at source for machines:

-Describing registry with "ORDO", "DCAT" & a "FAIR Data Point"

Marc Hanauer, Rajaram Kaliyaperumal

-Describing your registry access protocols

Esther van Enckevort

14:10 Hands on (Create a FAIR Data Point)

14:30 Break

14:45 Introduction to querying a FAIR Data Point

Mark Wilkinson

15:15 Hands-on Introduction to querying a FAIR Data Point

15:30 Using your FAIR metadata: exploring the FAIR Data Point via the Web

Rajaram Kaliyaperumal

15:45 Hands-on exploration of registry descriptions in a FAIR Data Point

16:00 Wrap-up; Questions & Answers

16:30 End of Day 4

DAY 5

1 October

10:00 Making Data Machine-readable & Hosting FAIR data: an example of a registry becoming FAIR

Martijn Kersloot

10:20 Hands on

10:40 Wrap-up

11:00 Using ontologies to describe data unambiguously for machines (and humans)

Ronald Cornet

11:30 Break

11:45 Introducing the EJP RD ontological model for "Common Data Elements"

Mark Wilkinson

12:00 How to apply the CDE model to your data

-Automatic conversion from tabular data in machine readable data with YARRML and Matey

Marc Wilkinson

12:30 Exploring linkable, machine readable CDEs with SPARQL: Hands-on demonstration

Marc Wilkinson

12:45 Break

14:30 Solving the game with FAIR Data & Metadata

Bruna Dos Santos Vieira, Marco Roos, Alberto Cámara, César Bernabé, Clemence le Cornec, Joeri van der Velde, Shuxin Zhang

15:00 FAIRification recap, implications for "registry managers" and project planning:

Esther van Enckevort, Marco Roos, Claudio Carta, Bruna Dos Santos Vieira

15:30 Hands on

15:50 Wrap-up

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

Erik Schultes, Barend Mons

16:20 Parking lot & Q&A next steps for your own FAIR registry

16:50 Evaluation of the 2nd training module and Satisfaction Survey

17:00 Concluding remarks

Domenica Taruscio, Marco Roos, Claudio Carta

17:10 End of the Course

SPEAKERS/TRAINERS

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

Johan de Graaf, ePAG Endo ERN, Dutch Pitutary Foundation, The Netherlands

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

Tiziana Grassi, National Centre For Rare Diseases, Istituto Superiore di Sanità, Italy

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

Diana Marinello, Azienda Ospedaliera Universitaria Pisana, Italy

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

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

Marco Roos, Leiden University Medical Centre, The Netherlands

Erik Schultes, GO FAIR, The Netherlands

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

Esther van Enckevort, University Medical Centre Groeningen, The Netherlands

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

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GENERAL INFORMATION

Venue: Online, on the Microsoft Teams Platform. The connection details will be sent by mail to the selected participants.

Participants

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 between teaching staff and participants a maximum of 30 attendees will be admitted to each training module.

Registration:

Registration is available for:

the first training module: "Rare Disease Registries", September 27-29, 2021;

the second training module: "FAIRification of Data", September 30-October 1, 2021;

the entire course: "Rare Disease Registries" and "FAIRification of data", September 27 – October 1, 2021.

The online registration form is available at the following link : ONLINE REGISTRATION until 11 July 2021.

An e-mail will be sent by 30 July 2021 to the selected participants.

Respondents who are not contacted by email should consider themselves not selected but will be kept on a waiting list until 12 September 2021.

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.

Selection of participants

A maximum of 30 attendees will be admitted to each training module. The selection process will be applied based on the participants' background, role with reference to registry activities, and involvement in ERNs.

Learning Assessment

At the end of each training module a learning assessment questionnaire and a satisfaction survey will be submitted to the participants.

Attendance Certificates

At the end of the course a certificate of attendance will be forwarded to the participants who attended 100% of the single training module or the entire course. No CME credits will be issued.

If you have any question please contact the course organiser Claudio Carta (claudio.carta@iss.it, with laura.cellai@iss.it in Cc).