International Summer School on Rare Disease Registries and FAIRification of Data

September 27 – October 1, 2021
Istituto Superiore di Sanità, Rome, Italy

GENERAL INFORMATION

Due to the force majeure situation and in order to allow 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 27 - October 1, 2021, 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/
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 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]

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".

SPECIFIC LEARNING OBJECTIVES

Registries are key resources to increase timely and accurate diagnosis, improve patient's 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 (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 (European Union) General Data Protection Regulation.

- During the second two days module "FAIRification of data", (September 30 - October 1, 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 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

LEARNING 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 and role with reference to registry activities, and on the involvement in ERNs.

REGISTRATION

Registration is possible 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.

REGISTRATION FORM 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 participants will be asked to submit a learning assessment, based on an online multiple-choice questionnaire and a satisfaction survey.

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 modules or the entire course program. No credits of Continuing Education in Medicine will be issued.
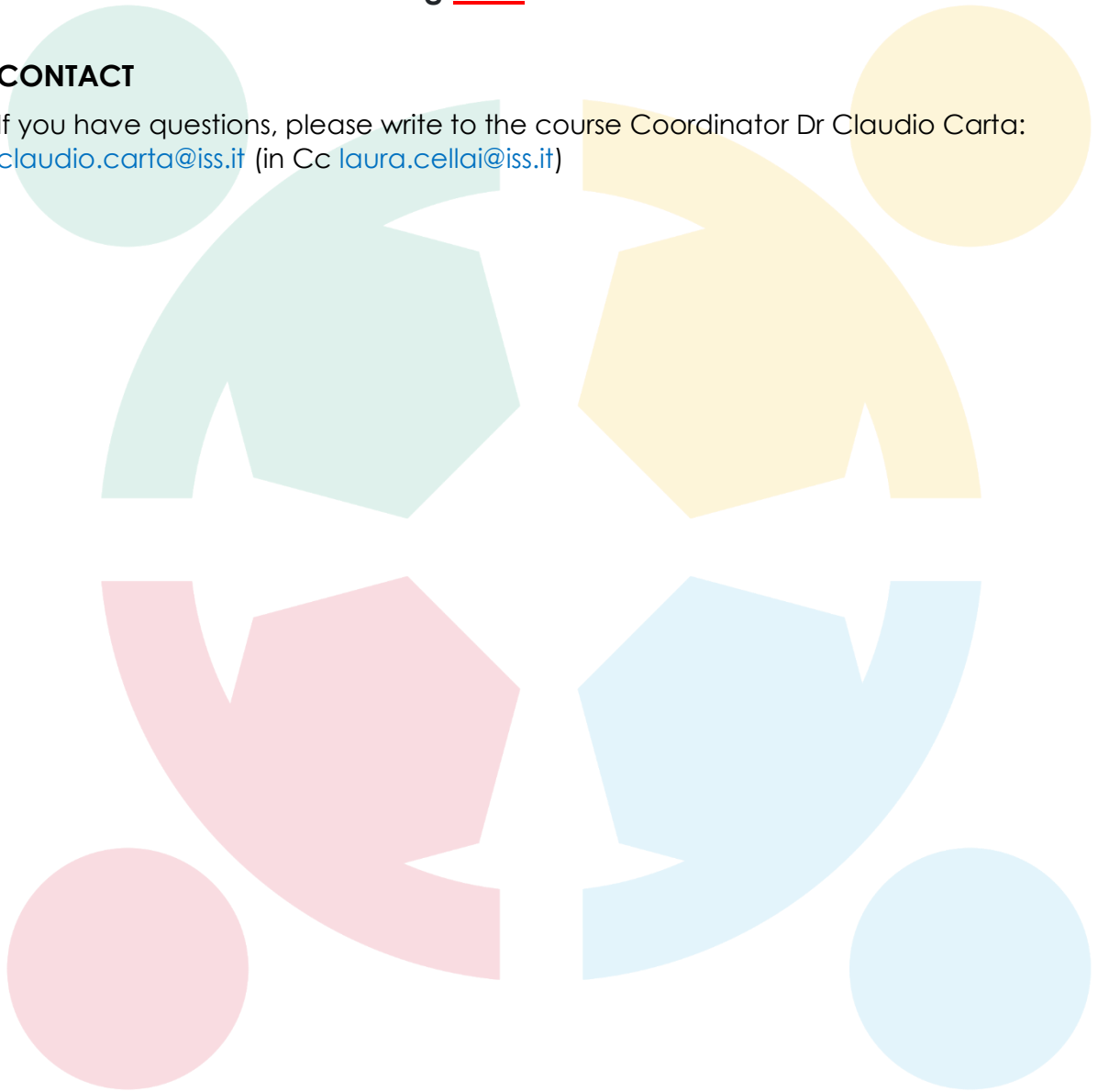
OFFICIAL LANGUAGE

English

VENUE

ONLINE

Important dates, deadlines, registration form, and further information, please visit the website at the following LINK

CONTACT

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

Programme of the Course

DAY 1   **1st Training Module, September 27, 2021**

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   **1st Training Module, September 28, 2021**

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   **1st Training Module, September 29, 2021**

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

DAY 4   **2nd Training Module, September 30, 2021**

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 Lesson 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 your 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   **2nd Training Module, October 1, 2021**

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 to machine readable data with YARRRML and Matey

Mark Wilkinson

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

demonstration

Mark 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

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; president of the Dutch Pituitary 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, Directeur technique Orphanet, INSERM, France

Rajaram Kaliyaperumal, Leiden University Medical Centre, The Netherlands

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

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