EUROPEAN JOINT PROGRAMME
RARE DISEASES

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.carta@iss.it

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
Domenica Taruscio

09:30 The European Platform on Rare Disease Registration (EU RD Platform)
Andri Papadopoulou

10:00 Introduction to Problem Based Learning and small groups
Claudio 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
Claudio Carta, Yllka Kodra, Marco Roos

14:30 Individual work Session with Facilitators; Fill in Surveys

15:00 RD registries in the Eastern EU: Situation, bottlenecks and opportunities
Rumen Stefanov

15:30 Aims, Governance & Sustainability
Joseph Giuliano, Paola 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
Yllka Kodra, Manuel Posada, Entela Xoxi

10:00 Coffee-break

10:15 Quality of Registries part 2
Yllka Kodra, Manuel Posada, Entela Xoxi

11:00 Ethics, GDPR and Informed Consent
Annalisa Landi, Marta Tomasi

12:30 Lunch

13:15 The FAIR guiding Principles
Claudio Carta, Marco Roos

13:30 A FAIR ecosystem to enable analysis, and reuse of sensitive rare disease data
M. Roos, David van Enckevort

14:00 Structuring data: Ontologies
Ronald Cornet

14:30 Roles of RD patients in registries & research - ePAGs in ERNs
Virginie Bros Facer

15:10 Experiences with RD registries: EuRRECa
Syed Faisal Ahmed

15:40 Experiences with RD registries: ERN PaedCan and the registry for very rare tumors
Gianni Bisogno

16:10 Experiences with RD registries: Unified European Registry for Inherited Metabolic Disorders registry - U-IMD
Florian Gleich

16:40 Introduction to Survey: FAIR Metrics
Claudio Carta, Annika Jacobsen

17:00 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 Introduction to Survey: Digital Environment for Training and Fill in Survey
Allegra Via

14:30 Surveys results and "QA"

14:50 Satisfaction Questionnaire

15:00 Closing 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
Claudio Carta, Marco Roos

09:40 Introduction to FAIR metrics assessment 1 and Individual hands-on
Annika Jacobsen

10:10 Coffee break

10:30 Introduction to drawing a conceptual model
Annika 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
Marco Roos

12:15 Clinical ontologies. What they are and where to look
Ronald 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
Marc Hanauer

15:00 Introduction to FAIRifier tutorial
Annika 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
Ronald Cornet, Marco Roos, Mark 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

11:45 Group report and sketch "your own data"_Experts and IT-Trainers

12:30 Individual hands-on survey on: FAIR metrics assessment 2

12:50 Individual hands-on survey on: Digital Environment for Trainings
Allegra Via

13:15 Lunch

14:00 Data FAIRification: Implications for "registry managers" and project planning
Claudio Carta, Marco Roos, David van Enckevort

14:45 Reflections on FAIR metrics 1&2
Annika Jacobsen

15:15 Evaluation of the 2nd Training Module: Satisfaction Questionnaire

15:30 Closing Remarks
Marco Roos, Domenica 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

Lieze Thielemans, Imperial College London, UK

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

FACILITATORS

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