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.cart@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.


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
# Preliminary Program of the Course

## DAY 1

**1st Training Module, September 23, 2019**

<table>
<thead>
<tr>
<th>Time</th>
<th>Activity</th>
</tr>
</thead>
<tbody>
<tr>
<td>08:45</td>
<td>Participants registration</td>
</tr>
<tr>
<td>09:10</td>
<td>Welcome address &amp; Faculty &amp; Presentation of the course by D. Taruscio</td>
</tr>
<tr>
<td>09:30</td>
<td>The European Platform on Rare Disease Registration (EU RD Platform) by A. Papadopoulou</td>
</tr>
<tr>
<td>10:00</td>
<td>Introduction to Problem Based Learning and small groups by C. Carta</td>
</tr>
<tr>
<td>10:15</td>
<td>Coffee-break</td>
</tr>
<tr>
<td>10:30</td>
<td>PROBLEM ANALYSIS (Session #1): Working in small groups with facilitators</td>
</tr>
<tr>
<td>12:30</td>
<td>Lunch</td>
</tr>
<tr>
<td>13:30</td>
<td>PROBLEM ANALYSIS (Session #2): Working in small groups with facilitators</td>
</tr>
<tr>
<td>14:00</td>
<td>Introduction to Survey: checklist for quality by C. Carta, Y. Kodra, M. Roos</td>
</tr>
<tr>
<td>14:30</td>
<td>Individual work Session with Facilitators; Fill in Surveys</td>
</tr>
<tr>
<td>15:00</td>
<td>RD registries in the Eastern EU: Situation, bottlenecks and opportunities by R. Stefanov</td>
</tr>
<tr>
<td>15:30</td>
<td>Aims, Governance &amp; Sustainability by J. Giuliani, P. Torrer</td>
</tr>
<tr>
<td>17:00</td>
<td>End of the day</td>
</tr>
</tbody>
</table>

## DAY 2

**1st Training Module, September 24, 2019**

<table>
<thead>
<tr>
<th>Time</th>
<th>Activity</th>
</tr>
</thead>
<tbody>
<tr>
<td>08:45</td>
<td>Welcome Participants</td>
</tr>
<tr>
<td>09:15</td>
<td>Quality of Registries part 1 by Y. Kodra, M. Posada, E. Xoxi</td>
</tr>
<tr>
<td>10:00</td>
<td>Coffee-break</td>
</tr>
<tr>
<td>10:15</td>
<td>Quality of Registries part 2 by Y. Kodra, M. Posada, E. Xoxi</td>
</tr>
<tr>
<td>11:00</td>
<td>Ethics, GDPR and Informed Consent by A. Landi, M. Tomasi</td>
</tr>
<tr>
<td>12:30</td>
<td>Lunch</td>
</tr>
<tr>
<td>13:15</td>
<td>The FAIR guiding Principles by C. Carta, M. Roos</td>
</tr>
<tr>
<td>13:30</td>
<td>A FAIR ecosystem to enable analysis, and reuse of sensitive rare disease data by M. Roos, D. van Enckevort</td>
</tr>
<tr>
<td>14:00</td>
<td>Structuring data: Ontologies by R. Cornet</td>
</tr>
</tbody>
</table>
14:30 Roles of RD patients in registries & research - ePAGs in ERNs_V. Bros Facer
15:10 Experiences with RD registries: EuRRECa_S.F. Ahmed
15:40 Experiences with RD registries: ERN PaedCan and the registry for very rare tumors_G. Bisogno
16:10 Experiences with RD registries: Unified European Registry for Inherited Metabolic Disorders registry - U-IMD_Florian Gleich
16:40 Introduction to Surveys: FAIR Metrics and Digital Environment for Trainings C. Carta, A. Jacobsen, A. Via
16:55 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 Surveys results and “QA”
14:45 Evaluation of the 1st Training Module: Satisfaction Questionnaire
15:00 Concluding 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 C. Carta, M. Roos
09:40 Introduction to FAIR metrics assessment 1 and Individual hands-on A. Jacobsen
10:10 Coffee break
10:30 Introduction to drawing a conceptual model_A. 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_M. Roos
12:15  Clinical ontologies. What they are and where to look_R. 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 M. Hanauer
15:00  Introduction to FAIRifier tutorial_A. 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 R. Cornet, M. Roos, M. 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
12:00  Group report and sketch “your own data”_Experts and IT-Trainers
12:45  Individual hands-on survey on: FAIR metrics assessment 2
13:00  Individual hands-on survey on: Digital Environment for Trainings
13:15  Lunch
14:00  Data FAIRification: Implications for “registry managers” and project planning C. Carta, M. Roos, D. van Enckevort
14:45  Reflections on FAIR metrics 1&2_A. Jacobsen
15:15  Evaluation of the 2nd Training Module: Satisfaction Questionnaire
15:30  Remarks and Conclusion _M. Roos, D. 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
Annikka 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
Marta Tomasi, University of Bolzano, Italy
Paola Torrieri, 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