GENERAL INFORMATION

Registries represent key resources for Rare Disease to increase timely and accurate diagnosis, improve patients management, tailor treatments, facilitate clinical trials, support healthcare planning and speed up research.

The "5th International Summer School on Rare Diseases and Orphan Drug Registries" aims to promote the establishment of Findable, Accessible, Interoperable, Reusable (FAIR) Rare Disease (RD) registries in compliance with IRDiRC and EU Recommendations. The Summer School is organized by the National Centre for Rare Diseases (ISS) in collaboration with Services and Centre for International Affairs (ISS), RD-Connect, Elixir, RD-Action, EURORDIS and endorsed by ICORD.

The Summer School will consist of plenary presentations and interactive small-group exercises, according to the Problem-Based Learning methodology.

The first part of the School (September, 18-20) will provide participants with useful tools and methodologies for establish a registry, the quality of the data collected and how a registry is turned into a FAIR resource. The second part (September 21-22) will be a hands-on experience (bring your own data), where the attendees work with FAIR data experts to make their data FAIR and linkable to other data that has been made FAIR before.

PARTICIPANTS and REGISTRATION

The Summer School is open to health professionals, researchers, medical specialists, registry curators, database managers and representatives of patients associations who are involved in or intend to establish a rare disease registry, mainly inside a European Reference Network [priority will be given to Registry Curators and IT support staff, ePAG members (or a patients/patient representatives linked with ePAGs)].

The Summer School will accept max 24-27 participants. A selection process will be applied based on the participant's background, role with reference to registry activities, and involvment in ERNs.

Registration is possible for the first part (September 18-20, 2017), the second part (September 21-22, 2017) or both.

Important dates and further information at www.iss.it/cnmr.

Online registration form is available at *https://it.surveymonkey.com/r/5th_Sum_School_BYOD* until June 30, 2017.

FEES AND COSTS Registration is free of charge. ISS does not cover any travel, subsistence and other costs incurred by the participants to attend the two events.

ASSESSMENT OF THE ACHIEVEMENT OF THE LEARNING OBJECTIVES The participants' performance will be mainly evaluated on the basis of the group work written solutions.

ATTENDANCE CERTIFICATES At the end of the Course a certificate of attendance will be handed to the participants who attended 100% of the 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 – 00161 Rome, Italy**.

CONTACTS

National Centre for Rare Diseases, Istituto Superiore di Sanità

www.iss.it/cnmr rareregistries-school@iss.it ☎ +39 06 4990 4418

Please check periodically on the CNMR-ISS website www.iss.it/cnmr any updates on the events.

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5th International Summer School

Rare Disease & Orphan Drug Registries

September 18-22, 2017

Rome, Italy

Endorsed by

ICORD
International Conference on Rare Diseases & Orphan Drugs

organised by

National Centre for Rare Diseases
CNMR

National Centre for Rare Diseases, Istituto Superiore di Sanità

in collaboration with

Services and Centre for International Affairs, Istituto Superiore di Sanità

RD-Connect elixir RD-ACTION DATA AND POLICIES FOR RARE DISEASES EURORDIS RARE DISEASES EUROPE

September 18-20, 2017

RARE DISEASE REGISTRIES

Day 1: Governance

Day 2: Quality and Legal issues

Day 3: FAIR Data and tools

OBJECTIVES

The Course will enable the participants to establish a registry on rare diseases. In particular, at the end of the course the participants will be able to:

- Describe the resources needed for the establishment of a rare disease registry;
- Describe the features of successful strategies to ensure long time sustainability of the registry, including data quality and FAIR Data

LEARNING METHOD

The training method will be Problem-Based Learning (PBL), a highly interactive and learner-centered approach where learning occurs by working in a small group assisted by a facilitator to develop a solution of a problem. Scientific articles, expert lectures, consultations and feed -back are the learning resources that will support the students in the problem solution.

SPEAKERS/TRAINERS

The teaching staff will include:

Virginie Bros-Facer, Eurordis
Claudio Carta, National Centre For Rare Diseases, Istituto Superiore di Sanità
Ronald Cornet, Academic Medical Center, Universiteit van Amsterdam
Marc Hanauer, Directeur technique Orphanet, Inserm
Victoria Hedley, Newcastle University
Yllka Kodra, National Centre For Rare Diseases, Istituto Superiore di Sanità
Simona Martin, Joint Research Centre, European Commission
Manuel Posada, Institute of Health Carlos III
Marco Roos, BioSemantics group, Leiden University Medical Centre
Domenica Taruscio, National Centre For Rare Diseases, Istituto Superiore di Sanità
David van Enckevort, University Medical Centre Groningen

September 21-22, 2017

BRING YOUR OWN DATA

Day 1: ***(i)*** **FAIRification workflow and tools: an Introduction**
(ii) **Ontologies and clinical ontologies. What they are and where to look**
(iii) **Hands on with FAIR data Trainers**

Day 2: ***(i)*** **FAIRification workflow**
(ii) **Implications for registry managers**
(iii) **Hands on with FAIR data Trainers**

OBJECTIVES

The BYOD will show practically how a rare disease registry can be a FAIR Resource. At the end of the BYOD the participants will be able to:

- Describe a FAIRification workflow for a rare disease registry
- Describe the resources needed to make a registry FAIR
- Describe some tools in order to FAIRifier a registry

LEARNING METHOD

The BYOD will consist of brief plenary introductions and practical working groups where participants see how to make their (sample) data FAIR and see how easy it becomes to answer difficult queries when a registry is FAIR.

SPEAKERS/TRAINERS

The teaching staff will include:

Claudio Carta, National Centre For Rare Diseases, Istituto Superiore di Sanità
Ronald Cornet, Academic Medical Center, Universiteit van Amsterdam
Marc Hanauer, Directeur technique Orphanet, Inserm
Annika Jacobsen BioSemantics group, Leiden University Medical Centre
Rajaram Kaliyaperumal, BioSemantics group, Leiden University Medical Centre
Marco Roos, BioSemantics group, Leiden University Medical Centre
Domenica Taruscio, National Centre For Rare Diseases, Istituto Superiore di Sanità
David van Enckevort, University Medical Centre Groningen
Mark Wilkinson, Centro de Biotecnología y Genómica de Plantas UPM-INIA (CBGP)

JUNE 30, 2017: ***Deadline for registration to "Rare Disease Registries", "Bring Your Own Data" or both!***

https://it.surveymonkey.com/r/5th_Sum_School_BYOD