

“RD-Connect BYOD Workshop to Link Rare Disease Registries”

September 29-30, 2016

National Centre for Rare Diseases, Istituto Superiore di Sanità
via Giano della Bella, 34 - Rome, Italy

Overview

The RD-Connect BYOD (Bring Your Data) Workshop to Link Rare Disease Registries will be a hands-on experience, where the attendees work with FAIR data experts to make their (sample) data FAIR and linkable to enable answering questions across resources and link to other data that has been made FAIR before.

The BYOD starts with a general introduction of the approach and an introduction of the cases by data owners.

After splitting up in working groups, one for each case, a data linkage expert explains the general SOP for making data linkable.

The SOP is subsequently customized for the case at hand in the working sessions.

The operating procedures will be evaluated at the end of the BYOD.

Thursday, 29 September 2016

- 08:30 - 09:00** Participants registration
- 09:00 - 09:15** Welcome address
Domenica Taruscio
- 09:15 - 09:30** Introduction of the workshop
Claudio Carta
- 09:30 - 10:45** Semantic Web, Linked Data and Ontologies for Beginners
Marco Roos
- 10:45 - 11:15** Coffee break
- 11:15 - 13:00** Use Case Introductions
Chairpersons: Claudio Carta and Marco Roos
- 13:00 - 13:30** Lunch
- 13:30 - 14:00** Workgroup/task/use case division
Claudio Carta and Marco Roos
- 14:00 - 15:45** Working session 1: Explanation of general SOP and tools for making data linkable at the source
Marco Roos and IT-Trainers
- 15:45 - 16:15** Break
- 16:15 - 17:00** Working session 2: attendees try to make their data linkable for CRQs following the general SOP together with data linkage experts
IT-Trainers
- 17:00 - 17:30** First impressions / progress report
Chairpersons: Claudio Carta, Marco Roos and Domenica Taruscio
- 17:30** End of the day

Friday, 30 September 2016

- 09:00 - 09:30** Answer questions from day 1
Chairpersons: Marco Roos and IT-Trainers
- 09:30 - 10:45** Working session 3: continuation from day 1
IT-Trainers
- 10:45 - 11:15** Coffee break
- 11:15 - 13:00** Working session 4: prepare demonstration of the results of the working sessions
IT-Trainers
- 13:00 - 13:30** Lunch
- 13:30 - 14:15** Use case presentations
Attendees and IT-Trainers
- 14:15 - 15:15** Evaluation of general SOP
Chairpersons: Claudio Carta, Marco Roos
- 15:15 - 16:15** What went well / Things to take a look at
Chairpersons: Claudio Carta, Marco Roos
- 16:15 - 17:00** Remarks and Conclusion
Claudio Carta, Marco Roos and Domenica Taruscio
- 17:00** End of the workshop

Data Linkage Experts

Ronald Cornet, Academic Medical Center, Universiteit van Amsterdam, Netherlands

Friederike Ehrhart, Maastricht University, Maastricht, Netherlands

Rajaram Kaliyaperumal, BioSemantics group, Human Genetics department, Leiden University Medical Centre, Leiden, Netherlands

Marco Roos, BioSemantics group, Human Genetics Department, Leiden University Medical Centre, Leiden, Netherlands

David van Enckevort, Department of Genetics, University Medical Centre Groningen, Groningen, Netherlands

Andra Waagmeester, Micelio, Ekeren, Belgium

Data Linkage "Friends"

Claudio Carta, Luca Ferrari, Sabina Gainotti, Yllka Kodra, Paola Torreri: National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

Scientific Directors

Domenica Taruscio, Director, National Centre for Rare Diseases Istituto Superiore di Sanità, Rome, Italy and RD-Connect WP2 Leader: "Databases and patient registries"

Marco Roos: BioSemantics group, Human Genetics Department, Leiden University Medical Centre, Leiden, Netherlands

Scientific Secretariat

Claudio Carta, National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

Mascha Jansen, Dutch Techcentre for Life Sciences, DTL, Amsterdam, Netherlands

Technical Secretariat

Giuseppe Bernardo, Luana Bernardo, Patrizia Crialese, Federico De Paulis, Stefano Diemoz,

Andrea Vittozzi: National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

GENERAL INFORMATION

PARTICIPANTS AND REGISTRATION

The Workshop is open to health professionals, researchers, medical specialists, medical students, registries curators, database managers and representatives of patient associations, who are involved in or intend to establish a rare disease patient registry.

A selection process will be applied based on the participant's background and role with reference to registry activities.

Online registration form, important dates and further information at www.iss.it/cnmr.

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 Workshop.

ATTENDANCE CERTIFICATES

At the end of the Workshop a certificate of attendance will be handed to the participants who attended 100% of the program.

OFFICIAL LANGUAGE

English.

VENUE

Aula Rossi, Istituto Superiore di Sanità, via Giano della Bella, 34 – 00161 Rome, Italy.

CONTACTS

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