RD-CONNECT WORKSHOP DATA LINKAGE AND ONTOLOGIES

September 24-25 2015, Rome, Italy

DAY1

8.30 Registration and pre-test

8.45 Welcome and presentation of the workshop objectives (***D. Taruscio***)

9:00 Data linkage for rare disease registries (***M. Roos, C. Carta***)

(i) Data linkage and ontologies (***M. Roos***)

9:45 Usefulness of ontologies for RDs (***E. López***)

10:00 *Coffee Break*

10:30 The Experimental Factor Ontology-EFO (***S. Sarntivijai***) (Remote)

10:50 Using EFO with the Human Phenotype Ontology (***S. Sarntivijai***) (Remote)

11:10 Orphanet Rare Disease Ontology, ORDO (***M. Hanauer***)

11:50 Demonstrations and questions

12:15 Data linkage tutorial (***M. Roos***)

12:30 *Lunch*

13:30 Tutorial with hands-on simulation (***Data linkage experts***)

(i) How to collect and harmonize data?

(ii) How to link data?

16:30 *End of the day*

DAY2

9:00 Wikidata a linked data platform (***A. Waagmeester***)

9:30 Post Test

10:00 *Coffee Break*

10:30 Tutorial with hands-on simulation (***Data linkage experts***)

(i) How to use linked data?

(ii) How to use existing linked data resources?

(iii) Nanopublications

12:30 *Lunch*

13:30 Evaluations, discussion, parking lot, remarks and conclusion

15:00 Free hands on

17:00 *End of workshop*

***Speakers, data linkage experts and "friends"*:**

- Luiz Bonino, semantic interoperability, Dutch Techcentre for Life Sciences, DTL, Amsterdam
- Claudio Carta, National Center for Rare Diseases, Istituto Superiore di Sanità, Roma,
- Stefano Diemoz, National Center for Rare Diseases, Istituto Superiore di Sanità, Roma
- Sabina Gainotti, National Center for Rare Diseases, Istituto Superiore di Sanità, Roma
- Marc Hanauer, Directeur technique / Chief technology, officer ORPHANET - INSERM US14
- Rajaram Kaliyaperumal, BioSemantics group, Human Genetics department, Leiden University Medical Centre
- Estrella Lopez, Institute of Rare Diseases Research (I I E R - I S C I I I), Madrid
- Marco Roos, BioSemantics group, Human Genetics department, Leiden University Medical Centre
- Sirarat Sarntivijai, Bioinformatician – Ontologist, European Bioinformatics Institute (EMBL-EBI)
- Domenica Taruscio, Director, National Center for Rare Diseases Istituto Superiore di Sanità, Roma
- Mark Thompson, BioSemantics group, Human Genetics department, Leiden University Medical Centre
- Paola Torreri, National Center for Rare Diseases, Istituto Superiore di Sanità, Roma
- Andrea Vittozzi, National Center for Rare Diseases Istituto Superiore di Sanità, Roma
- Andra Waagmeester, Micelio, Ekeren, Belgium