

## Language

The official language will be English.

## How much will it cost?

Registration is free of charge. The School does not cover any travel, subsistence and other costs incurred by the participants to attend the School. Specific support will be provided to EUROPLAN (<http://www.europlanproject.eu/>) Partners.

## Important communication

Given the interactive nature of the Course methodology, admitted participants wishing to obtain the Certificate of participation are expected to attend at least 90% of the Course scheduled hours. Sporadic presences are not allowed.

For more information visit the website  
[www.iss.it/cnmr](http://www.iss.it/cnmr)



### Course Director

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### Secretariat and Contacts

#### Scientific Secretariat

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## II International Summer School RARE DISEASE AND ORPHAN DRUG REGISTRIES

September 15-19, 2014

### Venue:

Casa dell'Aviatore  
Viale dell'Università, 20  
00185 Rome (Italy)

Organised by the  
National Centre for Rare Diseases  
Istituto Superiore di Sanità (ISS)



## Monday, 15 September

- 8.30 *Registration and pre-test*
- 9.15 Welcome and presentation of the course objectives
- 9.30 Introduction of participants and presentation of their activities (*D. Taruscio*)
- 10.30 *Coffee Break*
- 11.00 Registry types, Aims, Building a registry, Management, Sustainability (*L. Korngut*)
- 12:00 Epidemiological study design, data sources of population study variables(*Y. Kodra*)
- 13.00 *Lunch*
- 14.00 Cooperative learning on matters dealt with in the morning (CNMR, in collaboration with the speakers)  
*14:00 - 1<sup>st</sup> phase: Small groups*  
*15:00 - 2<sup>nd</sup> phase: Larger groups*  
*16:10 - 3<sup>rd</sup> phase: Plenary session*
- 17.00 *Adjourn*

## Tuesday, 16 September

- 8.30 Reference Standards and Catalogues (*P. Landais*)
- 10.30 *Coffee break*
- 11.00 Coding of rare disease (*P. Landais*)
- 12.00 Omics and links with biobanks and registries (*M. Calissano*)
- 13.00 *Lunch*
- 14.00 Cooperative learning on matters dealt with in the morning (CNMR, in collaboration with the speakers)  
*14:00 - 1<sup>st</sup> phase: Small groups*  
*15:00 - 2<sup>nd</sup> phase: Larger groups*  
*16:10 - 3<sup>rd</sup> phase: Plenary session*
- 17.00 *Adjourn*

## Wednesday, 17 September

- 8.30 Epidemiologic analyses, confounders, sample stratification (*F. Bianchi, M. Santoro*)
- 10.30 *Coffee break*
- 11.00 Personal Data Protection; Informed Consent; Registry termination (*S. Gainotti*)
- 13.00 *Lunch*
- 14.00 Cooperative learning on matters dealt with in the morning (CNMR, in collaboration with the speakers)  
*14:00 - 1<sup>st</sup> phase: Small groups*  
*15:00 - 2<sup>nd</sup> phase: Larger groups*  
*16:10 - 3<sup>rd</sup> phase: Plenary session*
- 17.00 *Adjourn*

## Thursday, 18 September

- 8.30 Patient unique identifier (*M. Posada*)
- 10.30 *Coffee break*
- 11.00 Quality assurance (*M. Posada*)
- 13.00 *Lunch*
- 14.00 Cooperative learning on matters dealt with in the morning (CNMR, in collaboration with the speakers)  
*14:00 - 1<sup>st</sup> phase: Small groups*  
*15:00 - 2<sup>nd</sup> phase: Larger groups*  
*16:10 - 3<sup>rd</sup> phase: Plenary session*
- 17.00 *Adjourn*

## Friday, 19 September

- 8.30 The application of the Human Phenotype Ontology (*M. Haendel*)
- 10.00 *Coffee break*
- 10.15 Data reporting and dissemination (*E. Bravo, P. De Castro*)
- 11.45 Discussion, evaluation questionnaire and post-test
- 12.30 Certificate delivery and greetings
- 13.00 *Lunch and Course closure*

## **Objectives and Methods**

*The II International Summer School on Rare Disease and Orphan Drug Registries will take participants through the main concepts and practical steps that must be undertaken in the establishment and management of a rare disease registry. The programme builds on the challenges posed by the evolving technological innovations, new data collection and sharing possibilities, legal requirements and security needs. The School will consist of frontal presentations followed by Cooperative Learning (CL) sessions, where participants work together to maximize their own and each other's learning potential. Basic elements of CL are positive interdependence, individual accountability, face-to-face interaction, group processing and exercise of small-group interpersonal skills.*

## **Speakers**

**Fabrizio Bianchi** - National Council of Research, Pisa, Italy  
**Elena Bravo** - Dep. of Cell Biology & Neuroscienze, ISS, Rome, Italy  
**Mattia Calissano** - MRC Centre for Neuromuscular Diseases at Newcastle, UK  
**Paola De Castro** – Publishing Unit, ISS, Rome, Italy  
**Sabina Gainotti** - National Centre for Rare Diseases, ISS, Rome, Italy  
**Melissa Haendel** - Oregon Health & Science University, USA  
**Yllka Kodra** - National Centre for Rare Diseases, ISS, Rome, Italy  
**Lawrence Korngut** - University of Calgary, Canada  
**Paul Landais** – Université de Montpellier, France  
**Manuel Posada** - Institute of Health Carlos III, Madrid, Spain  
**Michele Santoro** - National Council of Research, Pisa, Italy  
**Domenica Taruscio** - National Centre for Rare Diseases, ISS, Rome, Italy