

Course Director

Domenica TARUSCIO

Director, National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome

Didactic Coordinators

Giovanni DE VIRGILIO

Director, External Relations Office, Istituto Superiore di Sanità, Rome

Amalia Egle GENTILE

National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome

Scientific Secretariat

Claudio CARTA (coordinator), **Yllka KODRA**, **Adele ROCCHETTI**, **Paolo SALERNO**, **Paola TORRERI**
National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome

Daniela COCLITE, **Alice J. FAUCI**, **Alfonso MAZZACCARA**, **Antonello NAPOLETANO**
External Relations Office, Istituto Superiore di Sanità, Rome

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

CONTACTS

National Centre for Rare Diseases, Istituto Superiore di Sanità

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



4th International Summer School

Rare Disease & Orphan Drug Registries

GENERAL INFORMATION

OBJECTIVES

The Course will enable the participants to establish a patient registry for clinical research on rare diseases and related treatments. In particular, at the end of the course the participants will be able to:

- Describe the resources needed for the establishment of a clinical research registry;
- Describe the features of successful strategies to ensure long time sustainability of the registry, including registry purposes, data use, data sharing, dissemination activities, and the role of patient organisations;
- Describe the main steps for planning a registry and keeping control on its operations.

LEARNING METHOD

The training method will be Problem-Based Learning (PBL), a highly interactive and learner-centred 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.

PARTICIPANTS AND REGISTRATION

The Course 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 Course.

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.

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

September 26-28, 2016

Rome, Italy

PRELIMINARY PROGRAMME

endorsed by



organised by



National Centre for Rare Diseases, Istituto Superiore di Sanità

in collaboration with

External Relations Office, Istituto Superiore di Sanità

PROGRAMME

PRELIMINARY PROGRAMME

MONDAY

September 26, 2016

- 08:30 *Participants registration*
- 09:30 **Welcome and presentation of the course**
D. Taruscio
- 09:45 **Introduction to Problem-Based Learning**
G. De Virgilio
- 10:00 **PROBLEM ANALYSIS** (Session #1)_Working in small groups with facilitators
- 11:30 *Coffee-break*
- 11:45 **PROBLEM ANALYSIS** (Session #2)_Working in small groups with facilitators
- 13:00 *Lunch*
- 14:00 **Introduction to learning resources**
C. Carta, A.E. Gentile
- 14:15 **INDIVIDUAL WORK SESSION**
- 16:45 **MONITORING MEETING** with facilitators
- 17:30 *End of the day*

TUESDAY

September 27, 2016

- 08:30 **The European Platform for Rare Disease Registries**_ *S. Martin*
- 09:00 **The Pillars**_INTERACTIVE SESSION #1 with *J. Giuliano, P. Landais and D. Taruscio*
Aims, Sustainability, and Governance
- 10:30 *Coffee-break*
- 10:45 **Modern techniques and tools for data sharing**_INTERACTIVE SESSION #2 with *M. Roos*
Ontology, Interoperability and Linked Data
- 12:45 *Lunch*
- 13:30 **Strategies for Data Quality**_INTERACTIVE SESSION #3 with *Y. Kodra, M. Posada, M. Santoro*
Coding, Patient Unique Identifier, Quality assurance and Data elaboration essentials
- 15:00 *Coffee-break*
- 15:15 **Informed consent, dissemination and publication**_INTERACTIVE SESSION #4
with *P. De Castro, S. Gainotti*
Informed consent, Data reporting and dissemination
- 16:45 **Experiences with building and managing a registry**_INTERACTIVE SESSION #5
with *M. Crimi, M. Salvatore*, P. Torreri*
International disease registry, National registry, Patient driven registry
- 17:30 *End of the day*

WEDNESDAY

September 28, 2016

- 08:30 **Patients needs & experiences**_INTERACTIVE SESSION #6
with *F. De Angelis, N. Spinelli Casacchia**
Needs, strengths and weaknesses, Patients driven registry experience
- 10:00 *Coffee-break*
- 10:45 **PROBLEM SOLUTION**_Working in small groups with facilitators
- 13:30 *Lunch*
- 14:30 **PRESENTATION of GROUP SOLUTIONS AND FEEDBACK FROM PEERS AND EXPERTS**
- 16:30 *Satisfaction questionnaire*
- 17:00 **Concluding remarks**
D. Taruscio
- 17:30 *End of the course*

SPEAKERS

- Claudio Carta** - National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy
- Marco Crimi** - Ring 14 International Onlus, Reggio Emilia, Italy
- Fernanda De Angelis** - PARENT Project Italia, Rome, Italy
- Paola De Castro** - Publishing Unit, Istituto Superiore di Sanità, Rome, Italy
- Giovanni De Virgilio** - External Relations Office, Istituto Superiore di Sanità, Rome, Italy
- Sabina Gainotti** - National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy
- Amalia Egle Gentile** - National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy
- Joseph Giuliano** - Global Medical Operations & Patient Registries, Amicus Therapeutics, New Jersey, USA
- Yllka Kodra** - National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy
- Paul Landais** - University of Montpellier, Montpellier, France
- Simona Martin** - European Commission's Joint Research Centre, Ispra, Italy
- Manuel Posada** - Institute of Health Carlos III, Madrid, Spain
- Marco Roos** - Human Genetics Department, Leiden University Medical Centre, Leiden, Netherlands
- Marco Salvatore*** - National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy
- Michele Santoro** - Institute of Clinical Physiology, National Council of Research, Pisa, Italy
- Nicola Spinelli Casacchia*** - UNIAMO-Federazione Italiana Malattie Rare Onlus, Rome, Italy
- Domenica Taruscio** - National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy
- Paola Torreri** - National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

PBL facilitators

- Fabio Candura** - National Blood Centre, Istituto Superiore di Sanità, Rome, Italy
- Marta De Santis** - National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy
- Paolo Salerno** - National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy
- Lorenza Scotti** - External Relations Office, Istituto Superiore di Sanità, Rome, Italy

*to be confirmed