

## Language

The official language will be English.

## Methods of Assessment

There will be no formal examination. A Certificate of Attendance will be awarded only to those completing the full course.

## How much will it cost?

The course is available free of charge. Please take note that the Istituto Superiore di Sanità will provide refreshments (tea, coffee and biscuits) and lunch but will not pay for travel and accommodation expenses.

## How to apply

The application will be available at: [www.iss.it/cnmr](http://www.iss.it/cnmr)

## Important dates

24 April 2013 – application is open  
30 July 2013 – deadline for application submission  
1 August 2013 – confirmation of admittance  
16-20 September 2013 – course

For more information visit the website

[www.iss.it/cnmr](http://www.iss.it/cnmr)



### Course Director

**Domenica Taruscio**  
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### Contact/Info

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## International Summer School RARE DISEASE AND ORPHAN DRUG REGISTRIES

September 16-20, 2013

Istituto Superiore di Sanità  
Aula Marotta  
Viale Regina Elena, 299  
Rome (Italy)

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



## Monday, 16 September

8.30	Registration and pre-test
9.00	Welcome and presentation of the course objectives <i>Domenica Taruscio</i>
9.30	The role of registries in epidemiological, clinical and genetic research on rare diseases: case examples: 1)EUROCAT . <i>Fabrizio Bianchi</i>
10.00	2) <i>Facio Scapulo Humeral Dystrophy Registry. Rossella Tupler</i>
10.30	3)Patient driven registries: Duchenne and Becker registry. <i>Filippo Buccella</i>
11.00	Break
11:30	Defining the aim(s) of a registry and addressing study design, objectives and methods. <i>Paul Landais</i>
13.00	Lunch
14.00	Small group exercise
17.00	Adjourn

## Tuesday, 17 September

9.00	Defining the data elements, data-sources and variables. <i>Matthew Bellgard</i>
11.00	Break
11.30	Towards National and EU registry platforms <i>Domenica Taruscio, Luciano Vittozzi</i>
13.30	Lunch
14.30	Small group exercise
16.30	Adjourn

## Wednesday, 18 September

9.00	The main dimensions of quality. <i>Manuel Posada</i>
11.00	Break
11.30	The quality of registries in practice. <i>Yllka Kodra</i>
13.30	Lunch
14.30	Small group exercise
16.30	Adjourn

## Thursday, 19 September

9.00	Registry sustainability: funding, operation, termination. <i>Lawrence Korngut</i>
11.00	Break
11.30	Epirare survey on expired registries. <i>Emanuela Mollo</i>
12:30	The culture of data sharing. <i>Sabina Gainotti</i>
13.30	Lunch
14.30	Small group exercise
16.30	Adjourn

## Friday, 20 September

9.00	Impact of registries on researcher and patient communities. <i>Lawrence Korngut</i>
11.00	Break
11.30	Ethical and legal issues -data protection directive and the ethics of solidarity. <i>David Townend</i>
13:00	Evaluation questionnaire and post-test
14.00	Certificate delivery and greetings
14:00	End of course

## Description

The course will take the participants through the main concepts and practical steps that must be undertaken in the establishment and management of a rare disease registry to ensure its usefulness, soundness and sustainability.

The course will provide basic notions on the methodology of observational studies with a view to the specificity of rare disease registries, on the selection of data elements with a focus on the interoperability of rare disease registries, on quality assurance and on the technical and legal tools that must be adopted to protect patients' data confidentiality.

The course will consist of frontal presentations followed by small group exercises.

## Eligibility requirements

The course is open to the following profiles:

- professionals involved in the health care of rare disease patients;
- professionals working in public health institutions in the surveillance of rare diseases;
- persons working or volunteering for a patients' association;
- professionals working in the pharmaceutical industry and especially in the development or surveillance of orphan medicinal products.

Priority will be given to participants:

- 1) already involved in the conduction and management of a rare disease registry or patient data collection;
- 2) with a concrete intention to establish a new registry for a rare disease.

## Speakers / Tutors

*Matthew Bellgard – Murdoch University, Perth, Australia*  
*Fabrizio Bianchi – National Council of Research, Pisa, Italy*  
*Filippo Buccella – Parent Project Onlus, Italy*  
*Sabina Gainotti – National Centre for Rare Diseases, ISS, Rome, Italy*  
*Yllka Kodra – National Centre for Rare Diseases, ISS, Rome, Italy*  
*Lawrence Korngut – University of Calgary, Canada*  
*Paul Landais – University of Montpellier, France*  
*Emanuela Mollo – National Centre for Rare Diseases, ISS, Rome, IT*  
*Manuel Posada - Institute of Health Carlos III, Madrid, Spain*  
*Domenica Taruscio – National Centre for Rare Diseases,ISS, Rome,IT*  
*David Townend - University of Maastricht, Netherlands*  
*Rossella Tupler – University of Modena and Reggio Emilia, Italy*  
*Luciano Vittozzi – National Centre for Rare Diseases, ISS, Rome, Italy*