



**International Course**  
**Training on strategies to foster**  
**solutions of undiagnosed rare**  
**disease cases**

**27 – 29 April 2020**  
**Istituto Superiore di Sanità, Rome, Italy**

## GENERAL INFORMATION

Due to the force majeure situation, the international travel policies associated with COVID-19 and extraordinary measures to limit the spread of the virus, the Italian health authorities (including the Istituto Superiore di Sanità, ISS), recommend adopting a precaution approach and cancel the planned trips and events. In order to ensure the correct progress of the planned tasks of EJP RD, ISS ensures that the international course "Training on strategies to foster solutions of undiagnosed rare disease cases", 27-29 April 2020, ISS, Rome will be held ONLINE.

For this online edition will be observed the General Data Protection Regulation (EU Regulation 2016/679) (GDPR)

Participants who have been selected to attend the course will be informed of the means and tools for using the online version of the training course

## Programme of the Course

### DAY 1

April 27, 2020

#### Morning session

- 10:30 Welcome address & Faculty & Presentation of the course  
Domenica Taruscio
- 10:45 Presentation of the European Joint Programme on Rare Diseases  
Domenica Taruscio, Claudio Carta
- 11:00 Use Case from Telethon  
Vincenzo Nigro
- 11:30 Use Case from Institute of Rare Diseases Research & CIBERER, ISCIII  
Estrella Lopez
- 12:00 Questions & Answers Round Table between speakers & participants
- 13:00 End of morning session

#### Afternoon session

- 16:00 Use Cases from Undiagnosed Diseases Network International, part 1  
David Adams
- 16:20 Questions & Answers between speakers & participants
- 16:40 Use Cases from Undiagnosed Diseases Network International, part 2  
David Adams
- 17:00 Questions & Answers Round of Table between speakers & participants
- 17:30 End of the day 1

**DAY 2****April 28, 2020****Morning session**

10:45 Introduction of Day 2

Domenica Taruscio

11:00 Use Cases from Undiagnosed Diseases Network International

Marco Castori

11:30 Use Cases from Undiagnosed Diseases Network International

Roberta Fenoglio

12:00 Questions &amp; Answers Round of Table between speakers &amp; participants

13:00 End of morning session

**Afternoon session**

16:00 Undiagnosed Diseases Network International,

Domenica Taruscio

16:15 Undiagnosed Diseases Network Italy

Marco Salvatore

16:30 Solve-RD, Solving the unsolved Rare Diseases

Holm Graessner

17:00 Questions &amp; Answers Round of Table between speakers and participants

17:30 End of Day 2

**DAY 3****April 29, 2020****Morning session**

09:45 Introduction of Day 3

Domenica Taruscio

10:00 Tool Demo and Introduction from Institut IMAGINE

Patrick Nitschké

10:30 Hands-on exercise

11:00 Discussion between speakers &amp; participants

11:30 Solving RDs with the RD-Connect Genome-Phenome Analysis Platform

Sergi Beltran, Leslie Matalonga

12:00 Hands-on exercise

12:30 Discussion between speakers &amp; participants

13:00 End of morning session

### **Afternoon session**

- 15:30 Tool Demo on Phenome Central (or PhenoTips)  
Michael Brudno, Marta Girdea
- 16:00 Hands-on exercise
- 16:30 Discussion between speakers & participants
- 17:00 Closing remarks
- 17:30 End of the Course

### **SPEAKERS and TRAINERS**

**David Adams**, National Institutes of Health, NIH, Bethesda, MD, USA

**Sergi Beltran**, Centre Nacional d'Anàlisi Genòmica, CNAG-CRG, Barcelona, Spain

**Michael Brudno** University of Toronto, Canada

**Claudio Carta**, National Centre for Rare Diseases, ISS, Rome, Italy

**Marco Castori**, Casa Sollievo della Sofferenza, San Giovanni Rotondo, Italy

**Roberta Fenoglio**, University of Turin, Turin, Italy

**Marta Girdea**, The Hospital for Sick Children, Toronto, Canada

**Holm Graessner**, Eberhard Karls Universität Tübingen, EKUT, Tübingen, Germany

**Estrella Lopez**, Institute of Rare Diseases Research & CIBERER, ISCIII, Madrid; Spain

**Leslie Matalonga**, Centre Nacional d'Anàlisi Genòmica, CNAG-CRG, Barcelona, Spain

**Vincenzo Nigro**, Tigem and University of Napoli, Naples, Italy

**Patrick Nitschké**, Institute IMAGINE, Institute of Genetic Diseases, Paris, France

**Marco Salvatore**, National Centre for Rare Diseases, ISS, Rome, Italy,

**Domenica Taruscio**, National Centre for Rare Diseases, ISS, Rome, Italy

### **COURSE DIRECTOR**

**Domenica Taruscio**, National Centre for Rare Diseases, ISS, Rome, Italy

### **SCIENTIFIC SECRETARIAT**

**Claudio Carta** (Coordinator), **Marco Salvatore**,  
National Centre for Rare Diseases, ISS, Rome, Italy  
email: [claudio.cart@iss.it](mailto:claudio.cart@iss.it)

**Lorenza Scotti**, Research Coordination and Support, ISS, Rome, Italy

## **ORGANIZING SECRETARIAT**

*Linda Agresta, Daniela Bernardo, Laura Lee Cellai, Patrizia Crialesi, Stefano Diemoz,*  
National Centre for Rare Diseases, ISS, Rome, Italy  
email: [laura.cellai@iss.it](mailto:laura.cellai@iss.it)

## **OFFICIAL LANGUAGE**

Workshops will run in English.

## **VENUE**

ONLINE