



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

**12-14 April 2021**  
**Istituto Superiore di Sanità, Rome, Italy**

**ONLINE**

## 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 allow 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", 12-14 April 2021, ISS, Rome will be held ONLINE

### Programme of the Course

#### DAY 1

April 12, 2021

#### Morning session

- 9:30 Welcome address  
Domenica Taruscio
- 9.45 Overview of the European Joint Program on Rare Diseases  
Claudio Carta
- 10.00 The Undiagnosed Diseases Network International and the Italian National Undiagnosed Network coordinated by ISS  
Domenica Taruscio, Marco Salvatore
- 10:15 Solved and Unsolved Use Cases from ACURARE-ACIBADEM University, Rare Diseases and Orphan Drugs Application and Research Center, Turkey  
Ugur Ozbek, Yasemin Alanay,
- 11:00 Questions & Answers
- 11:15 *Break*
- 11:30 ACUgen – in house Exome and Genome analysis pipeline  
Özkan Özdemir
- 11:45 Hands-on exercise
- 12:30 End of morning session

#### Afternoon session

- 15:00 Use Cases from Solve-RD, Solving the unsolved Rare Diseases  
Richarda de Voer

15:45 Questions & Answers

16:00 *Break*

16:15 Solving RDs with the RD-Connect Genome-Phenome Analysis Platform

Sergi Beltran, Leslie Matalonga

16:30 Hands-on exercise

17:30 End of the day 1

## DAY 2

April 13, 2021

### Morning session

10:30 Use Cases from Instytut Pomnik- Centrum Zdrowia Dziecka, Poland

Piotr Socha

11:15 Questions & Answers

11:25 *Break*

11:35 Use Cases from Instytut Pomnik- Centrum Zdrowia Dziecka, Poland

Diana Kaminska

12:20 Questions & Answers

12:30 End of morning session

### Afternoon session

15:00 Use Cases from the "Undiagnosed Patients Program", Ospedale Pediatrico Bambino Gesù, Italy

Marco Tartaglia

15:45 Questions & Answers

16:00 Use Cases from the "Undiagnosed Patients Program", Ospedale Pediatrico Bambino Gesù, Italy

Marco Tartaglia

16:45 Questions & Answers

17:00 End of the day 2

## DAY 3

April 14, 2021

### Morning session

10:15 Use Cases from Aix Marseille Université, France

Christophe Béroud

11:00 Questions & Answers

11:20 *Break*

11:30 Tool Demo Introduction from Aix Marseille Université, France

Christophe Béroud

11:45 Hands-on exercise

12:30 End of morning session

### **Afternoon session**

15:00 Use Cases from Sir Ganga Ram Hospital, India

Ratna Dua Puri

15:45 Questions & Answers

16:00 *Break*

16:15 Use Cases from Sir Ganga Ram Hospital, India

Ratna Dua Puri

17:00 Questions & Answers

17:15 Closing remarks

17:30 End of the Course

### **SPEAKERS and TRAINERS**

**Yasemin Alanay** Acibadem Mehmet Ali Aydinlar University, Rare Disease and Orphan Drug Research and Application Center and School of Medicine, ACU/ACURARE, Istanbul, Turkey

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

**Christophe Béroud**, Human Genetics of Aix-Marseille University, AMU, Marseille, France

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

**Richarda de Voer**, Radboud University Medical Centre (Radboudumc), Nijmegen, Netherlands

**Diana Kaminska** Instytut Pomnik- Centrum Zdrowia Dziecka, Warsaw, Poland

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**Ratna Dua Puri**, Institute of Medical Genetics and Genomics, Sir Ganga Ram Hospital, New Delhi, India

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

**Piotr Socha**, Instytut Pomnik- Centrum Zdrowia Dziecka, Warsaw, Poland

**Marco Tartaglia**, Genetics and Rare Diseases Research Division, Ospedale Pediatrico Bambino Gesù, Rome, Italy

**Domenica Taruscio**, National Centre for Rare Diseases, CNMR, 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.carta@iss.it](mailto:claudio.carta@iss.it)

#### **ORGANIZING SECRETARIAT**

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**OFFICIAL LANGUAGE** Workshops will run in English.

**VENUE** ONLINE