



## **TRAINING ON STRATEGIES TO FOSTER SOLUTIONS OF UNDIAGNOSED RARE DISEASE CASES**

**12-14 April 2021**

organized by

**ISTITUTO SUPERIORE DI SANITÀ**

National Centre for Rare Diseases

In collaboration with the Partners of the European Joint Programme on Rare Diseases (EJP RD), Grant Agreement No 825575

### **PROGRAMME**

#### **Day 1, Monday 12 April 2021**

##### **Morning session**

- 09.30 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  
Uğur Özbek, 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 day 1

## **Day 2, Tuesday 13 April 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 Kamińska

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 day 2

## **Day 3, Wednesday 14 April 2021**

### **Morning session**

- 10:15 Use Cases from Aix Marseille Université, France  
Christophe Bérout
- 11:00 Questions & Answers
- 11:20 *Break*
- 11:30 Tool Demo Introduction from Aix Marseille Université, France  
Christophe Bérout
- 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 Bérout**, 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

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

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

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

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

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

**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

Tel. + 39 0649904016; email: [domenica.taruscio@iss.it](mailto:domenica.taruscio@iss.it)

#### **Scientific Secretariat**

*Claudio Carta (Coordinator), Marco Salvatore*, National Centre for Rare Diseases, ISS, Rome, Italy

Tel. + 0039 0649904369; email: [claudio.cart@iss.it](mailto:claudio.cart@iss.it)

#### **Organizing Secretariat**

*Laura Lee Cellai, Linda Agresta, Daniela Bernardo, Patrizia Crialesse, Stefano Diemoz*, National Centre for Rare Diseases, ISS, Rome, Italy

Tel + 0039 0649904422; email: [laura.cellai@iss.it](mailto:laura.cellai@iss.it)

**Venue:** online