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

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COURSE DIRECTOR

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SCIENTIFIC SECRETARIAT

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ORGANIZING SECRETARIAT

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OFFICIAL LANGUAGE Workshops will run in English.

VENUE ONLINE