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 & Answers Round of Table between speakers & 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 & 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 & 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 & 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
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OFFICIAL LANGUAGE

Workshops will run in English.

VENUE

ONLINE