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

3-5 April 2023
Istituto Superiore di Sanità, Rome, Italy

Endorsed by

This Training has received funding from the European Union’s Horizon 2020 Research and Innovation Programme under Grant Agreement No 825575 - European Joint Programme on Rare Diseases
GENERAL INFORMATION
INTRODUCTION AND OBJECTIVES

The International Course Training on strategies to foster solutions of undiagnosed rare disease cases is part of a series of training activities proposed by the European Joint Programme on Rare Diseases (EJP RD). EJP RD is a European Commission funded project (grant agreement No 825575, 2019 – 2023) with the goal “to create a comprehensive, sustainable ecosystem allowing a virtuous circle between research, care and medical innovation”. For more information about the EJP RD, see https://www.ejprarediseases.org/

This International course is part of WP14 of the EJP RD, “Training on Data Management & Quality”; Task 14.3 “Training on strategies to foster solutions of undiagnosed rare disease cases”. WP and Task Leader: Dr. Claudio Carta, ISS.

Course Director: Dr. Claudio Carta, ISS.

The Course is made up of 3 days of residential training organized by ISS in close collaboration with EJP RD Task Partners [EKUT, LBG (LBI-RUD), ACU/ACURARE, ISCIII, INSERM (AMU), FTELE, UMCG, IMAGINE, CNAG-CRG, IPCZD (CMHI)].

The course is endorsed by the International Collaboration on Rare Diseases and Orphan Drugs, [ICORD] and Undiagnosed Diseases Network International [UDNI].

Several initiatives have been undertaken at national and international level for undiagnosed rare diseases aimed at identifying clinical pathways and innovative methods to reach diagnosis. This course will illustrate methodologies and tools already used internationally and will provide participants with useful examples for the resolution of undiagnosed cases.

The course will provide participants, through the presentation of sample use cases that have long eluded diagnosis, with useful tools, instruments and knowledge on novel strategies to foster solutions of undiagnosed RD cases. Moreover, the course will facilitate networking among professionals involved in undiagnosed rare conditions.

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ACU/ACURARE Acibadem Universitesi, Istanbul, Turkey
CNAG-CRG Fundacio Centre de Regulacio Genomica, Barcelona, Spain
EKUT Eberhard Karls Universität Tübingen, Tübingen, Germany
FTELE Fondazione Telethon, Milan, Italy
ICORD International Collaboration on Rare Diseases and Orphan Drugs
IMAGINE Imagine Institut des Maladies Genetiques Necker Enfants Malades, Fondation, Paris, France
INSERM (AMU) Institut National de la Santé et de la Recherche Medicale, Marseilles, France
IPCZD (CMHI) Instytut Pomnik Centrum Zdrowia Dziecka, Warsaw Poland
ISCIII Instituto de Salud Carlos III, Madrid, Spain
ISS Istituto Superiore di Sanità, Rome, Italy
LBG (LBI-RUD) Ludwig Boltzmann Gesellschaft GMBH, Vienna, Austria
UMCG Academisch Ziekenhuis Groningen, Netherlands

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LEARNING METHOD
The in-person course is composed of presentations held by the experts and interactive question & answer sessions between speakers and participants. Tool demonstrations and hands-on exercises will be part of the training course as well.

PARTICIPANTS AND REGISTRATION
The training course will take place in-person at Istituto Superiore di Sanità (ISS, viale Regina Elena 299). The training is open to the international research community, to clinicians and to medical specialists who have experience and concrete interest in the diagnosis and research of rare diseases. To ensure active participation and exchange with teaching staff and participants a maximum of 30 participants will be admitted to the training course. A selection process will be applied based on the participants’ background in: genotypic and/or phenotypical identification of rare disorders; deep phenotyping; inferring variants; digital technologies in rare diseases. Priority will be given to participants involved in the European Reference Networks (ERNs) and in national and international Rare Disease Programs and Projects: Undiagnosed Diseases Network International (UDNI), Solving the Unsolved Rare Diseases (Solve-RD).

This course foresees: four fellowships for participants living in an EU-13 Country and Turkey. For more information about eligibility and criteria for selection, contact Claudio Carta at: claudio.carta@iss.it (in Cc laura.cellai@iss.it)

For each fellowship a maximum of 420 euros for round travel and 150 euros/night for hotel accommodation for a maximum of 3 nights are available.

Please note: inside ISS the wearing of FFP2 masks is mandatory. Social distancing will be required during the training course.

Participants are asked to bring their laptop for the hand-on sessions.

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REGISTRATION FORM For important updates, deadlines and for the online registration please visit the website at the following ONLINE REGISTRATION.

Deadline for registration: 15 February 2023.
Registration will remain open for the reserve list only until 2 March 2023.
An e-mail will be sent by 3 March 2023 to the participants selected to attend the course with and without travel and accommodation fellowship.

FEES AND COSTS
The course and registration are free of charge. The course organizers will not cover expenses incurred by the participants in any case.

LEARNING ASSESSMENT
At the end of the course participants will be asked to submit an online multiple-choice learning assessment questionnaire and a satisfaction survey.

ATTENDANCE CERTIFICATES
At the end of the course a certificate of attendance will be forwarded to the participants who attended the entire course programme. No credits for Continuing Education in Medicine will be issued.

OFFICIAL LANGUAGE
English

VENUE
Aula Bovet, Istituto Superiore di Sanità, Viale Regina Elena, 299- Rome, Italy

For important dates, deadlines, registration form, and further information, please visit the website at the following LINK.

CONTACT
If you have questions, please write to the course organizer Claudio Carta: claudio.carta@iss.it (in Cc: laura.cellai@iss.it)

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Programme of the Course

DAY 1 (CEST TIME)  April 3, 2023

09:30 Registration of the Participants
10:00 Welcome address & Faculty & Presentation of the course
   Director National Centre for Rare Diseases
10:15 Overview of the European Joint Programme on Rare Diseases
   Claudio Carta
10:30 Undiagnosed diseases: the family perspective
   Helene Cederroth (online)
11:00 Break
11:20 Overview of the Undiagnosed Diseases Network International
   Domenica Taruscio
11:40 Overview of the Undiagnosed Diseases Network Italy
   Marco Salvatore
12:00 Social and family impact of diagnostic delay of rare diseases: the Spain experience
   Juan Benito Lozano
12:30 Questions & Answers
12:45 Lunch
14:00 Solve-RD, Solving the unsolved Rare Diseases & Use Cases
   Katja Lohmann (online)
14:45 Solving RDs with the RD-Connect Genome-Phenome Analysis Platform
   Leslie Matalonga, Sergi Beltran
15:20 Break
15:30 Hands-on exercise
   Leslie Matalonga, Sergi Beltran
16:30 End of the day 1

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DAY 2 (CEST TIME)  
April 4, 2023

09:45 **Welcome to participants**
10:00 Use Cases from Telethon/TIGEM  
  **Vincenzo Nigro**
10:45 Use Cases from University of Tor Vergata  
  **Giuseppe Novelli**
11:30 Break
11:50 Use Cases from Instytut Pomińk-Centrum Zdrowia Dziecka  
  **Krystyna Chrzanowska**
12:30 Questions & Answers
12:45 Lunch
14:00 Use Cases from Université Aix Marseille  
  **Christophe Béroud**
14:30 Hands-on exercise  
  **Christophe Béroud**
16:00 **End of the day 2**

DAY 3 (CEST TIME)  
April 5, 2023

10:00 Use Cases from Hospital S. Camillo-Forlanini  
  **Francesca Clementina Radio**
10:45 Use Cases from Hôpital Necker Enfants Malades, Institut IMAGINE  
  **Rima Nabbout (online)**
11:30 Break
11:50 Use Cases from University of L’Aquila  
  **Chiara De Luca**
12:30 Questions and Answers
12:45 Lunch
14:00 Closing remarks
   Director of the National Centre for Rare Diseases, Domenica Taruscio, Claudio Carta

14:20 Learning assessment questionnaire & Satisfaction survey

15:00 Free Networking Attendees/Speakers

16:30 End of the Course

SPEAKERS

Sergi Beltran, Centre Nacional d’Anàlisi Genòmica, Barcelona, Spain

Juan Benito Lozano, Instituto de Investigación de Enfermedades Raras, ISCIII - Instituto de Salud Carlos III, Madrid, Spain

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

Claudio Carta, National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

Helene Cederroth, Wilhelm Foundation, Stockholm, Sweden

Krystyna Chrzanowska, Department of Medical Genetics, The Children’s Memorial Health Institute, Warsaw, Poland

Chiara De Luca, University of L’Aquila, L’Aquila, Italy

Katja Lohmann, Institute of Neurogenetics, University of Lübeck, Lübeck, Germany

Leslie Matalonga, Centre Nacional d’Anàlisi Genòmica, Barcelona, Spain

Rima Nabbout, Hôpital Necker Enfants malades, Université Paris Descartes, Institut Imagine, Paris, France

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

Giuseppe Novelli, University of Tor Vergata, Rome, Italy

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Francesca Clementina Radio, Sapienza University, San Camillo-Forlanini Hospital, Rome, Italy

Marco Salvatore, National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

Domenica Taruscio, Istituto Superiore di Sanità, Rome, Italy

COURSE DIRECTOR
Claudio Carta, National Centre for Rare Diseases, ISS, Rome, Italy

SCIENTIFIC SECRETARIAT
Federica Censi, Marco Salvatore, Domenica Taruscio, Fabrizio Tosto
National Centre for Rare Diseases, ISS, Rome, Italy

ORGANIZING SECRETARIAT
Linda Agresta, Laura Lee Cellai, Patrizia Crialese, Stefano Diemoz, Sandro Ghirardi, National Centre for Rare Diseases, ISS, Rome, Italy

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