EUROPEAN JOINT PROGRAMME
RARE DISEASES

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

ICORD
We inspire global collaboration among all rare disease stakeholders

UDNI
Undiagnosed Diseases Network INTERNATIONAL

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.

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 Pomnik-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

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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

DRAFT

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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