EUROPEAN JOINT PROGRAMME
RARE DISEASES

International Course

Training on strategies to foster solutions of undiagnosed rare disease cases

11-13 April 2022
Istituto Superiore di Sanità, Rome, Italy

ONLINE

Endorsed by

ICORD
We inspire global collaboration among all rare disease stakeholders

UDNI
Undiagnosed Diseases Network INTERNATIONAL

Endorsed and taught by selected members of the Undiagnosed Diseases Network International [UDNI] Board of Directors

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", 11-13 April 2022, ISS, Rome will be held ONLINE.

INTRODUCTION AND OBJECTIVES

The International Course "Training on strategies to foster solutions of undiagnosed rare disease cases" is a 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.

This course is part of the 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. Domenica Taruscio, ISS.

The Course is made up of 3 days of training organized by Istituto Superiore di Sanità (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 Conference of Rare Diseases and Orphan Drugs, [ICORD] and is also endorsed and taught by selected members of the Undiagnosed Diseases Network International [UDNI] Board of Directors.

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.

LEARNING METHOD

The 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 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 ONLINE 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).

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

FEES AND COSTS

The course and registration are free of charge.
The course organisers 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 a learning assessment, based on an online multiple-choice 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 of Continuing Education in Medicine will be issued.

OFFICIAL LANGUAGE

English.

VENUE

ONLINE

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 organiser Claudio Carta: claudio.carta@iss.it (in Cc laura.cellai@iss.it)

Programme of the Course

DAY 1 (CEST TIME) **April 11, 2022**

Morning session

09:45 Registration of the Participants

10:00 Welcome address

Domenica Taruscio

10:15 Overview of the European Joint Program on Rare Diseases

Claudio Carta

10:30 "Undiagnosed diseases: the family perspective"

Helene & Mikk Cederroth

10:50 Social and family impact of diagnostic delay of rare diseases: the Spain experience

Juan Benito Lozano

11:20 *Break*

11:40 Overview of Undiagnosed Diseases Network International & Italy

Domenica Taruscio, Marco Salvatore

12:10 NGS Strategies for gene discovery

Silvia Deaglio

12:50 Questions & Answers

13:00 *End of morning session*

Afternoon session

14:30 Undiagnosed /diagnosed: Use Cases part 1

David Adams

15:00 Questions &Answers

15:15 *Break*

15:30 Undiagnosed /diagnosed: Use Cases part 2

David Adams

16:00 Questions &Answers

16:30 *End of the day 1*

DAY 2 (CEST TIME) **April 12, 2022**

Morning session

10:00 Use Cases from IRCCS Santa Lucia

Carlo Caltagirone, Emiliano Giardina

10:45 Questions & Answers

11:00 *Break*

11:15 Solved Use Cases from Instytut "Pomnik-Centrum Zdrowia Dziecka"

Krystyna Chrzanowska

12:00 Questions &Answers

12:15 The IRDiRC Diagnostic Scientific Committee

Birutė Tumienė

12:30 *End of morning session*

Afternoon session

14:00 Solve-RD, Solving the unsolved Rare Diseases

Holm Graessner

14:30 Questions &Answers

14:45 Solving RDs with the RD-Connect Genome-Phenome Analysis Platform

Sergi Beltran, Steve Laurie

15:15 Hands-on exercise

16:00 *End of the day 2*

DAY 3 (CEST TIME) **April 13, 2022**

Morning session

10:00 Use Cases from Aix Marseille Université

Christophe Béroud

10:45 Questions &Answers

11:00 *Break*

11:15 Long read sequencing

Alexander Hoischen

12:00 Questions &Answers

12:30 *End of morning session*

Afternoon session

14:00 "Functional Models"

Kaya Bilguvar

14:45 Questions & Answers

15:00 *Break*

15:15 Genomic Data Analysis and Deep Learning

Julien Gagneur and Holger Prokisch

16:00 Questions &Answers

16:15 Learning assessment and satisfaction survey

16:30 Concluding remarks

Domenica Taruscio, Claudio Carta

16:45 *End of the Course*

SPEAKERS

David Adams, National Institutes of Health, NIH, Bethesda, MD, USA

Sergi Beltran, Centre Nacional d'Anàlisi Genòmica, CNAG-CRG, 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

Kaya Bilguvar, Department of Genetics, School of Medicine, Yale University, New Haven, CT, USA

Carlo Caltagirone, IRCCS Santa Lucia Foundation, Department of Clinical and Behavioral Neurology, Rome, Italy

Claudio Carta, National Centre for Rare Diseases, Rome, Italy

Helene Cederroth, Wilhelm Foundation, Stockholm, Sweden

Mikk Cederroth, Wilhelm Foundation, Stockholm, Sweden

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

Silvia Deaglio, Department of Medical Sciences, University of Turin, Turin, Italy

Julien Gagneur, Department of Informatics, Technical University of Munich, Garching, Germany

Emiliano Giardina, Laboratory of Genomic Medicine, UILDM, Fondazione Santa Lucia, Rome, Italy

Holm Graessner, Institute of Medical Genetics and Applied Genomics, University of Tübingen, Tübingen, Germany

Alexander Hoischen, Department of Human Genetics, Radboud University Medical Center, Nijmegen, the Netherlands

Steven Laurie, Centre Nacional d'Anàlisi Genòmica, CNAG-CRG, Barcelona, Spain

Holger Prokisch, Institute of Human Genetics, School of Medicine, Technische Universität München, Munich, Germany

Marco Salvatore, National Centre for Rare Diseases, Rome, Italy

Domenica Taruscio, National Centre for Rare Diseases, Rome, Italy

Birutė Tumienė, Vilnius University Hospital Santaros Klinikos, Vilnius, Lithuania

COURSE DIRECTOR

Domenica Taruscio, National Centre for Rare Diseases, ISS, Rome, Italy

SCIENTIFIC SECRETARIAT

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ORGANIZING SECRETARIAT

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