EUROPEAN JOINT PROGRAMME
RARE DISEASES

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
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. Domenica Taruscio, 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)] and with the endorsement of ICORD.

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 Conference 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

LEARNING METHOD

The first two days will be dedicated to plenary presentations and Problem-Based Learning (PBL). PBL is a highly interactive and learner-centred approach in which participants, working in small groups assisted by a facilitator, find the solution to a problem that will be discussed at the end of the session with the experts.

On the third day of the course, a hands-on experience with practical demonstrations of useful bio-informatics resources will be shown to the participants.

Participants are asked to bring their laptops for the PBL and the practical demonstration sessions.

PARTICIPANTS

The International 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 attendees will be admitted. 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 national and international Rare Disease Programmes and Projects: Undiagnosed Diseases Network International (UDNI), Solving the Unsolved Rare Diseases (Solve-RD) and the European Reference Networks (ERNs).
This course foresees four fellowships for participants resident, living and working in an EU13 Country (Bulgaria, Croatia, Cyprus, Czech Republic, Estonia, Hungary, Latvia, Lithuania, Malta, Poland, Romania, Slovakia, Slovenia). For more information about eligibility and criteria for selection, contact Claudio Carta at: claudio.carta@iss.it
For each fellowship a maximum of 350 euros for travel (flight and/or train round trip) and 120 euros/night for hotel accommodation for a maximum of 3 nights is available.

REGISTRATION

Online registration form is available at LINK

For the deadlines of the registration and updated info please visit LINK

An e-mail will be sent to the selected participants and to the attendees who have been selected to receive the fellowships within ten days of the closure of the registration.

Respondents who are not selected and contacted by email, will be kept on a waiting list until February 24, 2020.

UDNI Undiagnosed Diseases Network International
Solve-RD "Solving the unsolved rare diseases". Research project funded by the EC, 2018-2022
ERNs European Reference Networks

FEES AND COSTS

The course and the registration are free of charge. Coffee, refreshments and lunches will be offered during the course. Travel, accommodation and other costs incurred to attend the course are at the expense of the participants.
The course organisers will not cover expenses incurred by the participants in any case.

LEARNING ASSESSMENT

Group presentations are foreseen to assess the new learning.
A satisfaction questionnaire will also be submitted to the participants.

ATTENDANCE CERTIFICATES

At the end of the course a certificate of attendance will be handed to the participants who attended 100% of the course. No credits of Continuing Education in Medicine will be issued.

OFFICIAL LANGUAGE

English

VENUE

Aula Rossi, Istituto Superiore di Sanità, Via Giano della Bella, 34 Rome, Italy.

CONTACT

If you have questions please write to the course organiser Claudio Carta, PhD:
claudio.carta@iss.it

Program of the Course

DAY 1 **April 27, 2020**

08:45 Participants registration

09:10 Welcome address & Faculty & Presentation of the course
Domenica Taruscio

09:25 Presentation of the European Joint Programme on Rare Diseases
Domenica Taruscio, Claudio Carta

09:40 Introduction to Problem-Based Learning and small groups
Lorenza Scotti, Claudio Carta

09:50 Coffee-break

10:00 PROBLEM ANALYSIS Working in small groups with facilitators [Reading and analysis of the problem in groups with facilitators]

11:15 Use Case from Telethon
Vincenzo Nigro

12:00 Use Case from Instituto de Salud Carlos III
Estrella Lopez

12:45 Lunch

13:45 PROBLEM SOLUTION Working in small groups with facilitators

15:30 PRESENTATION OF GROUP SOLUTIONS AND FEEDBACK FROM PEERS AND EXPERTS

17:30 End of the day

DAY 2 **April 28, 2020**

08:45 Welcome to participants

09:00 PROBLEM ANALYSIS Working in small groups with facilitators [Reading and analysis of the problem in groups with facilitators]

10:15 Coffee-break

10:30 Use Cases from Undiagnosed Diseases Network International
David Adams, Marco Castori, Roberta Fenoglio

12:00 PROBLEM SOLUTION Working in small groups with facilitators

13:30 Lunch

14:30 PRESENTATION OF GROUP SOLUTIONS AND FEEDBACK FROM PEERS AND EXPERTS

16:30 Undiagnosed Diseases Network International
Domenica Taruscio

16:45 Undiagnosed Diseases Network Italy
Marco Salvatore

17:00 Solve-RD, Solving the unsolved Rare Diseases
Holm Graessner

17:30 End of the day

DAY 3 **April 29, 2020**

08:45 Welcome to participants

09:00 Tool Demo and Hands-on exercise from Institut IMAGINE
Patrick Nitschké

10:15 Coffee-break

10:30 Tool Demo and Hands-on exercise on Phenome Central (or PhenoTips)
Marta Girdea

11:45 Solving Rare Diseases with the RD-Connect Genome-Phenome Analysis Platform
Sergi Beltran, Leslie Matalonga

13:15 Lunch

14:15 Satisfaction Questionnaire

14:30 Closing remarks

15:00 Free Networking Attendees/Speakers/Facilitators

16:30 End of the Course

SPEAKERS/ TRAINERS

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

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

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 , Instituto de Salud Carlos III, 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,

Lorenza Scotti, Research Coordination and Support, ISS, Rome, Italy

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

FACILITATORS

Maria Cristina Barbaro, Scientific Communication Service, ISS, Rome, Italy

Laura Lee Cellai, National Centre for Rare Diseases, ISS, Rome, Italy

Marta De Santis, National Centre for Rare Diseases, ISS Rome, Italy

Margherita Genisio, 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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Lorenza Scotti, Research Coordination and Support, ISS, Rome, Italy

ORGANIZING SECRETARIAT

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