EUROPEAN JOINT PROGRAMME
RARE DISEASES

International Course

Training on strategies to foster solutions of undiagnosed rare disease cases

13-15 March 2024

Istituto Superiore di Sanità, Rome, Italy

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) 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)].

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 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.

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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PARTICIPANTS AND REGISTRATION

The training course will take place in-person at Istituto Superiore di Sanità (ISS, viale Regina Elena 299, Rome, Italy). 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 a 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.

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

REGISTRATION FORM

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

> Online Registration HERE

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.

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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 (CET TIME) **March 13, 2024**

09:30 Registration of the Participants

10:00 Welcome address & Faculty & Presentation of the course

Marco Silano

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

Claudio Carta

10:30 Undiagnosed diseases: the family perspective and Hackathon activity

Helene Cederroth (ONLINE)

11:00 Break

11:20 Undiagnosed Hackathon: procedure

Helene Cederroth (ONLINE)

11:50 Overview of the Undiagnosed Diseases Network National/International

Domenica Taruscio, Marco Salvatore

12:10 UDNI LMICs WG

Domenica Taruscio, Marco Salvatore

12:30 Questions & Answers

12:45 Lunch

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

Holm Graeßner

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

Leslie Matalonga

15:20 *Break*

15:40 Hands-on exercise

Leslie Matalonga

16:40 End of the day 1

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DAY 2 (CET TIME) **March 14, 2024**

09:45 Welcome to participants

10:00 Use Cases from ACURARE_ACIBADEM University: Solved Cases (part 1)

Yasemin Alanay

10:45 Use Cases from ACURARE_ACIBADEM University: Unsolved cases (part 2)

Ozlem Dogan

11:30 Break

12:00 Use Cases from the Memorial Children's Institute in Warsaw

Agnieszka Madej-Pilarczyk

12:45 Questons & answers

13:00 Lunch

14.00 Spain UDP programme

Beatriz Martínez-Delgado

14:15 Novel complex structural variant affecting USP9X gene as a cause of X-linked Intellectual developmental disorder.

Beatriz Martínez-Delgado

15:00 Use cases from Fondazione Policlinico Universitario A. Gemelli IRCCS

Wanda Lattanzi

15:45 Free Networking Attendees/Speakers

16:30 End of the day 2

DAY 3 (CET TIME) **March 15, 2024**

10:00 Use Cases from University Medical Center Groningen

Cleo van Diemen

10:45 Introduction to Variant Interpretation Pipeline, VIP

11:00 Break

11:30 Variant Interpretation Pipeline, VIP: Hands-on exercise

Cleo van Diemen

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12:30 Questions and Answers

12:45 Lunch

14:00 Closing remarks

14:20 Learning assessment questionnaire & Satisfaction survey

15:00 Free Networking Attendees/Speakers

16:00 End of the Course

SPEAKERS

Yasemin Alanay, ACURARE-Rare and Undiagnosed Diseases Center, Acibadem University, Istanbul, Türkiye

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

Helene Cederroth, Wilhelm Foundation, Stockholm, Sweden (ONLINE)

Ozlem Doğan, ACURARE-Rare and Undiagnosed Diseases Center, Acibadem University, Istanbul, Türkiye

Holm Graeßner, University of Tübingen, Tübingen, Germany.

Wanda Lattanzi, Fondazione Policlinico Universitario A. Gemelli IRCCS, Rome, Italy

Beatriz Martínez-Delgado, Molecular Genetics Unit and SpainUDP Program, of Institute for Rare Diseases Research (IIER), Instituto de Salud Carlos III, Madrid, Spain

Leslie Matalonga, CNAG-CRG, Centre for Genomic Regulation (CRG), The Barcelona Institute of Science and Technology, Barcelona, Spain

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Agnieszka Madej-Pilarczyk, Department of Medical Genetics, The Children's Memorial Health Institute, Warsaw, Poland

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

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

Domenica Taruscio, Former director National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

Cleo van Diemen, Department of Genetics, University of Groningen, Groningen, The Netherlands

COURSE DIRECTOR

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

SCIENTIFIC SECRETARIAT

Marco Salvatore, Domenica Taruscio

National Centre for Rare Diseases, ISS, Rome, Italy

ORGANIZING SECRETARIAT

Linda Agresta, Laura Lee Cellai, Patrizia Crialese, Stefano Diemoz, Donata Girolamo
National Centre for Rare Diseases, ISS, Rome, Italy

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