



EUROPEAN JOINT PROGRAMME  
RARE DISEASES

**3<sup>rd</sup> Training Course**

***“Quality assurance, variant interpretation and data management in the NGS diagnostics era”***

**October 27 –29, 2021**

**Istituto Superiore di Sanità, Rome, Italy**

**Endorsed by**



## GENERAL INFORMATION

Due to the force majeure situation and in order to allow the correct progress of the planned tasks of EJP RD, ISS ensures that the international course “Quality assurance, variant interpretation and data management in the NGS diagnostics era”, October 27-29, 2021, ISS, Rome **will be held ONLINE**.

### RELEVANCE, INTRODUCTION AND OBJECTIVES

The Training Course “**Quality assurance, variant interpretation and data management in the NGS diagnostics era**” 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”. For more information about the EJP-RD, see <https://www.ejprarediseases.org/>

In particular, this Course is a part of the WP14 on “Data Management & Quality Training”, which aims to organize residential training courses in different Countries. WP Leader: Dr. Claudio Carta, ISS, Task Leader Gert Matthijs, KU Leuven

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 Task Leader KU Leuven and, mainly, EJP RD Task Partners [EKUT Tübingen, ACU/ACURARE Istanbul, IPCZD(CHMI) Warsaw, CNAG-CRG Barcelona, INSERM (AMU) Marseille, UMC Groeningen]

Next Generation Sequencing (NGS) generates overwhelming amounts of data. Clinical and basic researchers are increasingly confronted with the complexity of genomic data. It is therefore of fundamental importance to provide researchers and clinicians with specific trainings on the interpretation of genetic variants and quality standards.

The course is endorsed by the International Conference for Rare Diseases and Orphan Drugs, European Society of Human Genetics, EuroGentest, Società Italiana di Genetica Umana and is also endorsed and taught by selected members of the Undiagnosed Diseases Network International Board of Directors.

### SPECIFIC LEARNING OBJECTIVES

The course will build on expertise gained by EuroGentest and help in the translation of research tools to diagnostic applications (in line with the IRDiRC objectives). The impact is on the quality and reliability of NGS results, obtained through rare disease research.

At the end of the training course participants will be able to:

- Perform a validation of a pipeline for NGS variants
- Achieve the management of a quality diagnostic laboratory
- Use international databases for rare diseases

## **LEARNING METHOD**

The training course will consist of plenary presentations, interactive question & answer sessions between speakers and participants, and hands-on trainings.

## **PARTICIPANTS AND REGISTRATION**

The training course is open to the international research community, clinicians, medical specialist, laboratory scientists (EBMG registered), junior laboratory scientists, clinical geneticists, policy makers and assessors for laboratory accreditation, and patient representatives, with a basic knowledge in biology or medicine.

To ensure active participation and exchange with teaching staff and participants a maximum of 30 participants will be admitted to the ONLINE training course.

**REGISTRATION FORM Important dates, deadlines, please visit the website at the following [LINK](#)**

## **SELECTION COMMITTEE**

A Scientific Selection Committee, including the 5 core partners for these series of training courses (ISS Rome, KU Leuven, EKUT Tübingen, ACU/ACURARE Istanbul, IPCZD-CHMI Warsaw,) will select the final list of participants.

## **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 a learning assessment, based on an online multiple-choice questionnaire will take place. A satisfaction survey will be also submitted.

## **ATTENDANCE CERTIFICATES**

At the end of the course a certificate of attendance will be forwarded to the participants who attended 100% of the training course. 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.cart@iss.it](mailto:claudio.cart@iss.it) (in Cc [laura.cellai@iss.it](mailto:laura.cellai@iss.it))



## Programme of the Course

### DAY 1 (CEST Time)

October 27, 2021

10:00 Welcome address

**Domenica Taruscio**

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

**Claudio Carta**

10:30 Clinical applications of NGS

**Silvia Deaglio**

11:00 Questions & Answers

11:15 Break

11:30 NGS techniques for gene discovery: detection and variant interpretation

**Achille Iolascon, Roberta Russo**

12:00 Hands-on

12:30 Break

14:00 Validation of pipelines and procedures

**Daniele Calistri**

14:30 Questions & Answers

14:45 Detection of non-coding and epigenetic variations in the genome

**Bernard Thienpont**

15:15 Questions & Answers

15:30 HTA of NGS

**Katherine Payne**

16:15 Questions & Answers

16:30 End of Day 1

### DAY 2 (CEST Time)

October 28, 2021

10:00 Diagnostic Laboratory quality assurance and management

**Gert Matthijs**

10:30 Questions & Answers

10:45 The Italian Experience on the external quality assurance of genetic testing

**Federica Censi, Fabrizio Tosto, Marco Salvatore and Domenica Taruscio**

11:15 Questions & Answers

11:30 Break

11:45 The role of bed-bench interactions for reporting NGS data in the clinical context and genetic counseling

**Marco Castori**

12:15 Questions & Answers

12:30 Break

14:30 Hands-on: Variant prioritization and HPO terminology

**Leslie Matalonga, Steven Laurie**

16:30 End of Day 2

### **DAY 3 (CEST Time)**

**October 29, 2021**

10:00 In-vitro Diagnostics

**Els Dequeker**

10:30 Questions & Answers

10:45 Beyond the exome

**Olaf Riess**

11:15 Questions & Answers

11:30 Break

11:45 Variant sharing and databases, and technological innovations

**Paolo Radice**

12:15 Questions & Answers

12:30 Break

14:30 The 1+ Million Genomes initiative

**Ivo Gut**

15:15 Question & Answers

15:30 Course Evaluation and Satisfaction Questionnaire

16:00 Closing Remarks

**Domenica Taruscio Gert Matthijs, Claudio Carta**

16:30 End of the Course

## **SPEAKERS**

**Daniele Calistri**, Istituto Scientifico Romagnolo per lo Studio e la Cura dei Tumori (IRST) IRCCS, Meldola, Italy

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

**Marco Castori**, Casa Sollievo della Sofferenza, San Giovanni Rotondo, Italy

**Federica Censi**, National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

**Silvia Deaglio**, University of Turin, Turin, Italy

**Els Dequeker**, Katholieke Universiteit Leuven, Leuven, Belgium

**Ivo Gut**, Centre Nacional d'Anàlisi Genòmica, Barcelona, Spain

**Achille Iolascon**, Department of Molecular Medicine and Medical Biotechnology, University of Naples Federico II, Naples, Italy

**Steven Laurie**, Centre Nacional d'Anàlisi Genòmica, Barcelona, Spain

**Leslie Matalonga**, Centre Nacional d'Anàlisi Genòmica, Barcelona, Spain

**Gert Matthijs**, Katholieke Universiteit Leuven, Leuven, Belgium

**Katherine Payne**, The University of Manchester, Manchester, UK

**Paolo Radice**, Fondazione IRCCS (Istituto Di Ricovero e Cura a Carattere Scientifico) Istituto Nazionale dei Tumori (INT), Milan, Italy

**Olaf Riess**, University of Tuebingen, Tuebingen, Germany

**Roberta Russo**, Department of Molecular Medicine and Medical Biotechnology, University of Naples Federico II, Naples, Italy

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

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

**Bernard Thienpont**, Katholieke Universiteit Leuven, Leuven, Belgium

**Fabrizio Tosto**, National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy

## **COURSE DIRECTOR**

Dr. Domenica Taruscio, ISS, Italy

### SCIENTIFIC SECRETARIAT

**Gert Matthijs**, KU Leuven, Belgium

**Claudio Carta** (Coordinator), **Federica Censi**, **Marco Salvatore**, **Fabrizio Tosto**

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### SCIENTIFIC SELECTION COMMITTEE

**Claudio Carta**, ISS, Rome, Italy

**Krystyna Chrzanowska**, IPCZD, Warsaw, Poland

**Holm Graessner**, University of Tübingen, Germany

**Gert Matthijs**, KU Leuven, Belgium

**Ugur Ozbek**, ACU/ACURARE, Istanbul, Turkey

**Domenica Taruscio**, ISS, Rome, Italy

### ORGANIZING SECRETARIAT

**Liliane Geyskens** KU Leuven, Belgium

**Linda Agresta**, **Daniela Bernardo**, **Laura Lee Cellai**, **Patrizia Crialesi**, **Stefano Diemoz**,  
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