QUALITY ASSURANCE, VARIANT INTERPRETATION AND DATA MANAGEMENT IN THE NEXT GENERATION SEQUENCING DIAGNOSTICS ERA

27 -29 October 2021

organised by

ISTITUTO SUPERIORE DI SANITÀ

National Centre for Rare Diseases

In collaboration with the Partners of the European Joint Programme on Rare Diseases (EJP RD), Grant Agreement No 825575

with the endorsement of: International Conference for Rare Diseases and Orphan Drugs,

Società Italiana di Genetica Umana

and also endorsed and taught by selected members of the Undiagnosed Diseases Network International Board of Directors

The endorsement of the European Society of Human Genetics, and of EuroGentest,

has been requested

Relevance

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

The training course is a part of a series of training activities proposed by the European Joint Programme on Rare Diseases (EJP RD), 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/.

Learning objectives

This training course is a part of Work Package 14 (WP Leader Claudio Carta, ISS) of the EJP RD, aiming at organizing residential training courses in different Countries on "Data Management and Quality Training".

Through the training course participants will learn to evaluate the pathogenetic nature and clinical significance of genetic variants, the criteria necessary for NGS analysis pipelines and the use of international databases. This is expected to impact on the quality and reliability of NGS results, obtained through rare disease research.

Specific aims

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

Training method
The course is composed of plenary presentations held by expert speakers, interactive question & answer sessions between speakers and participants and hands-on trainings.

PROGRAMME

Day 1

27 October

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 technique 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

28 October

10.00 Diagnostics 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, 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 Day2

Day 3

29 October

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 Questions & 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/TRAINERS

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

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

Marco Castori, Casa Sollievo della Sofferenza, Italy

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

Silvia Deaglio, University of Turin, Italy

Els Dequeker, Katholieke Universiteit Leuven, Belgium

Ivo Gut, Centre Nacional d'Anàlisi Genòmica, Spain

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

Steven Laurie, Centre Nacional d'Anàlisi Genòmica, Spain

Leslie Matalonga, Centre Nacional d'Anàlisi Genòmica, Spain

Gert Matthijs, Katholieke Universiteit Leuven, Belgium,

Katherine Payne, The University of Manchester, UK

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

Olaf Riess, University of Tuebingen, Germany

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

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

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

Bernard Thienpont, Katholieke Universiteit Leuven, Belgium

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

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General Information

Venue: Online, through Microsoft Teams Platform. The connection details will be sent by email to the selected participants.

Participants

The training course is open to the international research community, clinicians, medical specialists, laboratory scientists (EMBG registered), junior laboratory scientists, clinical geneticists, policy makers, assessors for laboratory accreditation and patient representatives with a basic knowledge in biology or medicine.
A maximum of 30 attendees will be admitted to the online training course

Registration

The online registration form is available at the following link: ONLINE REGISTRATION until 11 July 2021.

An email will be sent by 30 July 2021 to the selected participants.

Respondents who are not selected and contacted by email will be kept on a waiting list until 11 October 2021.

Fees and costs

The course and registration are free of charge.

The organisers will not cover expenses incurred by the participants in any case.

Selection of participants

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. The selection process will be carried out in agreement with Task Partners.

Learning assessment

A multiple-choice learning assessment questionnaire and a satisfaction survey will be submitted to the participants at the end of the training course.

Certificate of attendance

At the end of the course a certificate of attendance will be forwarded to the participants who attended 100% of the course. No CME credits will be issued.

For any other information, please write to the course Coordinator Dr. Claudio Carta (claudio.carta@iss.it, with laura.cellai@iss.it in Cc**).**