2nd TRAINING COURSE

"Quality assurance, variant interpretation and data management in the NGS diagnostics era"

12-14 October 2020

Due to COVID-19
the training course will be ON-LINE

GENERAL INFORMATION

INTRODUCTION AND OBJECTIVES

The Training Courses on Standards and quality of genetics/genomics data in laboratory and clinical research practice are a part of a series of programs put forth by the European Joint Programme for Rare Diseases (EJP RD). EJP RD is a European Commission co-funded project (GA 825575, 2019-2023).

The main objectives of the program WP14 are to decrease RD data fragmentation and increase data quality through training activities on data management & quality which will raise the level of capacities and help data sharing and networking within the RD community.

METHODOLOGY

The WP14 program will achieve the objectives through (i) the integration and implementation of existing and successful training activities and (ii) development of new specific trainings to address all objectives. This will ensure appropriate coverage of relevant aspects on data management & quality under a joint programme for meaningful interaction and comprehensive training across RD community in Europe.

THE TRAINING COURSES on Standards and quality of genetics/genomics data in laboratory and clinical research practice

The organisation of the specific training courses in program WP14.2 is coordinated by KU Leuven. Project partners are EKU Tübingen, ACU/ACURARE Istanbul, ISS Rome, IPCZD(CHMI) Warsaw, CNAG-CRG Barcelona, INSERM (AMU) Marseille, UMC Groningen.

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 importance to provide training on the interpretation of genetic variants and quality standards.

This is an annual course for scientists, bio-informaticians and clinicians with an interest in genomic technologies. The aim is to teach participants about the evaluation of the pathogenic nature and clinical significance of genetic variants, on the criteria that have to be set to the NGS analysis pipelines and on the use of international databases.

2020 – 2nd TRAINING COURSE

Due to COVID-19 this will be a 3-day ON-LINE training course on "Quality assurance, variant interpretation and data management in the NGS diagnostic era". Up to 40 participants can be accepted.

Over the years, the course content will shift to total genome interpretation and other "-omics" applications which will entail novel disease molecular mechanisms.

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. The next meetings will be hosted in Rome, Tubingen and Warsaw.

SELECTION COMMITTEE

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

ATTENDANCE CERTIFICATES

At the end of the training course a Certificate Of Attendance will be sent to the participants who attended the entire workshop.

No credits of Continuing Education in Medicine will be issued.

LANGUAGE

The training courses will be in English.

CONTACT

If you have questions, please write to the
organiser of this 2020 ON-LINE edition in Istanbul: Prof. Uğur ÖZBEK
or the coordination team in Leuven : Prof. Gert Matthijs

PROGRAM

See next pages.

Monday, 12th October 2020

(CET (Brussels); UCT+2)

09:00-09:30	Clinical applications of NGS	***Yasemin Alanay, Istanbul (Tr)***
09:30-10:00	Whole Genome Sequencing of rare diseases for diagnostics and gene discovery	***Lucy Raymond, Cambridge (UK)***
10:00-10:15	Group discussion	
10:15-10:30	**Break**	
10:30-11:00	Diagnostic Laboratory quality assurance and management	***Gert Matthijs, Leuven (B)***
11:00-11:30	Validation of an NGS variant pipeline/WGS	***Erika Souche, Leuven (B)***
11:30-12:00	Group Discussion	
12:00-14:00	**Lunch break**	

14:00-17:30 Hands on practice: Genome analysis and variant discovery

Hacettepe University Bioinformatics Team (Tr)
Can Koşukçu , Ceren Sucularlı, İdil Erte Yet , Gülşah Merve Kılınç

"In this hands-on practice we will introduce the Linux/Bash environment and basic commands including listing, copying and moving the files. Next, we will present the file formats that are used to store the raw and intermediate Next Generation Sequencing files as well as genomic variant data. We will demonstrate how to analyze the raw genome sequence data including the quality control, mapping to the reference genome and genomic variant discovery steps."

14:00-14:45	BLAST
14:45-15:45	Linux/Bash
15:45-16:00	Break
16:00-16:30	Introduction to NGS & Data types
16:30-17:30	Read mapping and variant (SNP/Indel) discovery in whole genome sequence data

Tuesday, 13th October 2020

(CET (Brussels); UCT+2)

09:00-09:30	Indels and CNV's by NGS, Large Sequence Variations	***Can Alkan, Ankara (Tr)***
09:30-10:00	How to make your data FAIR?	***Uğur Sezerman,Istanbul (Tr)***
10:00-10:15	Group discussion	
10:15-10:30	**Break**	
10:30-11:00	HTA of WGS in genome diagnostics prior to genetic counselling	***Uğur Özbek, Istanbul (Tr)***
11:00-11:30	Data formatting and sharing solutions in WGS	**Özkan Özdemir, Istanbul(Tr)**
11:30-12:00	Group Discussion	
12:00-14:00	**Lunch Break**	
14:00-14:30	International guidelines for variant annotation – Using ClinVar as a resource to support variant interpretation	***Steven Harrison, Cambridge (USA)***
14:30-16:30	Hands-on: Variant prioritization and HPO terminology	***Leslie Matalonga/Steven Laurie, Barcelona (E)***

Wednesday, 14th October 2020

(CET (Brussels); UCT+2)

09:00-09:30	Validation of non-invasive prenatal testing and low pass sequencing from chromosome analysis	***Murat Çetinkaya (Tr)***
09:30-10:00	Novel genomic applications in preimplantation genetic testing and prenatal diagnosis	***Eftychia Dimitriadou, Leuven (B)***
10:00-10:15	Group Discussion	
10:15-10:30	**Break**	
10:30-11:00	Single molecule sequencing – PacBio	***Kornelia Neveling, Nijmegen (NL)***
11:00-11:30	Group Discussion & Closing of training course	

Scientific Selection Committee
Prof. Gert Matthijs, KU Leuven, Belgium
Prof. Ugur Ozbek, ACU/ACURARE, Istanbul, Turkey
Prof. Claudio Carta, ISS, Rome, Italy
Prof. Holm Graessner, University of Tübingen, Germany
Prof. Krystyna Chrzanowska, IPCZD, Warsaw, Poland
Co-Partners
Prof. Christophe Béroud, AMU, Marseille, France
Prof. Sergi Beltran, CNAG, Barcelona, Spain
Prof. Morris Swertz, UGroningen, Netherlands