2nd TRAININGCOURSE

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

12-14 October 2020, Istanbul

Deadline Registration: 20 July 2020

Please note that a mitigation plan is in place due to COVID-19.

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

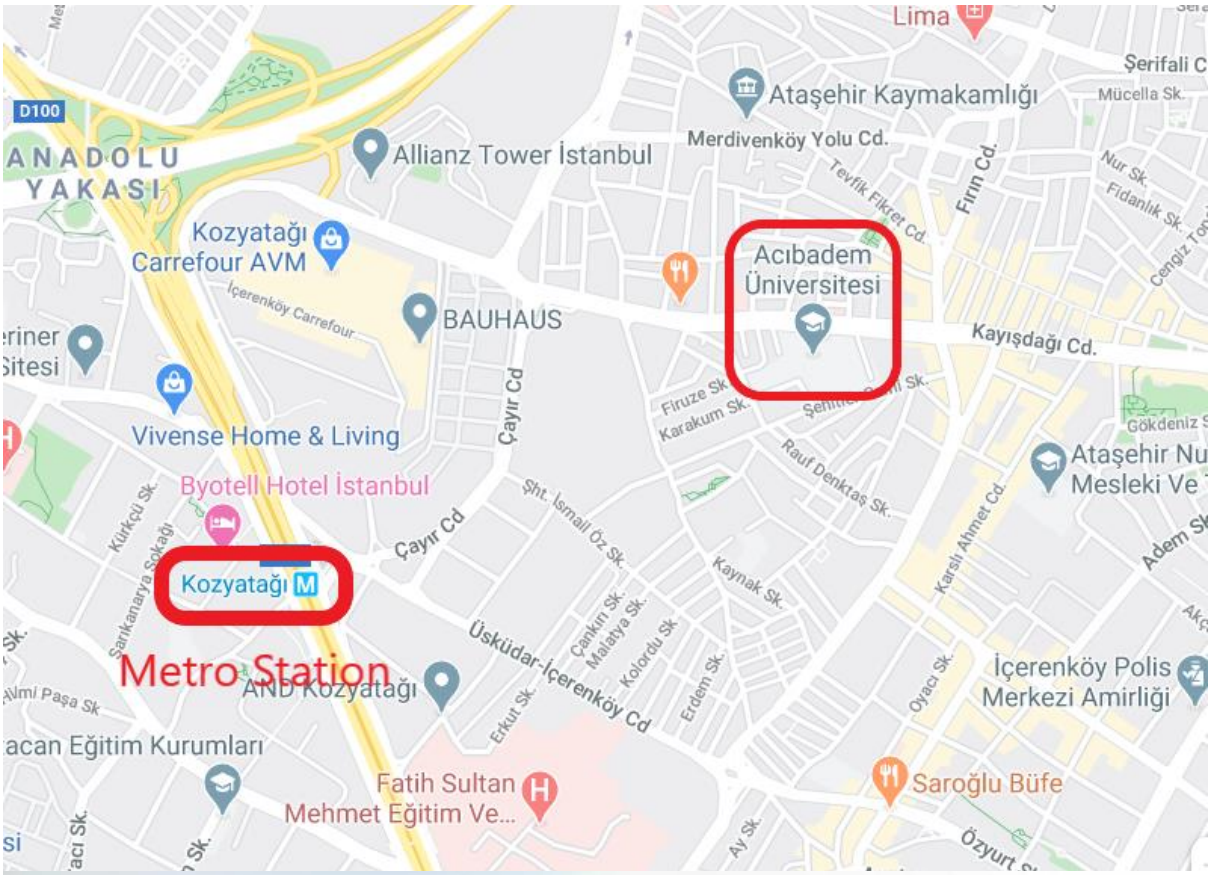
Theme of this 3-day residential training course is **"Quality assurance, variant interpretation and data management in the NGS diagnostic era"** and can host 35 participants.
The course will build on expertise gained by EuroGentest and help in the translation of research tools to diagnostic applications (in line with the IRDiRC objectives). The impact is on the quality and reliability of NGS results, obtained through rare disease research.

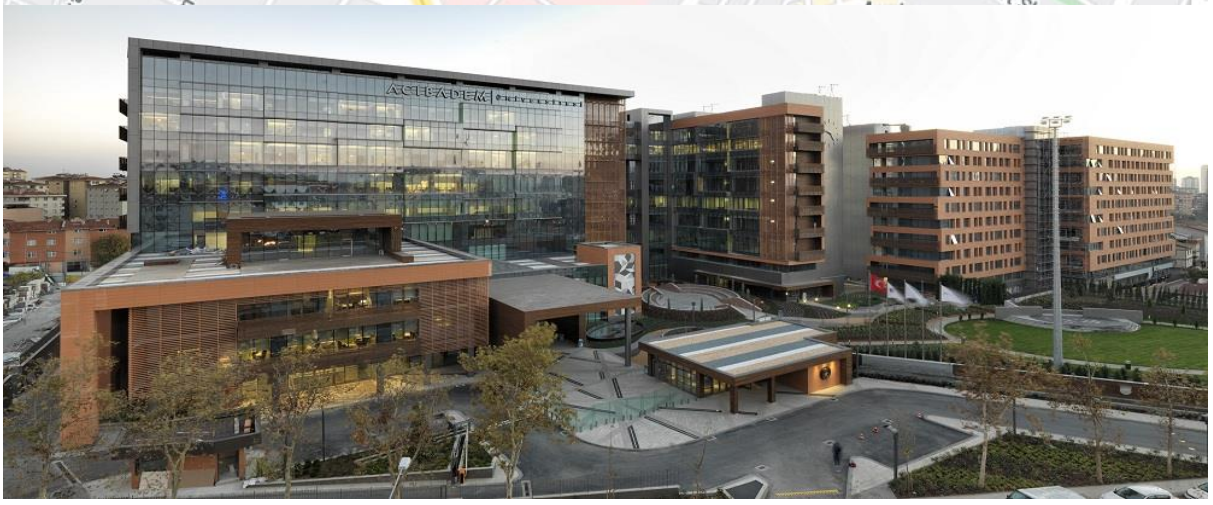
The next meetings will be hosted in Rome, Tubingen and Warsaw.

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

VENUE

ACIBADEM Üniversitesi, Kerem Aydınlar Kampüsü,
Kayışdağı Cad. No:32, **Ataşehir/İstanbul, Türkiye**





PARTICIPANTS

The training courses are accessible to laboratory scientists (EBMG registered), junior laboratory scientists, clinical geneticists, other medical specialists in training, policy makers and assessors for laboratory accreditation, and patient representatives, with a basic knowledge in biology or medicine. 60% of participants should belong to the first two categories.

The training course will consist of **lectures and hands-on trainings**, with an interactive and learner-centred approach.

Participants are asked to **bring their laptops,** if possible.
For the hands-on sessions, a class room with PC's will be available.

To ensure active participation and exchange with teaching staff and fellow participants, a **maximum of 35 participants** will be admitted to the training course.

REGISTRATION – Application deadline: 20 July 2020

If you wish to attend this training course, please complete the online registration and submit your application before 20 July 2020.
Please note that a mitigation plan is in place due to COVID-19

FEES AND COSTS

The training course and registration is **free of charge.**
Coffee, refreshments and lunches will be offered during the course.

Participants are expected **to arrange their own travel, accommodation** and other expenses to attend the training course.

The course organisers will not cover expenses incurred by the participants.

TRAVEL FELLOWSHIPS

Two travel fellowships are available covering one round trip flights (for a max. of €350) and up to three nights of hotel accommodation (for a max. of €120/night).
Especially participants from EU-13 countries *(Estonia, Latvia, Lithuania, Poland, Czech Republic, Slovakia, Hungary, Slovenia, Bulgaria, Romania, Malta, Cyprus, Croatia)* are encouraged to subscribe.

If you wish to **apply for a travel fellowship** please mark this in the online registration tool.

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 and the 2 travel fellowships.

Before the end of August, all registered participants will be informed about the selections.

HOTEL ACCOMMODATION

Participants are kindly asked to arrange their own hotel accommodations.

PUBLIC TRANSPORT

The Kerem Aydınlar Campus is located 1 km from Kozyatağı metro station.
Several buses leave from Kadıköy and Üsküdar bus stations to the Campus.

Buses from Kadıköy – 14A, 19T, 19S
Buses from Üsküdar – 11T, 320A

You can get off the bus at "Acıbadem Üniversitesi" stop.

You can download the **app called "MOBIETT"** for bus times and routes.

ATTENDANCE CERTIFICATES

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

No credits of Continuing Education in Medicine will be issued.

LANGUAGE

The training courses will be in English.

CONTACT

If you have questions, please contact the organiser of this training course, Prof. Uğur ÖZBEK at ACURARE, or the coordinator Prof. Gert Matthijs at KU Leuven.

PROGRAM

Monday, 12th October 2020

09:00-09:45	Clinical applications of NGS	**Yasemin Alanay, Istanbul (TR)-TBC**
09:45-10:30	Whole Genome Sequencing of rare diseases for diagnostics and gene discovery	**Lucy Raymond, Cambridge (UK)-TBC**
10:30-11:00	**Coffee break**	
11:00-11:45	Diagnostic Laboratory quality assurance and management	**Gert Matthijs, Leuven(B)**
11:45-12:30	How to make your data FAIR?	**Uğur Sezerman,Istanbul (TR)-TBC**
12:30-13:15	**Lunch**	
13:15-17:00	Hands on practice: Genome analysis and variant discovery "In this hands on practice we will introduce the Unix/Bash environment and basic linux commands including listing, copying and moving the files. Next, we will present the file formats that are used to store the sequencing and 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."	**Hacettepe Üniversity Bioinformatics Team (TR)** **Can Koşukçu** **Ceren Sucularlı** **İdil Erte Yet** **Gülşah Merve Kılınç**
13:15-13:45	Unix/Bsh	
13:45-14:15	Data types	
14:15-14:30	**Coffee Break**	
14:30-17:00	Read mapping and variant (SNP/Indel) discovery in whole genome sequence data	

A group dinner will be organized

Tuesday, 13th October 2020

09:00-09:45	International guidelines for variant annotation – Using ClinVar as a resource to support variant interpretation	**Steven Harrison, Cambridge (USA)-TBC**
09:45-10:30	Indels and CNV's by NGS, Large Sequence Variations	**Can Alkan, Ankara (TR)-TBC**
10:30-11:00	**Coffee break**	
11:00-12:00	Panel: Genome diagnostics and importance of variant sharing *Moderator; Nurten Akarsu*	
11:00-11:30	HTA of WGS in genome diagnostics prior to genetic counseling	**Uğur Özbek, Istanbul (TR)**
11:30-12:00	Data formatting and sharing solutions in WGS	**Uğur Sezerman, Istanbul (TR)-TBC**
12:00-13:30	**Lunch**	
13:30-16:00	Hands-on: Variant prioritization and HPO terminology	**Leslie Matalonga/Steven Laurie, Barcelona (E)-TBC**
16:00-17:00	Panel: Would you sequence your genome and publish the data	**Gert Matthijs, Leuven(B)**
20:00	**All speakers are invited for dinner**	

Wednesday, 14th October 2020

09:00-09:45	Validation of non-invasive prenatal testing and low pass sequencing from chromosome analysis	**Murat Çetinkaya (TR)-TBC**
09:45-10:30	Novel NGS application in PGD/PND (genomic applications in prenatal diagnosis)	**TBA (TR)**
10:30-11:00	**Coffee break**	
		Kornelia Neveling, Nijmegen (NL)-TBC
11:00-11:45	Single molecule sequencing – PacBio	
11:45-12:30	TBA	
12:30-13:30	**Lunch**	
13:30	Old City Guided tour	

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

Local Organising Committee
Prof. Ugur Ozbek, ACU/ACURARE, Istanbul, Turkey
Özden Hatırnaz NG
Gizem Şentürk
Sezer Akyoney