1st TRAINING COURSE

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

23-25 October 2019, Leuven

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.

2019 – 1st TRAINING COURSE

This first 3-day residential training course on ***"Quality assurance, variant interpretation and data management in the NGS diagnostic era"*** will host 20 participants.

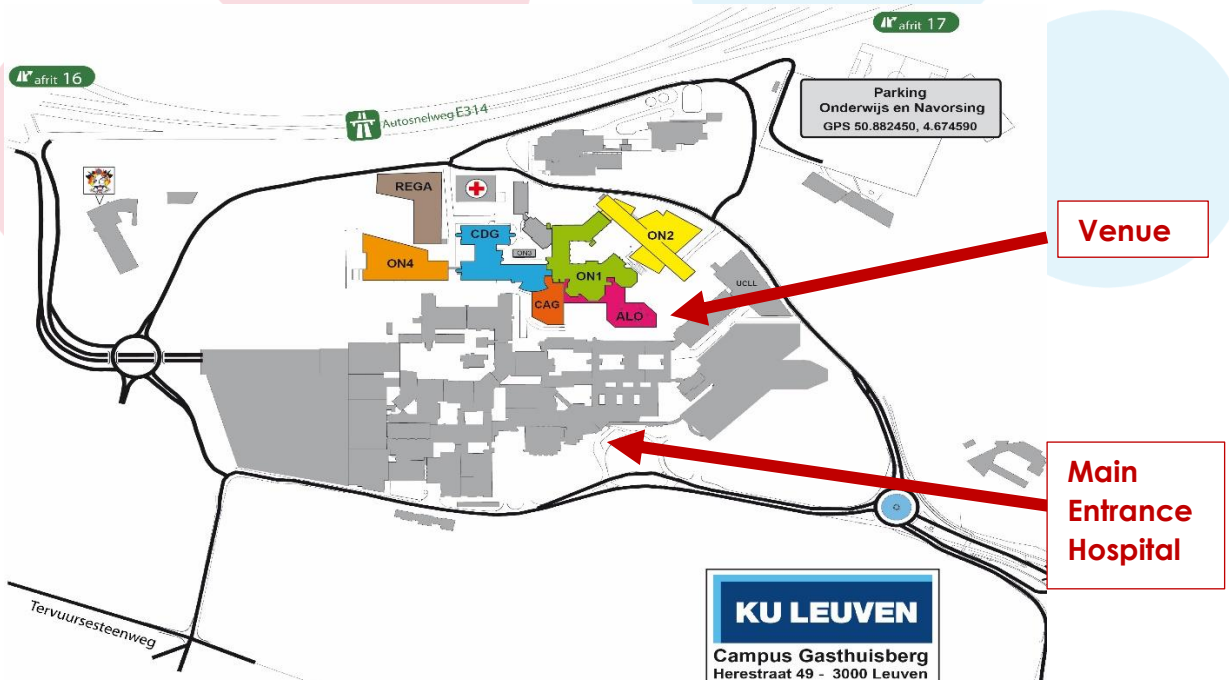
The course will build on expertise gained by EuroGentest and help in the translation of research tools to diagnostic applications (in line with the IRDiRC objectives). The impact is on the quality and reliability of NGS results, obtained through rare disease research.

The next meetings will be hosted in Istanbul, 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

KU Leuven, Campus Gasthuisberg,
Herestraat 49, **Onderwijs & Navorsing**
Building: **ALO**
3000 Leuven, Belgium



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 1st training course will consist of **lectures and hands-on trainings**, with an interactive and learner-centred approach.

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

To ensure active participation and exchange with teaching staff and fellow participants, a **maximum of 20 participants** will be admitted to the training course.

REGISTRATION - Application deadline 31 July 2019

If you wish to attend to this training course, please complete the online registration fields and submit your application before 31 July 2019.

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 to cover 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.

After July 31, all registered participants **will be informed** if they are selected to attend this training course.

HOTEL ACCOMMODATION

Participants are kindly asked to arrange their own hotel accommodations.
For hotels in Leuven, see: https://www.visitleuven.be/en/lodging

PUBLIC TRANSPORT

The Campus Gasthuisberg is located 4 km from Leuven train station.
Several buses leave from the central bus station (in front of train station) to Gasthuisberg about every ten minutes.

Bus 3-CAMPUS stops right next to the building 'Onderwijs en Navorsing & ALO', make sure to remain seated until you reach the very last stop called 'Leuven Gasthuisberg Campus'.

Bus 600-BUITENRINGBUS (bus gate **12**) leaves the station every 10 minutes.
First stop is Gasthuisberg, in front of the main hospital entrance.

You can take any bus that stops at the main entrance of the University Hospital Gasthuisberg and ask at the reception for directions to the ALO building.

Information on public transportation by bus is available at De Lijn and for trains at Belgian Rail. See Google maps : https://goo.gl/maps/rwpvNXBDg3tKExPL9

ATTENDANCE CERTIFICATES

At the end of the training course a Certificate Of Attendance will be handed out 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 first training course, prof. Gert Matthijs

PROGRAMME

See next pages.

EUROPEAN JOINT PROGRAMME RARE DISEASES

ESHG EUROPEAN SOCIETY OF HUMAN GENETICS

EuroGentest

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

Leuven, 23-25 October 2019

Wednesday 23th October 2019

12:00-13:00	**Registration & Coffee**	
13:00-14:00	Clinical applications of NGS	***Jeroen Breckpot, Leuven (B)***
14:00-15:00	Whole Genome Sequencing of rare diseases for diagnostics and gene discovery: coding and non-coding	***Kathleen Freson, Leuven (B)***
15:00-15:30	**Coffee break**	
15:30-16:30	Indels and CNV's	***Alejandro Sifrim, Leuven (B)***
16:30-17:30	Validation of an NGS variant pipeline / WGS	***Erika Souche, Leuven (B)***
18:30-19:30	**City walk Leuven**	**Guided tour**

Thursday 24th October 2019

09:00-09:45	Validation of low pass sequencing from chromosome analysis	***Björn Menten, Ghent (B)***
09:45-10:30	International guidelines for variant annotation – Using ClinVar as a resource to support variant interpretation	***Steven Harrison, Cambridge (USA)***
10:30-11:00	**Coffee break**	
11:00-13:00	Hands-on: Variant prioritization and HPO terminology	***Leslie Matalonga/Steven Laurie, Barcelona (E)***
13:00-14:30	**Lunch**	
14:30-15:30	HTA of WGS in genome diagnostics prior to genetic counseling	***Katherine Payne, Manchester (UK)***
15:30-16:00	**Coffee break**	
16:00-16:45	Variant data sharing by clinical laboratories through public databases: consent, privacy and further contact for research policies	***Mahsa Shabani, Leuven (B)***
16:45-17:30	Panel Debate: "Would you sequence your genome and publish the data"	***Gert Matthijs, Leuven (B)***

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Friday 25th October 2019

09:00-09:45	Validation of non-invasive prenatal testing	***Nathalie Brison/Kris Van Den Bogaert, Leuven (B)***
09:45-10:30	Data sharing solutions in WGS	***Christophe Béroud, Marseille (F)***
10:30-11:00	**Coffee break**	
11:00-11:45	Novel NGS application in PGD/PND (genomic applications in prenatal diagnosis)	***Joris Vermeesch, Leuven (B)***
11:45-12:30	Single molecule sequencing - PacBio	***Kornelia Neveling, Nijmegen (NL)***
12:30-14:00	**Lunch**	