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First Report on Course on interpretation of genetic variants and quality standards

Organisation name of lead beneficiary for this deliverable:

7 – KU Leuven

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Dissemination level:

PU

Introduction and Objective

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), a European Commission (EC) co-funded project (Grant Agreement 825575, 2019-2023).

The training course that is the object of the present deliverable falls under the action of WP 14, "Training on data management and quality", Task 14.2 "Standards and quality of genetics/genomics data in laboratory and clinical practice". Task leader is KU (Katholieke Universiteit) Leuven.

The main objectives of the program of 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.

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

Methodology

Definition of the training programme

The programme of the training was defined as first step. The training course, "Quality assurance, variant interpretation and data management in the NGS diagnostics era", in task 14.2 was coordinated by KU Leuven, together with task partners EKU Tübingen, ACU/ACURARE Istanbul, ISS Rome, IPCZD (CHMI) Warsaw, CNAG-CRG Barcelona, INSERM (AMU) Marseille, UMCG Groningen.

The course is an annual training for scientists, bio-informaticians and clinicians with an interest in genomic technologies.

The aim of the course is to teach participants about the evaluation of the pathogenetic 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.

The edition of the course that was held on 23-25 October 2019 in Leuven, represented the first edition of the training courses of task 14.2 and was the first 3-day residential training course on "Quality assurance, variant interpretation and data management in the NGS diagnostics era".

The course had the aim of building on expertise gained by EuroGentest and helping in the translation of research tools to diagnostic applications (in line with the IRDiRC objectives).

The course was expected to have an impact on the quality and reliability of NGS results obtained through RDs research.

Selection of the Speakers

Speakers were selected on the base of their expertise on the aims of the course. Most of the speakers were from KU/UZ (Universitair Ziekenhuis) Leuven, as these institutions have gained great expertise on the subject. The following speakers, selected from among EJP RD Partners, were invited: Leslie Matalonga and Steven Laurie from CNAG-Barcelona; Christophe Béroud from AMU-Marseille.

Selection of the training method

A lecture within the scope of the course and hands-on practice in variant interpretation and genome analysis was selected as training method,

Moreover 3 case studies developed by RD Connect were produced and handed to the selected participants as preparatory materials.

Dissemination of the course

The course was disseminated online through different websites, event calendars, newsletter such as EJP RD, the European Society of Human Genetics, Inserm, KU Leuven, the American Society of Human Genetics

The internal mailing within KU/UZ Leuven was also used for the dissemination of the training course.

Selection of participants

The participants that were selected to attend the training course had, although presenting a heterogeneous background, a permanent clinical or laboratory experience. Other selection criteria were the promotion of the RDs, young researchers in the field of RDs, researchers involved in translational research.

Participants coming from EU-13 countries (Estonia, Latvia, Lithuania, Poland, Czech Republic, Slovakia, Hungary, Slovenia, Bulgaria, Romania, Malta, Cyprus, Croatia) were especially encouraged to attend the course and 2 fellowships were reserved to applicants working and living in a EU-13 country.

A satisfaction questionnaire for participants was prepared.

Results and Discussion

78 applicants filled in the registration form correctly. Applications submitted after the deadline were not taken into account.

No applications were submitted from policy or patient representatives.

22 applicants confirmed attendance after selection.

3 apologized later that they could not attend the training due to lack of funding or to local situations (so 14% of the selected applicants dropped out) and 1 attendant (2% of the effectively expected participants) did not show up at the training, so that 18 participants finally attended the training course.

10 participants were researchers and 8 were clinical researcher. One of the participants was an EJP RD co-partner (Krystyna Chrzanowska, Warsaw).

The 2 fellowships were assigned to participants coming from Romania and Hungary.

Participants were from 13 different countries in particular participants from EU13 Countries came from Estonia, Latvia, Poland, Hungary, Bulgaria, Romania, Croatia

The training course was published on different media channels: Facebook and Twitter via EJP RD and KU Leuven/Center Human Genetics.

Below some of the answers of the participants from the satisfaction questionnaire:

- *"I believe the program was very well structured to cover the road topic of NGS."*

- *"I was happy to solve several problems or unanswered questions from my practice. "*

- *"I enjoyed that the speakers were interactive and adjusted the presentation to accommodate the questions from the audience."*

- *"I really enjoyed the course. The speakers were all interesting and shared their own experience with NGS and its applications in diagnostics. The setting was perfect and the organizers welcoming."*

- *"Very interesting course. all aspects covered were really to the point and very well covered"*

- *"Yes. I really enjoyed learning. There was a good balance between known and unknown information. Lecturers greatly managed to keep things at once simple and engaging. They were really competent and devoted."*

- *"I very much liked the course. It was very interesting and helped me to broaden my perspectives on NGS applications in medical genetics and possible range of difficulties you may encounter and solutions to different problems. I found the hands-on workshop very useful."*

- *"Of course, I liked it! I have some background in this area because I work with NGS and variant interpretation. However, it was an excellent opportunity*

to learn about the future of NGS and new applications of this technique. For me the talks about bioinformatics were the most relevant be-cause I don´t know much about bioinformatics."

Conclusions and next actions

The training course has been successfully delivered.

Speakers are submitting their presentations as pdfs in order to publish the training material on the EJP RD website.

Some speakers provided a short abstract of their lecture. This material will be ideally distributed via a final mailing list to the attendees, together with this abstract overview, a list of participants and the information about the published materials regarding the training course.

According to participants' feedback the hands-on session will be extended to 3/4 hours in the next training editions. The speakers of the 2019 edition convened to participate to the edition of 2020.

The next edition of the course will take place in Istanbul on 12-14 October 2020. The programme is in preparation.

The following editions of the training course will be hosted in Rome, Tübingen and Warsaw.

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