EJP RD

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EJP RD Deliverable 14.4

Second Report on course on interpretation of genetic variants and quality standards (M60)

Task Leader: KULeuven,
Participants: EKUT, ACU/ACURARE, ISS, IPCZD (CMHI), CNAG-CRG, INSERM (AMU), UMCG

Title Training Course of Task 14.2:

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

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, titled "Quality assurance, variant interpretation and data management in the NGS diagnostics era" is the object of the present deliverable that 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 Leuven university.

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, and 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 editions of the course held in 2020 (online, organized by ACU/ACURARE Istanbul), 2021 (online, organized by ISS Rome), 2022 (online, organized by EKUT Tübingen) and in 2023 (in-person organized by IPCZD, Warsaw), represented subsequent editions of the training courses of task 14.2 , of which the first edition was held at KU Leuven university in 2019. They all had the format of a 3-day training course on "Quality assurance, variant interpretation and data management in the NGS diagnostics era".

Following the pandemic situation of COVID19, three editions (at ISS Rome in 2020, at ACU/ACURARE Instanbul in 2021 and at EKUT Tübingen in 2022) have been necessarily adapted into an online format, thanks also to the availability of the speakers and organisers. We are glad that in 2023, the course in Warsaw was an in-person edition again.

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 shown 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. Next to speakers from the local institute, different speakers from Leuven were involved in each of the courses, due to great expertise on the subject. Different speakers, selected from among EJP RD Partners in Belgium, Turkey, Italy, Germany, Poland and Spain, were involved.

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 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 .

Selection of participants

The participants that were selected to attend the training course had, although presenting an 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 and Turkey (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 and Turkey.

A satisfaction questionnaire for participants was used after each edition of the course.

Results and Discussion

An overview of the number of trainees in Y2-Y5:

	Selected # candidates	# participated	Countries
Istanbul (2020-online)	38 /75	34	Austria, Belgium, Bulgaria, Cyprus, France, Ireland, Italy, Latvia, Lithuania, Malta, Ireland, Poland, Portugal, Romania, Spain, TK, Russia
Rome (2021-online)	37	26	Germany, Hungaary, Italy, Brasil, Romania, Portugal, Estonia, France, Germany, Israel, India,
Tübingen (2022-online)	36 /83	26/36 attended for 3days	Lithuania, Germany, Italy, TK, Spain, Portugal, Romania, Brasil, Belgium, Poland, UK, Finland, Ireland, France, Norway, Mexico, Sudan
Warsaw (2023 – in-person)	29/42	26	**Poland, Cyprus, Slovakia, Lithuania, Romania, Austria, Ireland, Germany**

The satisfaction surveys were highly positive. Some comments from the satisfaction surveys in Y2-Y5:

- *"Like the possibility to get acquainted of how other institutions perform the testing and (or) validation"*

- *"The speaker's presentations were very explanatory"*

- *„Like the utility of RNASeq in identifying unsolved cases and quality reassurance session"*

- *„Experts from the field share successes and failures"*

– *„Like the entire organization, the speakers and the content of the presentations"*

– *„Speakers from different fields (bioinformatics, doctors, labororatory staff), varied programme"*

- *"I found out who is involved in NGS diagnostics and what is the level of NGS diagnostics in Europe"*

- *"It allows me to know better about basics of NGS & variant classification and was great to be aware of progress in this field*

Some comments for improvement:

- *Still a need for longer hands-on sessions*

- *Major need for longer hands-on sessions or even longer training courses*

- *General need of more info on bio-informatic tools, which was implemented to the last course in 2023*

- *Need of future training courses on "Quality assurance, variant interpretation and data management in the NGS diagnostics era"*

Conclusions

This deliverable is focus on the 4 editions held from 2020 till 2023 but all the 5 training editions planned in the Grant have been successfully delivered (one per year).

Speakers were submitting their presentations as pdfs in order to publish the training material on the EJP RD website, in close folder with limited access to participants.

Some speakers provided a short abstract of their lecture. This material is offered to the attendees, together with this abstract overview and a list of participants.

According to participants' feedback after the first edition, the hands-on session was extended in time in the next training editions.

The course content was adapted annually according to the evolutions in genomics and NGS diagnostics era.