First course basics in human genetic diagnostics – A course for Clinical Laboratory Geneticists (CLGs) in education, Nicosia, Cyprus, 20.-24. June 2016

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The first course “Basics in human genetics diagnostics - A course for CLGs in education” took place in the Cyprus Institute of Neurology and Genetics in Nicosia (CING) and was organized by Drs. Thomas Liehr (Jena, Germany), Leonidas A. Phylactou and Carolina Sismani (both Nicosia, Cyprus). The course was enabled in first place by a generous sponsoring of the European Society of Human Genetics (ESHG), being interested in a sound education of Clinical Laboratory Geneticists (CLGs) in all (European) countries. Also CLGs coming from countries without a national education scheme for CLGs need such courses to be eligible for the European registered CLG title (ErCLG – see https://www.eshg.org/clg.0.html). Besides, the support from Carl-Zeiss Jena, DAKO – an Agilent Technologies Company, Illumina, MetaSystem, MRC-Hollan, Oxford Gene Technology and Cytocell and ZytoVision GmbH and the fees of the overall 30 participants enabled the realization of this five day course. Nonetheless, the financial frame would have been still extremely dense, if CING would not have provided a lecture hall free of charge, and if not all speakers just claimed their expenses and did not ask for being paid. Thanks for all these efforts to all involved parties!

Before talking about the course itself, I would like to thank again CING for hospitality and well organization of everything around the course – main work was done here by Carolina Sismani with support of Leonidas A. Phylactou and his CING team. The course schedule developed together by Thomas Liehr, Leonidas A. Phylactou and Carolina Sismani, was dedicating each of the five days to a special field. Every day was divided to four major parts: two of them were lecturers about theoretical basics of certain topics; one part was a workshop with discussion of possible situation of case analysis using genetic diagnostic tools; each day ended with a written test, where participants were asked to answer to several questions concerning day topics. In 4/5 tests participants needed to have >50% of the available points, which was managed by all of us.

Day 1 was devoted to the basics of human genetics, the history of genetic diagnostics from

karyotyping to NGS, and the inheritance patterns. Here, the participants got a chance not only to listen about genetic counselling and syndromology, but also due to a workshop to empathize themselves in the place of counselor and a patient, construct a family pedigree and perform own risk assessments.

Day 2 was devoted to cytogenetics from history to application in diagnostics and research. During the workshop participants were asked to do a karyotype and to pass a quest about ICSN nomenclature.

Day 3 was about molecular cytogenetics from FISH to array-CGH, with introduction to main approaches in these fields and discussion of possible simple and complex cases results. During a workshop on array CGH, case-results were discussed. Lecturers about basics of molecular genetics, knowledge of monogenic syndromes and PCR techniques completed this day.

The presentation of PCR techniques and their possible applications in diagnostics was continued in the day 4. Knowledge about MPLA and related approaches was given to the participants with several case presentations and deep analysis and discussion of the results.

Day 5 was the last one and was very intense. We heard about sequencing techniques, NGS and their application, non-invasive prenatal diagnostics, biochemistry genetics and metabolic disorders, basics of epigenetics and gene therapy, futures of diagnostics using proteomics knowledge. After a very interesting presentation made by Helena Kääriänen about future of human genetics services we all could exchange and discuss about this topic from the view of the different countries being represented by the participants, i.e. Armenia, Bulgaria, Canada, Cyprus, Finland, Greece, Italy, Latvia, Romania, Russia, Sweden, Ukraine, and USA.

Although we had an intense course and tight scheduled of lectures and workshops, There was also always well planned breaks and thus time for inter-participant and lecturer conversations; and this is most important for communication with other scientists and colleagues.

In conclusion I would like to thankful all organizers for this course that provided a comprehensive introduction to genetic diagnostic tools that maybe helpful for beginner CLGs to choice his/her own future specialty and for advanced researchers to refresh knowledge and keep update.

Figure:

Participants and organizers of the course in the lobby of the CING.