3Gb-Test NGS Course, April 20 - 23, 2015, Prague - Report

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In April 2015, I attended the 4-day NGS course titled: "Next-Generation Sequencing in a Diagnostic Setting" in Prague, Czech Republic.

The NGS course was very well organized and, even though I came to it as a beginner, I had no problems advancing through the presentations thanks to the carefully planned and highly motivating introduction to the NGS world given on the first day. The introduction included: NGS terminology; information on the types of instruments being used in NGS; type of variants; how properly work in the wet-lab; and, an in-depth view of exome sequencing.

In the days following the introduction, we were shown informative presentations and provided with key information or tips not often available in user manuals including how to: accurately deal with NGS in detail; handle variant calling; and, deal with the data, results and unsolicited results. Moreover, the speakers took us through the importance of considering the ethical aspects of NGS, guidelines and ISO accreditation for diagnostic laboratories.

The quality of the speakers was excellent and I greatly appreciated the many opportunities we had to converse with them and ask questions. At the end of the course, a section was dedicated to Questions & Answers, where we were able to discuss any questions or comments we were unable to solve before then.

In addition to the information we received in the presentations, there were practical "hands-on" sessions on how to find variants after receiving all the data from NGS in different platforms (e.g. CLCBio, Phenosystems, Ingenuity and LOVD) allowing us to understand and integrate the valuable information learned in the presentations.

As a newcomer to NGS, I did have two major concerns about NGS: one was the technical aspects on the workflow to obtain successful results with NGS; and the other one, was how to apply NGS in a diagnostic setting caring for patient data and give quality results. Therefore, I came into the course with many expectations and those expectations were definitely met.

My overall feeling after completing the course is that there is still a place for improvement to apply NGS in genetic diagnostics, however, I am confident it is moving in the right direction.

One final note: The most important message I got from this course was: "please, share your data!", we all need to build good (complete/extensive/thorough) databases to help obtain the best diagnosis we can for patients in need. NGS is definitely the future in medicine and these types of excellent courses are essential and a must for people involved in genetic diagnostics.