NGS course - Next-generation sequencing in a diagnostic setting

September 8 – 11, 2014
Divani Acropolis Hotel, Athens Greece

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DAY 1 September 8

14.00 Welcome
(Joanne Traeger-Synodinos, Athens, Greece)

14.10 Introduction; NGS terminology, types of variants
(Johan den Dunnen, Leiden, The Netherlands)

14.45 Characteristics of the sequencing methods - Illumina, Ion Torrent, SOLiD, Pacific Biosciences *(tbc)*

15.30 Break

16.00 The critical steps in the process; based on 3Gb-test deliverable D1.1
(Erika Souche, Leuven, Belgium)

16.45 Making the DNA sequence-ready; wet lab procedures
(Bart Jansen, Leiden, The Netherlands)

17.30 Close

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DAY 2 September 9

09.00 PCR based targeting; gene set analysis
(Erika Souche, Leuven, Belgium)

09.45 Exome sequencing; targets, hybridisation capture
(*Hans Scheffer, Nijmegen, The Netherlands)*

10.30 Break

11.00 The data and how they flow; formats, pipelines, storage and maintenance
(Jeroen Laros, Leiden, The Netherlands)

11.45 ELSI aspects; ethical, legal and social issues
(Anne Cambon-Thomsen, Toulouse, France)

12.30 Break

14.00 Calling variants; from simple (SNVs) to complex (SVs)
(Christian Gilissen, Nijmegen, The Netherlands)

Afternoon practicals
Spaces limited, participants should bring their own computer

14.45 Practicals

15.30 Break

16.00 Practicals

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3Gb-TEST Regional Meeting - Next-generation sequencing and genomic applications in disease and health: has the future arrived?

Acropolis Museum, Athens, Greece
17.00-21.00

Chairs: Emmanuel Kanavakis (University of Athens, Greece), Yannis Georgiou (University of Ioannina, Greece)

17.30 Introducing NGS diagnostic applications in human genetics: the 3Gb-test project
(Bert Bakker, Leiden, the Netherlands)

18.00 NGS in clinical diagnostics: current trends
(Hans Scheffer, Nijmegen, The Netherlands)

18.30 NGS for free fetal DNA analysis
(Elles Boon, Leiden, The Netherland)

19.00 *Refreshment Break*

Chairs: Sohia Kitsiou-Tzeli (University of Athens, Greece), Aspasia Tsezou (University of Thessaly, Greece)

19.30 The applications of next generation sequencing in the analysis and study of single cells. *(Thierry Voet, KU Leuven, Belgium)*

20.00 Ethical aspects of NGS at the intersection of research and clinics
(*Eva Winkler, Heidelberg, Germany)*

20.30 Future trends, a discussion
(Johan den Dunnen, Leiden, The Netherlands)

21.00 Closing of meeting
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DAY 3 September 10

09.00 Thresholds for calling variants *(Chris Mattocks, Salisbury, UK)*

09.45 Annotation and prioritization of variants; incl. prediction software
(Periklis Makrythanasis, Geneva, Switzerland)

10.30 Break

11.00 Lessons learned from cystic fibrosis: from the CFTR1 to CFTR2 database
(Milan Macek, Prague, Czech Republic)

11.45 Share your findings; DNA diagnostics = share variants & phenotype
(George P. Patrinos, Patras, Greece)

12.30 Break

14.00 Whole genome versus whole exome sequencing; pros/cons both methods
(Christian Gilissen, Nijmegen, The Netherlands)

Afternoon practicals
Spaces limited, participants should bring their own computer

14.45 Practicals

15.30 Break

16.00 Practicals

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DAY 4 September 11

09.00 Functional validation of variants; RNA, protein, expression, functional assays
(Bert Bakker, Leiden, the Netherlands)

09.45 de novo genome assembly versus mapping to a reference genome as the method to use to identify the variants present
(tbc)

10.30 Break

11.30 Future developments
(Bart Jansen, Leiden, The Netherlands)

11.45 Evaluation of course and Q&A session

12.30 Meeting ends