**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