Clinical and quality issues when introducing new technologies in genomics

Friday May 30, 2014
ESHG Conference - Satellite meeting
MiCo Milan, Italy

Registration Fee: EUR 50.-

PROGRAMME

09.00	Registration
09.30	Welcome Coffee
10.00-10 :30	Get together and introduction
10.30-11.00	**New technologies, new possibilities** *Hans Scheffer (Department of Human Genetics, Radboud university medical center, Nijmegen, The Netherlands)* How is diagnostics changing medical practice? Diagnostic utility and (preliminary) evidence for better care.
11.00-11.30	**The challenges of Clinical reporting: What findings should be fed back to patients and families? Insights from the Deciphering Developmental Disorders Project** *Wendy Jones (Wellcome Trust Sanger Institute, Hinxton, UK)* On the clinical challenges, and how to report results and other findings to patients and families.
11.30-12.00	**New technologies, new quality issues** *Els Dequeker (Biomedical Quality Assurance Research Unit, Department of Public Health and Primary Care, University of Leuven, Belgium)* How to adapt a quality system to a novel technology? Don't forget that existing rules still apply.
12.00-12.30	**A contribution to European Guidelines on diagnostic NGS** *Gert Matthijs (Center for Human Genetics, University of Leuven, Belgium) on behalf of EuroGentest's working group on guidelines for diagnostic NGS.*
12.30-13.00	**Cross border testing in Europe – several problems to be solved?** *Pia Pohjola and Helena Kääriäinen (National Institute for Health and Welfare, Helsinki, Finland) on behalf of the European Union Committee of Experts on Rare Diseases (EUCERD).*
13.00-14.00	Lunch

Workshop 1: Genomic data to laboratory report: tools and quality issues

14.00-15.30

Moderated by Bert Bakker, Jan Traeger-Synodinos and Ros Hastings
Practical workshop to share emerging guidelines on reporting and collect evidence from the field (i.e. how people practice it currently).This workshop will discuss whole genome analysis (NGS and microarray) and cover variant calling parameters, quality control, confirmation/follow up, predicting functional effects of DNA variants and the limitations of NGS/microarray analysis and laboratory report content.

15.30 – 16.00

Coffee Break

Workshop 2: Ethical and societal aspects of genome diagnostics

16.00 – 17.30

Moderated by Anne Cambon-Thomsen (Toulouse) and Martina Cornel (Amsterdam)
What kind of guidance do we need to properly introduce NGS into medical practice? Practical workshop to collect participant's views after an Introductory talk on issues and existing frameworks for translation of WGS into clinical practice and two discussant presentations on examples of experiences in national contexts (UK and Italy).

16.00 – 16.20
Introductory talk (based on a work performed within 3Gb-TEST)
Recommendations for whole genome sequencing – putting the cart before the horse?
Samantha Leonard (Inserm, University Toulouse III Paul Sabatier and hôpitaux de Toulouse, France, 3GbTEST)

16.20 – 16.35
'Realising Genomics': the ethical, legal and social challenges. A PHG Foundation perspective
Alison Hall (PHG Foundation, Cambridge, UK**) –** Discussant -

16.35 – 16.50
An Italian clinician experience and views
Francesca Forzano, Galliera hospital, Genova, Italy - Discussant

16.50 – 17.30
General round table discussion
introduced by *Anne Cambon-Thomsen* (5 min) and concluded by *Martina Cornel* (5 min)

17.30-18.00

Concluding remarks