

**Programme 2nd Joint TECHGENE and EuroGentest (EUGT2) meeting
'PROGRESS IN QUALITY ASSURANCE AND TECHNICAL DEVELOPMENTS
IN GENETIC TESTING'**

Thursday, January 19, 2012, Nijmegen (The Netherlands)

➤ 08.00 - 09.00 Registration / Coffee

Highlights of the TECHGENE project

- 09.00 - 09.10 Hans **SCHEFFER** (Radboud University Medical Centre, Nijmegen):
Welcome and brief introduction to TECHGENE
- 09.10 - 09.35 Marcel **NELEN** (Radboud University Medical Centre, Nijmegen):
"Exome sequencing in clinical practice"
- 09.35 - 09.55 Anne **CAMBON-THOMSEN** (INSERM, Toulouse):
"Ethical considerations on genome sequencing: results of the survey"
- 09.55 - 10.15 Christian **GILISSEN** (Radboud University Medical Centre, Nijmegen):
"Data analysis and variant prioritization in exome sequencing"

➤ 10.15 - 10.45 Coffee

Highlights of the EuroGentest2 project

- 10.45 - 10.55 Gert **MATTHIJS** (University of Leuven):
Brief introduction to EuroGentest
- 10.55 - 11.10 Sarah **BERWOUTS** (University of Leuven):
"Report on the EUGT survey on quality assurance in European genetic laboratories"
- 11.10 - 11.20 Dragica **RADOJKOVIC** (Laboratory of Molecular Biology, Belgrade):
"Implementation of QA in eastern European laboratories"
- 11.20 - 11.30 David **BARTON** (National Centre for Medical Genetics, Dublin):
"The importance of the (revision of) the IVD Directive for genetic laboratories"
- 11.30- 11.40 Joerg **SCHMIDTKE** (Institute of Human Genetics, Hannover):
"Clinical Utility Gene Card: a tool for quality in genetic diagnostics"

Keynote lecture

- 11.40-12.15 Cecile **JANSSENS** (Erasmus University Medical Centre , Rotterdam):
"Understanding the results of (commercial) susceptibility tests"

➤ 12.15-13.30 Lunch

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Workshop 1: "Prenatal and neonatal genomic screening?"

- 13.30 - 14.00 Ghislaine **CLERET DE LANGAVANT** (Commissaire à la santé et au bien-être, Montréal):
"Experiences of a nation-wide prenatal screening program for trisomy 21"
- 14.00 - 14.20 Marta **RODRIGUEZ DE ALBA** (Fundación Jiménez Díaz, Madrid):
"NIPD (Non Invasive Prenatal Diagnosis): are we ready to move from rare monogenic diseases and sex determination to aneuploidy screening?"
- 14.20 - 14.40 Rosalind **HASTINGS** (John Radcliffe Hospital, Oxford):
"External quality assessment for genome-wide assays: lessons from a scheme on constitutional molecular karyotyping"
- 14.40 - 15.00 Chris **MATTOCKS** (NGRL Salisbury):
"Quality assurance in NGS"
- 15.00 - 15.30 **Coffee**

Workshop 2: "Future developments in diagnostic testing – What is next?"

- 15.30 - 17.00 Forum discussion: A discussion of current and future developments in diagnostic testing in genetics, including NGS-based diagnostics and in vitro diagnostics: what is expected and needed, and what will be offered in the (near) future? A follow up to a discussion on NGS held two years ago.

Representatives from patient organizations, clinical geneticists, clinical laboratory geneticists, genome research laboratories, genetic societies, and commercial companies in medical technology.

Panelists: David **BARTON** (National Centre for Medical Genetics, Dublin), Martina **CORNEL** (VU Medical Centre, Amsterdam), Lisbeth **TRANEBJÆRG** (University of Copenhagen), Joris **VELTMAN** (Radboud University Nijmegen Medical Centre), Ning **LI** (BGI Europe, Copenhagen), Elsbeth **VAN VLIET** (VSOP, Dutch Patients Alliance for Rare and Genetic Conditions).

Moderator: Han **BRUNNER** (Radboud University Nijmegen Medical Centre).

- 17.00 **End of the symposium**