Newsletter December 2011

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YOUR NEWS

Are you a EUGT2 participant? Let us know if you are involved in upcoming evens or have a new publication. We will publish this on the website and in the newsletter!

IN SHORT

EuroGentest2 is a European coordination action for the harmonization and further improvement of genetic services, especially genetic testing across Europe.

UPCOMING EVENTS

- Joint EuroGentest/TECHGENE Scientific Symposium, 19 Jan
- 6émes Assises de Génétique Humaine et Médicale, 2-4 Feb
- Up Close and Personalized, International Congress on Personalized Medicine, 2-5 Feb
- VII International Conference on Rare Diseases and Orphan Drugs, 4-6 Feb
- Workshop: An introduction to Quality Assurance in Genetic Diagnostic Laboratories, 6-7 Febr and 9-10 Feb
- Rare Cancers Conference 2012, 10 Feb
- BASIC WORKSHOP - Accreditation for beginners, how to implement ISO 15189, 23-24 Feb
- RE(ACT) Congress, 29 Feb-2 Mar

PARTICIPANTS ONLY

Documents for EUGT2 participants are found on the participant page of the EuroGentest website.

If you cannot access these documents, make sure you are registered on the website and contact our webmaster for participant access.

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! RhesusD genotypic workshop: Call for participants

Part of the remit of EuroGentest is to standardise and disseminate non-invasive prenatal diagnosis in EU.

We will be running a RHD genotyping workshop as part of the programme of activities of EuroGentest. This will involve the provision of reagents and maternal plasma samples from Rh negative woman for participants to then carry out DNA extraction and real time PCR arrays in their own laboratories. The *RHD* assay to be used was one previously standardised in the SAFE network of excellence, and involves the analysis of RHD exons 5 and 7 by real time PCR.

Participant labs will be expected to provide their own DNA extraction kits and methodology. We would prefer the use of Qiagen's QIAamp® Circulating Nucleic Acid kit but understand that not all labs may have access to the required vacuum manifold. Results will be assessed centrally with the intention of publication, with a meeting to discuss outcomes in Plymouth UK mid to late 2012.

For those interested in involvement please fill in the form available on the website and send to Prof. Neil Avent (neil.avent@plymouth.ac.uk). To download the form click here.

Meeting report - 3rd Meeting of the CAPABILITY-Network and the Institute for Health and Consumer Protection (IHCP) on Genetic Testing in Emerging Economies (GenTEE)

The GenTEE (Genetic Testing in Emerging Economies) consortium held its 3rd meeting on 28th October to 1st November, 2011, in Beijing, P.R. China. The meeting was organized locally by Prof. Nanbert Zhong, Center of Medical Genetics, Peking University Health Science Center, Beijing, P. R. China and internationally by the EU Joint Research Institute in Ispra, Italy, the Institute of Health and Consumer Protection (IHCP), and the Women's Health Research Unit at the Westfaelische Wilhelms-Universitaet (WWU), Muenster, Germany.

GenTEE is an international networking partnership that brings together geneticists from Europe, and nine International Cooperation Partner Countries (ICPC) including:

Argentina, Brazil, China, Ecuador, Egypt, India, Oman, Philippines and South Africa and EuroGentest2 WP 8 ("Best Practice Guidelines for Provision of Clinical Genetic Service") to

- document and compare current practice and the state of genetic service provision in the nine ICPC
- accelerate information exchange on best practices among countries/regions

GenTEE and its networking activities stand in the tradition of previous projects funded by the European Commission on the collection of comparative data on genetic services development, namely the "Concerted Action on Genetics Services in Europe" Survey (CAGSE, funded by FP5), the Institute for Prospective Technological Studies (IPTS) Survey and Report "Towards quality assurance and harmonisation of genetic testing services in the EU" (2003) that led to EuroGentest(1) and the CAPABILITY project, a Specific Support Action for EuroGentest(1).

The major objective of the workshop was to discuss the preliminary outcome of a joint survey on genetic service and testing development in the nine participating ICPC and to prepare the survey outcome report to be published by the IHCP in spring 2012.

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Meeting report - EuroGentest/CanGèneTest Ancillary event at the 12th ICHG/ASHG Meeting

At the 12th International Congress of Human Genetics held in Montreal in October 2011, EuroGentest2 and APOGEE-Net/CanGèneTest jointly organized a session entitled "Challenges in the Translation of Genomic Innovations into Clinical Care". The session was opened by François Rousseau (Université Laval) who presented the APOGEE-Net/CanGèneTest Network and its role in addressing the challenges of translating genomic and genetic innovations into the health care system. Gert Matthijs (University of Leuven) presented the EuroGentest Network and its role in the improvement and standardization of quality in genetic testing. Joris Vermeesch (University of Leuven) described two pilot studies of external quality assessment of constitutional molecular karyotyping, and Helena Kääriäinen (University of Helsinki) described the impact of the increasing importance of genetic testing in health care. Daniel Reinharz (Université Laval) presented cost/effectiveness and cost/utility simulations for selected genetic and genomic health innovations, and Renaldo Battista (Université de Montréal) discussed the role of health technology assessment in the translational process.

The session was a success as it attracted over 60 participants despite the concurrent presentation of several other events, underlining the importance of these questions to the medical and scientific community. One of the conclusions of this event was that there is strong complementarity between the activities of each Network and that there is room for even more synergy between the different teams working in this area. More information on the session can be obtained by following this link:

http://www.eurogentest.org/web/info/public/ICHG_AncillaryEvent2011.xhtml

Meeting report - Joint expert meeting WP7&8 (Unit 2) EUGT2 report – Zaandam (Holland) November 2011

A productive meeting with 26 enthusiastic experts was organized by the researchers from work package 7&8 (Unit 2) of Eurogentest2 (EUGT2). Subjects of discussion during the meeting were quality in genetic counselling for pre-symptomatic testing and best practice for provision of clinical genetic service.

Work package 8 leader Martina Cornel opened with an overview of goals of EUGT2 and the objectives of the meeting. This was followed by three presentations of experiences with testing for monogenetic subtypes. The presentations included examples from oncology (Ignacio Blanco), cardiology (Johan Brandt) and MODY (Maggie Shepherd) and showed that different fields ask for different approaches and have their own opportunities and threats when it comes to good genetic services. Marcel Nelen then presented his experiences with Next Generation Sequencing (NGS) for diagnostics, which described the strengths and weaknesses of the method and gave rise to a discussion about informed consent.

After an entertaining evening programme and a good night's sleep, two parallel workshops were chaired by Heather Skirton (WP7), Ulf Kristofferson and Martina Cornel (WP8) with the goal of describing principles of good practice for the two work package subjects. Main themes discussed were: principles, objectives, logistics and content of genetic counselling for pre-symptomatic testing and informed consent for NGS, cooperation between different actors in genetic services and informing relatives.

A more detailed report of the discussions and conclusions will soon be available on the website.

! Newly published Clinical Utility Gene Cards for:

Acrodermatitis enteropathica – *Dec 2011*

Alport syndrome – *Dec 2011*

Trimethylaminuria – *Nov 2011*

To download click here.

Other news

- Connect to EuroGentest on LinkdIn
- In the participant section of the EuroGentest website, some interesting general documents from the **EC website** are highlighted and you can find information on the 18 month reporting and on time recording *(participants only)***.**