Genetic Testing in Europe
Network for test development, harmonization, validation and standardization of services

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Newsletter November-December 2013

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

- E-course on the new version of ISO 15189, 10 Jan
- 25 th Annual Meeting of the German Society of Human genetics, 19-21 Mar
- Human Genome Meeting 2014, 27-30 Apr

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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Do you wish to receive this newsletter and stay up to date with the activities of EuroGentest2? Please register on our website.

The future of EuroGentest

EuroGentest has been providing services to the genetics community since 2005. December 31st 2013, the funding offered by the European Commission will come to its end. We would like to thank all the EuroGentest partners for their continuous efforts in working towards our common goal: good quality in genetic testing across borders in Europe!

The EuroGentest project will not end for good. EuroGentest will integrate with the European Society of Human Genetics. We look forward to continue and serve the genetics community in this a effort with the ESHG from 2014 on!

See you again in 2014!

The EuroGentest Management team in Leuven (Belgium)

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EuroGentest E-courses

In January a new E-course will be available "on the new version of ISO 15189". This will be the fourth e-course of EuroGentest.

Up till now 137 unique participants from 49 different countries participated in our online courses. Each course consisted of a live or recorded presentation, a quiz and a forum discussion. We have noticed that there still is a high demand for information on quality management and working towards accreditation and validation. After a first evaluation we can conclude that a live presentation is preferred by the participants. Also the discussion after the webinar and possibility to ask live questions to the presenter are highly appreciated and are more interactive then the discussions on the forum, which are more difficult to maintain. The quiz on the other hand is a good and easy tool to test knowledge and is frequently used. In the future we will try to offer you new e-courses to provide you more information on quality in an easy accessible and interactive way.

- January 10th, 14:30h (CET) – E-course on the new version of ISO 15189 – Register here!

Two major publications of the GenTEE report and CAPABILITY country reports

The European Commission's Joint Research Centre (JRC) published the final report of the GENTEE project, entitled "Genetic Testing in Emerging Economies", which represents the first worldwide effort to systematically survey and assess the current state of medical genetic services in emerging economies. Click here for the report.

Shorter versions of single country reports were published in a special issue "CAPABILITY and "Genetic Testing in Emerging Economies (GenTEE)" of the Journal of Community genetics (Volume 4 Number 3 July 2013). Click here for this special issue.

23andme

23andme has halted sales of its health-related consumer genomics service after a warning letter from the FDA. This signals a new milestone in the road to a coherent regulatory framework for consumer genomics. It is now over a year since 23andme filed for regulatory approval and the FDA's letter reveals that things have not been going well between the company and the agency. The FDA's action has sparked discussion about what standards they are requiring 23andme to meet. The specifics are shrouded in the confidentiality of the regulatory process but when the FDA held an advisory panel on consumer genomics in March 2011 the critical question it posed to the assembled experts was whether this class of tests should be held to the FDA's statutory standard i.e. should be able to provide "clinically significant results". Unsurprisingly the panel was not willing to operate a policy of genetic exceptionalism for consumer genomics and affirmed that this standard should be applied. But genetic exceptionalism was precisely what 23andme were asking for at the meeting: they suggested that FDA needed to redefine clinical validity to deal with their class of tests. The FDA's new warning letter suggests that much of the tension between company and agency is at this sticking point.

Stuart Hogarth

PEGASUS

The PEGASUS project (for PErsonalized Genomics for prenatal Aneuploidy Screening USing maternal blood) proposes to carry out an independent study that will validate the performance and utility of new genomic technologies for screening for aneuploidies in pregnant women using fetal DNA present in the maternal blood. The team of researchers will identify an evidence-based cost-effective approach for implementation of this new technology in the Canadian health care system. They will develop decision-making tools that will assist couples in making informed decisions, as well as educational tools for health care professionals, all integral components of the implementation of genomics-based non-invasive prenatal diagnosis. The deliverables of this project will enable decision makers, pregnant women and their partners to make informed choices pertaining to prenatal genetic screening and diagnosis, such as screening for Down syndrome, and reduce the risk to pregnancies associated with amniocentesis.

More information can be found on: www.pegasus.nhs.uk

Yves Labelle, CanGèneTest (www.cangenetest.org)

! New Clinical Utility Gene Cards

Hyperlipoproteinemia, TYPE II – November 2013

Click here for the full list of CUGC's.

Publications

By EuroGentest members:

- Current issues in medically assisted reproduction and genetics in Europe: research, clinical practice, ethics, legal issues and policy European Society of Human Genetics and European Society of Human Reproduction and Embryology by Heather Skirton, Helena Kääriäinen, Milan Macek Jr., Martina Cornel, Joyce Harper, Mike Morris, Pascal Borry, Jorge Sequeiros and others.
- Early evaluation and value-based pricing of regenerative medicine technologies by Wolf Rogowski and others.
- Can I get a retweet please? Health research recruitment and the Twittersphere. By Heather Skirton, Leigh Jackson, Lesley Goldsmith and others.
- An iterative consensus-building approach to revising a genetics/genomics competency framework for nurse education in the UK by Heather Skirton and others.
- Implementing noninvasive prenatal fetal sex determination using cell-free fetal DNA in the United Kingdom by Celine Lewis, Lyn Chitty and others.
- A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases by Hans Scheffer and others.
- Utility and limitations of genetic/genomic information and testing by Heather Skirton and Leigh Jackson.
- Non invasive prenatal diagnosis of aneuploidy: next generation sequencing or fetal DNA enrichment? By Neil Avent and others.
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For your interest:

- Non-invasive prenatal testing using massively parallel sequencing of maternal plasma DNA: from molecular karyotyping to fetal whole-genome sequencing
- The Effects of a Genetic Counseling Educational Program on Hereditary Breast Cancer for Korean Healthcare Providers
- Perspective on the Technical Challenges Involved in the Implementation of Array-CGH in Prenatal Diagnostic Testing
- A Genetic Counselor's Guide to Using Next-Generation Sequencing in Clinical Practice
- ACMG Standards and Guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: revision 2013
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Other

- Connect to EuroGentest on LinkdIn 
- Read more about EuroGentest