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

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Newsletter October 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

- NGS 2013 Manchester: Applications & Bottlenecks, 5-6 Nov
- 14th EMBO/EMBL Science & Society Conference: Public and Private Health – Genomics, Medicine and Society, 7-8 Nov
- Epigenomics of Common Diseases, 7-10 Nov
- 5th Pan Arab Human Genetics Conference (PAHGC) and the Golden Helix Symposium 2013, 17-19 Nov
- **BASIC WORKSHOP - Validation of diagnostic tests in clinical molecular genetics, 21 Nov**
- 5th TECHGENE knowledge network meeting: Implementation of new Next Generation Sequencing techniques, 21 Nov
- **EuroGentest workshop on European guidelines for Next Generation Sequencing, 21 Nov**
- **E-Course on validation, 29 Nov**

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

New feature of the CUGC's: NGS panel database

Unit 2 of the EuroGentest2 initiative commissions the establishment of the Clinical utility gene cards (CUGCs). CUGCs are disease-specific guidelines regarding the clinical utility of genetic testing. They cover all elements relevant for assessing risks and benefits of genetic test application.

With the long-term goal of adapting the CUGC format to next-generation sequencing (NGS) technologies, we have built up a NGS panel database. In this database we present data from NGS providers including panel name, tested genes and disease of interest. Our main purpose is to give users the opportunity to quickly identify diagnostic options according to the different search terms: disease, gene, OMIM number, panel name, provider and Orpha number. Any overlap or gap of tested genes between different panels can easily be determined by comparison.

As of October 11 2013, we identified 25 laboratories having launched a total of 776 clinical NGS tests covering 2236 genes and 1114 diseases. The list will be updated regularly. A prototype of our data collection is available at our project website: http://www.eurogentest.org/index.php?id=668

We encourage NGS providers from the commercial and academic sectors to contact us regarding their current services in order to include them in this database: eurogentest@mh-hannover.de

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EuroGentest workshop: Implementation of clinical genetic databases

This workshop will provide training for current and future implementers of accessible or shared databases for collection of actionable clinical genotype and phenotype data. Training will cover:

- Tools and resources used in the process of creating and running a database
- Standards important for databasing
- Strategies for successful planning and implementation of a database project

The workshop will take place **December 17** and will be organised by NGRL Manchester (Manchester Centre for Genomic Medicine) as part of the EuroGentest project.

The workshop will take place in **Nowgen's** dedicated bioinformatic training suite and will include practical computer-based training and group exercises to explore the resources available and the issues relating to sustainable database implementation.

Click here for more information and to register for the workshop.

EU Regulation on in-vitro Diagnostic Medical Devices – information on voting in EP

The European Commission issued a proposal for a new Regulation on in vitro diagnostic devices in September 2012. At the committee stage in the European Parliament, the Rapporteur Peter Liese, MEP introduced an entirely new article into this proposed Regulation. The article is focused on the practice of genetic medicine, and seeks to regulate what happens in clinics and hospitals where patients may have a genetic test as part of their medical care.

In May 2013, the European Society of Human Genetics issued a position statement opposing this new article, stating that the provisions of the article were "unworkable in the daily practice of genetic medicine."

A version of the Regulation containing this new article was recently passed by the committee, and will be voted on at the European Parliament on October 21. It is very important that this article is removed at the European Parliament vote, or it may become European law.

Click here for more information.

participant access.

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! New Clinical Utility Gene Cards

Huntington's disease - Oct 2013

16p13.11 microdeletion syndrome – Oct 2013

Xeroderma pigmentosum – Oct 2013

Cystinosis – Sep 2013

von Hippel-Lindau (VHL) – Aug 2013

Progressive familial intrahepatic cholestasis type 1 – Aug 2013

Progressive familial intrahepatic cholestasis type 2 – Aug 2013

Progressive familial intrahepatic cholestasis type 3 – Sep 2013

Choroideremia – Aug 2013

Click here for the full list of CUGC's.

Publications

By EuroGentest members:

- Criteria for fairly allocating scarce health-care resources to genetic tests: which matter most? By Wolf Rogowski, Joerg Schmidtke and others.
- Patient compliance based on genetic medicine: a literature review by Joerg Schmidtke and others.
- Public attitudes towards preventive genomics and personal interest in genetic testing to prevent disease: a survey study by Martina Cornel and others.
- Offering prenatal diagnostic tests: European guidelines for clinical practice guidelines by Lyn Chitty, Heather Skirton, Lesley Goldsmith, Leigh Jackson and Celine Lewis.
- Fair allocation of health-care resources: finding a model that does not disenfranchise users of genetic services. A commentary on Rogowski et al.... by Heather Skirton.
- Factors affecting the clinical use of non-invasive prenatal testing: a mixed methods systematic review by Heather Skirton.
- Dispelling myths about rare disease registry system development by Ségolène Aymé and others.
- Current landscape and new paradigms of proficiency testing and external quality assessment for molecular genetics by Rob Elles and others.

For your interest:

- Predictive genetic testing for complex diseases: a public health perspective
- Direct-to-Consumer Genetic Testing: A Comprehensive View
- Genomic Counseling: Next Generation Counseling
- ACMG Standards and Guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: revision 2013
- Use of preimplantation genetic diagnosis for serious adult onset conditions: a committee opinion

Other

- Connect to EuroGentest on LinkdIn
- Read more about EuroGentest