



# Newsletter September 2013



## YOUR NEWS

Are you a EUGT2 participant? [Let us know](#) if you are involved in upcoming events 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

- 29. Ernst Klenk Symposium in Molecular Medicine 29 sep-1 Oct
- Recent Developments in Neurofibromatoses and Rasopathies, 29 Sep – 1 Oct
- ATELIER DE PERFECTIONNEMENT: En bonne voie vers l'accréditation? 3-4 Oct
- Best Practice Meeting – PGD arrays, 3 Oct
- 2nd International Workshop "Rare Disease and Orphan Drug Registries, 21-22 Oct
- 63rd Annual ASHG meeting, 22-26 Oct



## 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.



## UNSUBSCRIBE

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## REGISTER

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

## Call for expressions of interest as Commission appointees to the new Commission expert group on rare diseases

The European Union Committee of Experts on Rare Diseases ([EUCERD](#)) has come to the end of its three year mandate during which they have assisted the European Commission with the preparation and implementation of Community activities in the field of rare diseases, in cooperation and consultation with the specialised bodies in Member States, the relevant European authorities in the fields of research and public health action and other relevant stakeholders acting in the field.

The European Commission acknowledges the value and importance of the tasks taken up by EUCERD and published on [30 July 2013 a Decision](#) to establish a "Commission expert group on rare diseases", clearly illustrating the continuing need for support in this area. In this decision general rules regarding the criteria and procedures to appoint members are written down.



The Commission now calls upon European level patients' organisations, professional associations, scientific societies and associations producing products or providing services in the field of rare diseases to express their interest and appoint their representatives.

[Click here for more information.](#)

## How to prioritize genetic tests?

Resources are too limited to provide all beneficial genetic testing services available the next decade. Clinics may lack personnel and have waiting lists. Low and middle income countries may depend on commercial offers to be paid by patients themselves. Ethical and economic reflection on prioritization criteria is needed.

In a consensus process oriented at the principles of "Accountability for Reasonableness" points to consider for prioritization were developed. Participants collaborated in EuroGentest and/or the Public and Professional Policy Committee (PPPC) of the European Society of Human Genetics (ESHG).

We now invite the ESHG membership to comment on the current version of the document, which will afterwards be sent to the ESHG Board for approval and to the European Journal of Human Genetics for review.

Comments in the discussion forum are welcomed until the **end of September 2013**.

--> [click here to go to the document on the ESHG website and comment on it](#)

Also, suggestions can be sent by email to [Franziska Severin](#). After approval of the ESHG Board all comments will be available from the lead author.

## EuroGentest interactive workshops

**21 Nov 2013 - [BASIC WORKSHOP - Validation of diagnostic tests in clinical molecular genetics](#)**

Meet experienced people in this field, learn about regulatory requirements (ISO 15189) or how to design a proper validation plan for new molecular genetic tests [\[more\]](#)

### Upcoming E-courses:

- End of October 2013: E-course on the accreditation process
- December 2013: E-course on the new version of ISO 15189– similarities and differences

More info on: <http://www.eurogentest.org/index.php?id=651> or mail: [workshops@eurogentest.org](mailto:workshops@eurogentest.org)

## Patients at risk of inherited cardiac conditions and sudden death - a case based course for clinical practice in primary care

If you are a primary care physician or nurse, you may already have had patients asking about inherited cardiac diseases, such as cardiomyopathy or sudden adult death due to arrhythmia.

In this course, the aim is to help you to determine the level of risk for your patients with a family history of cardiac disease, and find out how to refer and manage them. The course has been organised by a practising general practitioner and a genetic counsellor. It is entirely case based and will include conversations with patients about care.

This course is residential, running from 1700hrs on Wednesday 13th November 2013 and finishing on Friday 15th November at 1400hrs.

The venue will be Zaandam Hotel, Amsterdam. The hotel is easy to reach from Amsterdam Schiphol airport.

The costs is: 460€ per person, which includes cost of the course, accommodation for two nights, two three course dinners, breakfast and lunch during the course.

Click [here](#) for more information.

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## ! New Clinical Utility Gene Cards

[3M syndrome-update 2013](#) – July 2013

[Alaïlle Syndrome \(ALGS\)](#) – June 2013

[Beckwith-Wiedemann syndrome](#) – June 2013

[Hypophosphatasia - update 2013](#) – Aug 2013

[Vici syndrome](#) – July 2013

Click [here](#) for the full list of CUGC's.

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## Publications

### *By EuroGentest members:*

- [Recommendations for reporting results of diagnostic genetic testing \(biochemical, cytogenetic and molecular genetic\)](#) by Els Dequeker, Mireille Claustress, Ros Hastings, David Barton and others.
- [EURO-WABB: an EU rare diseases registry for Wolfram syndrome, Alstrom syndrome and Bardet-Biedl syndrome](#) by Ségolène Aymé and others.
- [The European BRCA patent oppositions and appeals: coloring inside the lines](#) by Gert Matthijs and others.
- [Reflecting on earlier experiences with unsolicited findings: Points to consider for next generation sequencing and informed consent in diagnostics](#) by Tessel Rigter, Lidewij Henneman, Ulf Kristoffersson, Pascal Borry, Martina Cornel and others.  
! Watch [also this video](#) where author Tessel Rigter presents this publication.

### *For your interest:*

- [Genetic tests obtainable through pharmacies: the good, the bad, and the ugly](#)
- [Ethical, Legal, and Counseling Challenges Surrounding the Return of Genetic Results in Oncology \(restricted access\)](#)
- [ACMG clinical laboratory standards for next-generation sequencing](#)
- [Genomic Testing: The Clinical Laboratory Perspective](#)

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## Other

- Genetic Counseling Program Gives Cigna Customers Increased Access to Genetic Counselors
- Click here for reports on the [ISCN educational session](#) and [the PWG Genetics and Society workshop](#) (during annual ECA meeting) in- Dublin (Ireland) June 29 2013.

- Connect to EuroGentest on [Linkdn](#)
- Read more [about EuroGentest](#)

