



# Newsletter March 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

- [Genomic Disorders 2013, 10-12 Apr](#)
- 8th International Meeting on CNVs and Genes in Intellectual Disability and Autism, 12-13 Apr
- HGM 2013 and 21st ICG, 13-18 Apr
- First International GENCODYS Conference, 14-17 Apr
- First International Rare Diseases Research Consortium Conference, 16-17 Apr
- 12th International Symposium on Mutation in the Genome, 22-26 Apr
- Train the Trainers Courses, 27 Apr-17 May



## 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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## Report of the EuroGentest symposium in Prague, March 7-8, 2013



We would like to thank all speakers and participants for a very successful conference! It was filled with inspiring presentations and lively discussions.

Also a big thank you to our colleagues in Prague for the warm welcome!

Some presentations are already available on the website for the participants of the symposium, the rest will follow as we receive approval from the speakers. Click [here](#) to view the presentations (and log-in) and a short report.

*Access to the presentations is limited to event participants only.*

## EuroGentest at the European Society of Human Genetics in Paris

**June 8-11, 2013** the European Society of Human Genetics is organizing its yearly conference in Paris (France) and as previous years EuroGentest will be there with a joint booth with the ESHG and Orphanet. For this we would like to call upon all EuroGentest partners to join us at the booth so we can address the people interested in our activities in an optimal manner!

There will also be several workshops organized by EuroGentest during the meeting, please take a look at [our webpage](#) for an overview of our activities during [ESHG2013](#).

We look forward to welcoming you at our booth in Paris!



## Workshop report: "Preimplantation genetic diagnosis and gamete testing"

The objectives of this second meeting organized by Workgroup 10 of EuroGentest 2 were to edit some guidelines for PGD for triplet repeat disorders (particularly for Myotonic Dystrophy type 1, Huntington Disease and Fragile X Syndrome) in order to define the basis for an international harmonization of technological protocols. This meeting was held in Montpellier (France) on February 5th 2013.

Several experts from 9 PGD centres in Europe (Athens, Brussels, London, Maastricht, Madrid, Montpellier, Paris, Rome and Strasbourg) as well as the scheme director of EQA (UK NEQAS) for monogenic disease PGD attended the meeting.

In the morning, each participant presented his activity, the techniques used and results obtained.

In the afternoon, a round table was organized to discuss different points: strategies to use (direct or indirect analysis); definition of « informative » microsatellite markers; localization and genetic map of the markers used; inclusion criteria depending on the number of informative markers; strategies to use in the case of a lack of informativity and specific recommendations depending on the triplet repeat disorder.

A draft of the guidelines has been written and should be finalized before summer 2013.

## EuroGentest upcoming workshops

- **Workshop on Interpretation of diagnostic genetic results (biochemical, cytogenetics and molecular).** [Friday 7th June 2013 - ESHG Conference – Satellite meeting - Paris, France](#)
- **Advanced Workshop: Are we on the right track towards accreditation?** [2-3 May 2013 – Barcelona \(Spain\)](#)
- **E-Course: The accreditation process in detail.** [April-May 2013](#)
- **Atelier de perfectionnement: en bonne voie vers l'accréditation.** [3-4 October 2013, Paris \(France\)](#)
- **Validation of diagnostic tests in clinical molecular genetics.** [21-22 November, 2013 Venue to be determined](#)



## REGISTER

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

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## ! New Clinical Utility Gene Cards

[Achromatopsia - update 2013](#) – Mar 2013

[Long-QT syndrome \(types 1–13\)](#) – Mar 2013

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

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## Publications

### *By EuroGentest members:*

- [Genetics and democracy-what is the issue?](#) By Ulf Kristoffersson and others.
- [Eliciting preferences for priority setting in genetic testing: a pilot study comparing best-worst scaling and discrete-choice experiments](#) by Joerg Schmidtke, Wolf Rogowski, Franziska Severin and others.
- [A systematic review of factors influencing uptake of invasive fetal genetic testing by pregnant women of advanced maternal age](#) by Heather Skirton and others.
- [Preparation and validation of the first WHO international genetic reference panel for Fragile X syndrome](#) by David Barton, Rob Elles and others.

### *For your interest:*

- [Discovering misattributed paternity in genetic counselling: different ethical perspectives in two countries](#)
- [Ethical and policy issues in genetic testing and screening of children](#)
- [Physicians' preparedness for integration of genomic and pharmacogenetic testing into practice within a major healthcare system](#)

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## Other

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

