



Newsletter May 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

- Workshop on Interpretation of diagnostic genetic results (biochemical, cytogenetics and molecular) June 7
- Clinical Applications of Next Generation Sequencing, June 8
- European Human Genetics Conference 2013, June 8-11
- Belgian Medical Genomics Initiative Kickoff Meeting, June 18
- 2013 International Conference on Genomics in Europe, [June 25-28](#)
- Functional Genomics and Systems Biology training course, 19-28 June
- 5th International Congress of Molecular Medicine, 27-30 June
- 9th European cytogenetics conference, 29 June- 2 July



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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German policy documents on the regulation of genetic testing



In April 2013, two policy documents on the regulation of genetic services have been issued in Germany, one by the German Ethics Council and one by the Genetic Diagnostics Commission. The first one expresses recommendations on genetic diagnosis in general, firstly for improvement of the provision of information for the public and in training. The second document, is the Tri-annual Report of the Genetic Diagnostics Commission.

Click [here](#) to read a commentary on both documents, written by Jörg Schmidtke (Hannover) and Clemens Müller (Würzburg).

ACMG guidelines for genetic counselling



A Working Group of the ACMG (American College of Medical Genetics and Genomics) has spent a year developing a set of recommendations for reporting incidental findings. While the situation with regard to notifying patients of incidental findings arising from exome and genome sequencing (defined by the ACMG as clinical sequencing) requires clarification, it seems that the ACMG has gone well beyond the issue of incidental findings with these recommendations. From my perspective some of the guidance appears to contravene ethical practice.

Click [here](#) to read the full comment written by Heather Skirton.

Visit EuroGentest at the booth in Paris ESHG2013

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 (booth # 460).

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!



! New Clinical Utility Gene Cards

[Johanson-Blizzard syndrome](#) – May 2013

[Alström syndrome - update 2013](#) – Apr 2013

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

Publications

By EuroGentest members:

- [Whole-genome sequencing in health care](#) by Martina Cornel, Pascal Borry, Ros Hastings, Hans Scheffer and others.
- [Non-invasive prenatal diagnosis for single gene disorders: experience of patients](#) by Celine Lewis, Lyn Chitty and others.
- [Noninvasive prenatal testing: the paradigm is shifting rapidly](#) by Lyn Chitty and others.
- [A framework to start the debate on neonatal screening policies in the EU: an Expert Opinion Document](#) by Martina Cornel, Tessel Rigter and others.
- [The clinical implementation of non-invasive prenatal diagnosis for single-gene disorders: challenges and progress made](#) by Lyn Chitty and others.

For your interest:

- [Next generation sequencing \(NGS\) strategies for the genetic testing of myopathies](#)
- [Best ethical practices for clinicians and laboratories in the provision of noninvasive prenatal testing](#)
- [Noninvasive Prenatal Molecular Karyotyping from Maternal Plasma](#)



REGISTER

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

Other

➤ **The EuroGentest website has transferred to a new server.** We have done many efforts to maintain the look and feel of the website as it is, so you as a user will notice as little as possible of this change. However, the url's of the internal pages are changed and some pages will look a bit different. Please [contact us](#) if you experience any problems.

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

