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

*If you cannot see this newsletter properly view the online version*

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**Newsletter May 2012**

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**EuroGentest at the The European Human Genetics Conference 2012**

From 23 until 26th of June 2012, the European Society of Human Genetics (ESHG) will organize their annual meeting in Nuremberg (Germany), gathering professionals from all areas in genetic testing.

EuroGentest will be represented at the joint booth with Orphanet and ESHG at the exhibition hall during the whole conference. Please come and visit us for information about the patient leaflets, clinical utility gene cards, workshops and other EuroGentest activities.

In addition, many EuroGentest participants will present their work during the several symposia, workshops, education sessions and satellite meetings.

Click here for more information.

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**EuroGentest2 Workshop on Prenatal diagnostic testing guidelines: report**

One of the key deliverables for WP8, ‘Quality in genetic services’ was the publication of European guidelines on prenatal diagnostic testing.

On the 2nd and 3rd May, a group of invited experts participated in a workshop held in London to develop a set of recommendations to guide prenatal diagnostic testing in Europe. The 15 participants from eight countries were: Heather Skirton, Lyn Chitty, Katia Bilardo, Lesley Goldsmith, Helena Kaarialainen, Ouli Kamarainen, Susan Kelly, Faustina Lalatta, Celine Lewis, Milan Macek, Olav Petersen, Thomy de Ravel, Marta Rodríguez de Alba, Maria Soller and Sally Taffinder. The relevant disciplines of medical genetics, fetal medicine and laboratory genetics were well-represented.

We agreed that the focus of the guidelines was prenatal diagnosis for women whose fetus is at increased risk of a specific condition. Such testing may be performed using invasive (such as amniocentesis, chorionic villus sampling or fetal blood sampling) or less invasive procedures (such as analysis of cell-free fetal DNA in maternal blood or fetal imaging). The guidelines do not refer to antenatal screening tests. Following presentations on the way in which prenatal diagnostic testing is offered in the eight different countries, we focussed on preparing guidelines under four main topics: the objective of prenatal diagnostic testing, general principles underpinning prenatal diagnostic testing, logistical issues involved in testing and topics for counselling in prenatal diagnosis.

A draft document has been prepared and will be circulated to relevant professional organisations in the countries of Europe. We look forward to receiving comments from colleagues in the field.

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**! Newly published Clinical Utility Gene Cards for:**

Centronuclear and myotubular myopathies – May 2012

Check the EuroGentest website for a full list of CUGC’s.

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**Publications**

*By EuroGentest members:*

- The fate and future of patents on human genes and genetic diagnostic methods by Gert Matthijs and others.
- Refining noninvasive prenatal diagnosis with single-molecule next-generation sequencing by Neil Avent.
- Editorial comment on Single molecule sequencing of free DNA from maternal plasma for noninvasive trisomy 21 detection by Bert Bakker and others.
- Suggested components of the curriculum for nurses and midwives to enable them to develop essential knowledge and skills in genetics by Heather Skirton and others.
- Next-generation genetic testing for retinitis pigmentosa by Hans Scheffer and others.

*For your interest:*

- Personal omics profiling reveals dynamic molecular and medical phenotypes.
- Telegenetics: a systematic review of telemedicine in genetics services.
The EuroGentest website has a new homepage. For more information on how to achieve an optimal view, click here.