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

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Newsletter October 2012

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

- 8th ISNS European Neonatal Screening Regional Meeting, 4-6 Nov
- Annual Meeting of the American Society of Human Genetics, 6-10 Nov
- Steering Committee meeting EUGT2, 16 Nov
- Third International Conference of the Cyprus Society of Human Genetics, 16-18 Nov
- Workshop on Prioritizing genetic tests, 28-29 Nov
- 7th Fraunhofer Life Science Symposium Leipzig 2012, 29-30 Nov
- 3rd annual World Orphan Drug Congress, 29-30 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 participant access.

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

Your opinion needed: Views on providing genetic tests

Research into molecular genetics is leading to large numbers of new genetic tests available in health care practice. At the same time there are demands in many European countries to contain costs of public health care. As a consequence, situations may exist or arise where not all desirable tests can be funded by existing health care budgets.

To work towards an ethically reflected guidance on how to deal with scarce health care resources EuroGentest researchers in collaboration with the Public and Professional Policy Committee of the European Society for Human Genetics are currently conducting an online survey on this difficult topic. We invite you to participate in the short anonymous online survey available under http://ww3.unipark.de/uc/providingtests/

The results of this survey will be discussed at a closed workshop on November 28th, for more information click here.

Preliminary program available for the 3rd EuroGentest meeting (Prague)

A preliminary program is now available revealing the first speakers for the 3rd EuroGentest meeting, co-organized this year by APOGEE-Net/CanGeneTest.

No admission fee, but on-line registration is required.

.IRDiRC: an update

The International Rare Diseases Research Consortium (IRDiRC) has been created to increase the international collaboration on rare diseases research and bring together funding bodies and researchers. The ultimate goal is to develop 200 new therapies and to provide diagnostics for all rare genetic diseases by 2020. This consortium is an initiative of the European Commission and the National Institutes of Health (NIH,USA). Three scientific committees have been set-up in the consortium: diagnostics, therapies and horizontal aspects.

The Executive Committee of IRDiRC met on the 25th and 26th of September 2012 in Evry, France. The consortium has received a grant from the European Commission's DG Research which allows it to be supported from now on by a Scientific Secretariat located at the Rare Disease Platform in Paris. The contract for IRDiRC has been assigned to INSERM (the French National Institute for Health and Medical Research) with Dr. Ségolène Aymé as project leader and to the Rare Disease Foundation with Prof. Nicolas Lévy leading this project for the foundation. This new resource will definitely be of great help for IRDiRC to achieve its goals.

More information on IRDiRC can be found on their website.

! New Clinical Utility Gene Cards

Osteogenesis imperfecta– **Sept 2012**

Familial polycythaemia vera **– Oct 2012**

Incontinentia pigmenti **– Oct 2012**

Campomelic dysplasia **– Oct 2012**

Click here for the full list of CUGC's.

Publications

By EuroGentest members:

- The changing landscape of genetic testing and its impact on clinical and laboratory services an research in Europe by Ros Hastings, Brian Fowler, Egbert Bakker, David Barton, Joerg Schmidtke Martina Cornel and others.
- The introduction of arrays in prenatal diagnosis: a special challenge by Ros Hastings and others.
- The challenge of implementing genetic tests with clinical utility while avoiding unsound applications b Martina Cornel and others.
- Non-invasive prenatal diagnosis of single-gene disorders from maternal blood by Marta Rodriguez d Alba and others.
- Translation of a research-based genetic test on a rare syndrome into clinical service testing, with soto syndrome as an example by Helena Kääriäinen and others

For your interest:

- Ethical issues with newborn screening in the genomics era
- Specific guidelines for assessing and improving the methodological quality of economic evaluations o newborn screening

Other

- Connect to EuroGentest on LinkdIn
- Read more about EuroGentest
- The EuroGentest website has a new look! Check out the new introduction pages: Genetic Laboratories Health Professionals and Patients, Public and Policy.