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

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Newsletter July 2013

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

- Clinical practice guidelines on rare diseases, July 8-12
- Human Genome Analysis: Genetic Analysis of Multifactorial Diseases, July 24-30
- International Symposium of Personalized Medicine, 16-17 Aug

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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JRC publishes a report entitled 'Genetic testing offer in Europe' with the main outcomes of an experts' meeting held in November 2012

On 21 May 2013, the European Commission's Joint Research Centre (JRC) published a scientific and policy report entitled "Genetic Testing Offer in Europe", which summarises the main outcomes of an experts' meeting which took place at the JRC-Ispra site on 19-20 November, 2012.

The workshop was organised by the JRC in collaboration with the European Union Committee of Experts on Rare Diseases' (EUCERD's) and EuroGentest to discuss the quality of genetic testing and the future organisation of genetic testing in Europe. The report also highlights specific points which potentially deserve future attention by the European Commission. It gathered experts and stakeholders in the field of genetic testing, including representatives from the European Commission (JRC, Directorates-General for Research & Innovation and Health and Consumers).

The report presents developments and expert recommendations on a selection of topics, including organisation of genetic testing for rare diseases in Europe, quality assurance, next generation sequencing and direct-to-consumer genetic testing. The selection was based on the importance and urgency of the matter and the need and opportunity for action at the European level, and the likelihood for successful intervention.

Click here for more information and to download the report.

The First International Rare Disease Research Consortium (IRDiRC) conference: report and guidelines

The International Rare Diseases Research Consortium (IRDiRC) was initiated by the European Commission and the US National Institutes for Health Research and launched in April 2011 to foster international collaboration in the rare diseases field. Their first international conference took place in Dublin (Ireland) 16-17 April 2013 and was organised by the European Commission in association with the Irish Presidency of the European Union.

IRDiRC

The conference aimed to gather stakeholders active in the rare disease area from across the globe. In addition to a top-level programme taking stock of advances toward IRDiRC goals, it provided ample opportunities to network with the international rare disease community.

Click here for more information and for the report and abstracts of the conference.

EUCERD held it's 8th meeting and adopts two sets of recommendations

The European Union Committee of Experts on Rare Diseases (EUCERD) held its 8th meeting on 5-6 June 2013, marking the end of its three-year mandate. This last meeting was proof of the effectiveness of the Committee in successfully bringing together representatives of the European Commission, Member States, patient organisations, Industry and experts in the field of public health and research, as it saw the adoption of two sets of recommendations, on patient registration and data collection and indicators for national plans and strategies respectively.

Click here for more information and to download the recommendations.

! New Clinical Utility Gene Cards

Arrhythmogenic right ventricular cardiomyopathy (ARVC) – June 2013

Hereditary thrombocythemia – June 2013

Tuberous sclerosis complex (TSC1, TSC2) – June 2013

Click here for the full list of CUGC's.

Publications

By EuroGentest members:

- Health needs assessment for medical genetic services for congenital disorders in middle- and low-income nations by Ulf Kristoffersson, Jörg Schmidtke, Alistair Kent 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.

For your interest:

- A tiered-layered-staged model for informed consent in personal genome testing
- Survey of European clinical geneticists on awareness, experiences and attitudes towards direct-to-consumer genetic testing
- Non-invasive Prenatal Testing: Technologies, Clinical Assays and Implementation Strategies for Women's Healthcare Practitioners
- Molecular genetic testing and the future of clinical genomics

Other

- **Orphanet** has launched the Dutch language site.
- **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 LinkdIn
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